Instand-NGS4P OMC Questionnaires

The final questionnaires addressed to users (66 pages), patient associations (37 pages) and solution providers (58 pages) prepared by the project consortium experts and uploaded to Survey Monkey are displayed.

USER'S Questionnaire INSTAND-NGS4P

PROJECT INTRODUCTION

INSTAND-NGS4P is an EU-funded Pre-Commercial Procurement (PCP) project for improving cancer patient's benefit from Next Generation Sequencing (NGS). Driven by patient and clinical needs, two innovative NGS workflows from sample pre-analytics to medical decision-making will be developed for routine diagnostics of common and rare adult and paediatric cancers complying with the IVDR. The developed workflows will compile information from cancer gene testing, pharmacogenetics testing and e-medication in proper presentation to medical doctors for supporting therapy decision-making at bedside widely applicable in health systems.

The EU-co-funded PCP project provides funding for a public consortium to define unmet medical and technical needs based on an Open Market Consultation, which lays the foundation for a call for tenders addressing solution providers (companies) to develop their products to better meet user needs. At three cut-off periods, companies responding to this call will be evaluated regarding their ability to answer these users' needs from design perspective until the product phase. The total funding allocated to companies for product development (in total 8.55 M€) will finally lead to two integrated standardized NGS workflows, including decision support.

The different lots of this project cover the entire workflow for integration and standardization of targeted and whole genome DNA sequencing and decision making at the bedside:

- lot 1: Pre-sequencing (specimen collection, nucleic acid isolation, library preparation)

- lot 2: Sequencing

- lot 3: Bioinformatics analysis

- lot 4: Integrated reporting

The procurement will take the form of a pre-commercial procurement (PCP) under which R&D service contracts will be awarded to a number of solution providers in parallel in a phased approach. This will make it possible to compare competing alternative solutions.

Each selected operator will be awarded a framework agreement that covers 3 R&D phases.

The 3 phases are:

- Design of lots
- Prototype of lots
- Fully integrated NGS workflow.

Each of the 3 phases will address all 4 lots.



USER'S Questionnaire INSTAND-NGS4P

PURPOSE OF THE QUESTIONNAIRE

This questionnaire is part of the Open Market Consultation (OMC) which aims at refining the clinical, patient and technical needs defined by the project consortium, as well as the emerging solutions available or under development to address these needs. For this purpose, three questionnaires were developed for three different stakeholder groups – users, solution providers and patient associations. The feedback collected from the questionnaires will support the subsequent preparation of the call for tenders. The questionnaires also allow disclosure of confidential information to the project consortium, which will be treated with high confidentiality. Aggregated data from the results of the survey will potentially be published in an anonymous way, excluding the information labelled as confidential.

The personal information collected through the questionnaires will be exclusively used for the purposes of the project, will not be shared with third parties, and will be deleted after the closure of the project.

GUIDANCE

The questionnaire is addressed to all solution providers in the field of NGS, and is divided into the following sections: General questions, Lot 1, Lot 2, Lot3, Lot4. After answering the general questions, you will be asked for each section whether you want to fill it in or not. We would highly appreciate if you would take the time to fill in the section(s) related to your expertise. However, if you do not find the question relevant, feel free to select N/A (not applicable) as an answer or skip the question and to move on to the next one.

Please submit the questionnaire by May 31st, 2021. Thank you in advance for contributing.

* 1. Organisation profil	e
Organisation name	
Address	
City/Town	
Country	
* 2. Contact person	
First name	
Family name	
Email Address	
Phone Number	

USER'S Questionnaire INSTAND-NGS4P
General questions
* 3. How is your organization positioned in the NGS scene (more than one answer possible):
Provider
Manufacturer
Includes Research & Development
Hospital/medical centre
Academic hospital
Diagnostic laboratory
Patient organization
Other (please specify)

* 4. What is your current position in your organization?

5. Are you currently performing production/diagnostic activities for products used or diagnostics in the NGS workflow in (if yes, please specify the percentage in the textbox):

Europe	
North America	
Asia	
South America	
Australia	

* 6. Are you currently performing research and development activities for products/diagnostics used in the NGS workflow in (if yes, please specify the percentage in the textbox):

Europe	
North America	
Asia	
South America	
Australia	

* 7. Are you using NGS sequencing for research or for diagnostics?

- O Yes
- O No
- __N/A
- * 8. Do you have access to sequencing facilities?
 - Yes
 - O No
 - __N/A
- * 9. Do you perform sequencing in house or externally?
 - O In house
 - Externally
 - ON/A
- * 10. What platform do you use?
 - 🔵 Illumina
 - Olon Torrent
 - Gene-Reader

```
Oxford Nanopore
```

MGI

- 🔵 Pac Bio
- 🔿 N/A
- Other (please specify)
- * 11. How many samples are you typically sequencing per week?
 - C Less than 1
 - 0 1-10
 -) 10-50
 - 50-100
 - More than 100
 - () N/A

* 12. How interested are you in whole exome sequencing? (0-not interested in all, 5 - very interested)						
0	1	2	3	4	5	N/A
\bigcirc	\odot	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 13. How interes	sted are you in g	gene panels? (0	-not interested i	in all, 5 - very int	erested)	
0	1	2	3	4	5	N/A
\bigcirc	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 14. How interested are you in whole genome sequencing? (0-not interested in all, 5 - very interested)						
0	1	2	3	4	5	N/A
\bigcirc	\odot	\odot	0	\odot	\odot	\bigcirc

- * 15. Are you a user (current or future) of NGS in the clinical setting?
 - 🔵 Yes
 - O No

USER'S Questionnaire INSTAND-NGS4P
LINICAL NEEDS
16. Do you currently employ NGS in your clinical practice?
⊖ Yes
No
For clinical research purpose only
17. Do you currently base clinical decisions in daily clinical practice on NGS?
⊖ Yes
No
☐ I am not sure
18. How many times do you perform NGS during the patient's journey?
 At diagnosis
 At diagnosis and progression
O As a monitoring tools
19. Do you or your center/hospital participate in precision oncology trials?
⊖ Yes
⊖No
() I am not sure
No, but would be interested in
If yes, please specify:
20. What is the turn around time (from sample collection) in which you expect to receive an NGS report?
○ <72h

72h-1 week
1-2 weeks
2-3 weeks
3-6 weeks
>6 weeks
Not important

21. Which type of cancers do you mainly treat?
O Adult
Childhood
O Adult and childhood
22. If you treat adult cancers, which kind of cancers do you treat?
Lung
Breast
Colo-rectal
Bilio-pancreatic
Gastric
Melanoma
Adult sarcoma
Prostate
Bladder
Gynecological
Head and neck
Other (please specify)
23. If you treat childhood cancers, which kind of cancers do you treat?
All
AML
CML
Lymphoma
Neuroblastoma
Ewing Sarcoma

Osteosarcoma

Other (please specify)

Tumor Blood Bone marrow r (please specify). If blood or bone marrow, please specify sample collection tubes: How are the samples processed? Formalin-fixed and paraffin embedded (FFPE) Fresh frozen Fresh refrigerated Don't know Other (please specify)	
Blood Blood Bone marrow r (please specify). If blood or bone marrow, please specify sample collection tubes: How are the samples processed? Formalin-fixed and paraffin embedded (FFPE) Fresh frozen Fresh refrigerated Don't know Other (please specify)	
 Bone marrow r (please specify). If blood or bone marrow, please specify sample collection tubes: How are the samples processed? Formalin-fixed and paraffin embedded (FFPE) Fresh frozen Fresh refrigerated Don't know Other (please specify)	
r (please specify). If blood or bone marrow, please specify sample collection tubes: How are the samples processed? Formalin-fixed and paraffin embedded (FFPE) Fresh frozen Fresh refrigerated Don't know Other (please specify)	
How are the samples processed? Formalin-fixed and paraffin embedded (FFPE) Fresh frozen Fresh refrigerated Don't know Other (please specify)	
How are the samples processed? Formalin-fixed and paraffin embedded (FFPE) Fresh frozen Fresh refrigerated Don't know Other (please specify)	
How are the samples processed? Formalin-fixed and paraffin embedded (FFPE) Fresh frozen Fresh refrigerated Don't know Other (please specify)	
Formalin-fixed and paraffin embedded (FFPE) Fresh frozen Fresh refrigerated Don't know Other (please specify)	
Fresh frozen Fresh refrigerated Don't know Other (please specify)	
Fresh refrigerated Don't know Other (please specify)	
Don't know Other (please specify)	
Other (please specify)	
Intermediate	
) Low	
High	
y very nigh	
Which information do you need covered by an NGS workflow?	
Risk stratification	
Prognostic factors	
Predisposition/Hereditary syndromes	
Therapeutic targets	
Pharmacogenetics	

28. V	Which types of genetic variants are relevant for cancer predisposition?	
	Single nucleotide variants (SNV)/small insertions and deletions (INDEL)	
	Copy number aberrations (CNV)	
	Other (please specify)	
L		
29. V	Which types of genetic variants are relevant as actionable targets?	
	which types of genetic variants are relevant as actionable targets.	
	Single nucleotide variants (SNV)/small insertions and deletions (INDEL)	
	Single nucleotide variants (SNV)/small insertions and deletions (INDEL) Copy number aberrations (CNV)	
	Single nucleotide variants (SNV)/small insertions and deletions (INDEL) Copy number aberrations (CNV) Fusions	
	Single nucleotide variants (SNV)/small insertions and deletions (INDEL) Copy number aberrations (CNV) Fusions Overexpression	
	Single nucleotide variants (SNV)/small insertions and deletions (INDEL) Copy number aberrations (CNV) Fusions Overexpression Other (please specify)	
	Single nucleotide variants (SNV)/small insertions and deletions (INDEL) Copy number aberrations (CNV) Fusions Overexpression Other (please specify)	

30. How important is it to include the level of evidence of genetic variants for suggested targeted treatments in the report (ie clinical variants actionable/clinical trials restricted/research mutations only) ?

Very low
O Low
 Intermediate
High
🔵 Very high

31. How important is it to include the level of evidence of single and combined targeted treatments in the report?

- O Very low
- O Low
- Intermediate
- High
- Very high

32. How important is it to include pharmacogenetic information in the report?

O Very low
O Low
Intermediate
High
⊖ ^{Very high}

33. What kind of pharmacogenetic information do you think is useful (please select all that apply)?

Avoiding toxicity

Evaluation of drug-drug interactions

Drug dose assessment

Other (please specify)

34. How important is it to include response predictions to targeted therapies in a report?

\bigcirc	Very low
0	Low
\bigcirc	Intermediate
O	High
\bigcirc	Very high

35. If it is not an approved drug for the use in pediatric patients, how important is it to provide information on

recommended treatment schedules and dose modifications?

Very low Low Intermediate High Very high

36. Is it important to state clearly in the report suspect germline mutations?



Only for restricted cancer type, specify

37. Is it important to quote in the report suspect clonal hematopoiesis?



38. How important is it to determine and clarify the allele frequency of single gene alteration in the report) Very low O Low Intermediate High () Very high 39. How is it important to list all VUS in the final report? O Very low O Low Intermediate High Very high 40. How important is it to report "complex" biomarker like TMB/HR/DNA Repair deficiency 🔵 Very low O Low Intermediate High Very high

41. How important is it to include the possibility to match genetic findings to clinical trial active for enrollment in your country?

O Very low

O Low

Intermediate

High

Very high

42. Do you currently participate in molecular tumor boards?

O Yes

No

No, but interested in

If yes, please specify which and if they are virtual

43. If you have any further comments in the context of clinical needs. Please let us know.

44. Is this information confidential?



USER'S Questionnaire INSTAND-NGS4P

Introduction lot 1

* 45. Would you like to answer questions related to Lot 1 (Pre-Analytics and Library preparation)?



ot 1						
HALLENGE 1:						
nproving the analytic	cal performance by s	tandardizing and	or simplifying the	e pre-analytical p	rocesses	
46. Most commo	only used/produce	ed samples for	NGS analysis			
Which are the ty	ypes of specimens	s most commor	nly analyzed in	your Lab?		
FFPE						
Frozen tissue	es					
Blood						
FNAs						
Plasma						
N/A						
Other (please	e specify)					
+1. IS UIC [[[[U[[[iation above conii	dential?				
Yes, it is cont No, it can be mong the following, pla ocessing standards. F B. FFPE	fidential shared ease indicate the spec Please weight each of t	dential? simen(s) you consi them on the basis	der most challengi of their importance	ng in terms of defii from 1 (lower) to t	ning detailed pre-a 5 (higher).	nalytical
Yes, it is cont No, it can be mong the following, pla ocessing standards. F B. FFPE	fidential shared ease indicate the spec Please weight each of t	dential? simen(s) you consi them on the basis 2	der most challengi of their importance 3	ng in terms of defii from 1 (lower) to t 4	ning detailed pre-a 5 (higher). 5	nalytical N/A
Yes, it is cont No, it can be mong the following, pla ocessing standards. F B. FFPE	fidential eshared ease indicate the spec Please weight each of t 1	dential? simen(s) you consi them on the basis 2	der most challengi of their importance 3	ng in terms of defin from 1 (lower) to s 4	hing detailed pre-a 5 (higher). 5	nnalytical N/A
Yes, it is cont Yes, it is cont No, it can be mong the following, plu ocessing standards. F B. FFPE DNA RNA	fidential shared ease indicate the spec Please weight each of t 1	dential? timen(s) you consi them on the basis 2	der most challengi of their importance 3	ng in terms of defin from 1 (lower) to 5 4	hing detailed pre-a 5 (higher). 5	nalytical N/A
Yes, it is cont No, it can be mong the following, pla ocessing standards. F B. FFPE DNA RNA 9. Frozen tissues	fidential eshared ease indicate the spec Please weight each of t 1	dential? simen(s) you consi them on the basis 2	der most challengi of their importance 3	ng in terms of defin from 1 (lower) to 5 4	hing detailed pre-a 5 (higher). 5	nnalytical N/A
Yes, it is cont No, it can be mong the following, pla ocessing standards. F B. FFPE DNA RNA 9. Frozen tissues	fidential eshared ease indicate the spec Please weight each of t 1	dential? simen(s) you consi them on the basis 2	der most challengi of their importance 3 O	ng in terms of defin from 1 (lower) to s 4	hing detailed pre-a 5 (higher). 5 0	nnalytical N/A
 Yes, it is cont Yes, it is cont No, it can be mong the following, playocessing standards. F B. FFPE DNA P. Frozen tissues DNA 	fidential eshared ease indicate the spec Please weight each of t 1	dential? simen(s) you consi them on the basis 2 2 2 2	der most challengi of their importance 3 0 3 3	ng in terms of defin from 1 (lower) to 9 4 0	ning detailed pre-a 5 (higher). 5 0	nnalytical N/A

50. Blood/plasma						
	1	2	3	4	5	N/A
DNA						\bigcirc
RNA	\odot	\odot	\odot	\odot	\odot	\odot
cfDNA						\bigcirc
cfRNA	0	\odot	0	0	0	\bigcirc
51. Fine Needle Aspirates	;					
	1	2	3	4	5	N/A
DNA						0
RNA	0	0	0	0	0	0
52. Circulating Tumor Cel	ls					
	1	2	3	4	5	N/A
DNA						0
RNA	0	0	0	0	0	\odot
53 Extracellular vesicles						
	1	2	3	4	5	N/A
DNA		_		·	Ū	0
RNA	0	0	0	0	0	0
	0	0	0	0	~	
54. Saliva						
	1	2	3	4	5	N/A
DNA						0
55. Other						
	1	2	3	4	5	N/A
DNA						\bigcirc
RNA	\bigcirc	\odot	\odot	\bigcirc	\odot	0

56. If you selected other, please specify

PROBLEM: Preanalytical processing procedures can cause variations in the test result

SOLUTION: Standardisation according to ISO and CEN documents

Is your organization aware of the following standards? Please select the first box if there is awareness and the second if your organization works according to or applies the standard:

57. ISO 15189 Medical Laboratories - Requirements for quality and competence



58. ISO 20166-1:2018 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for formalin-fixed and paraffin-embedded (FFPE) tissue - Part 1: isolated RNA



59. ISO 20166-3:2018 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for formalin-fixed and paraffin-embedded (FFPE) tissue - Part 3: isolated DNA



60. ISO 20184-1:2018 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for frozen tissue - Part 1: isolated RNA



61. ISO 20186-1:2019 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for venous whole blood - Part 1: isolated cellular RNA



62. ISO 20186-2:2019 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for venous whole blood - Part 2: isolated genomic DNA



63. ISO 20186-3:2019 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for venous whole blood - Part 3: isolated circulating cell free DNA from plasma

Aware

Applies

64. CEN/TS 17390-1:2020 (WI=00140123) Molecular in vitro diagnostic examinations - Specifications for preexamination processes for circulating tumor cells (CTCs) in venous whole blood - Part 1: Isolated RNA

Aware Applies

65. CEN/TS 17390-2:2020 (WI=00140125) Molecular in vitro diagnostic examinations - Specifications for preexamination processes for circulating tumor cells (CTCs) in venous whole blood - Part 2: Isolated DNA

Aware Applies

66. Do you adhere to other ISO standards for NGS pre-analytics or library preparation?

O Yes

If yes, please specify

67. Does your organization see ISO standardization as a good and useful approach?

) Yes

No

Comment

PROBLEM Many quality issues in pre-analytics arise even before nucleic acid extraction, during the whole process starting from sample collection, processing and storage.

SOLUTION: An entry-level quality check to confirm the sample is fit for purpose.

68. In your opinion, this would be:



Helpful

Crucial to develop

69. Which parameters	beyond adherence to standards would be needed to be checked
for fixed, paraffin- embedded samples	
for fresh frozen tissue	
for white blood cells (PBMC)	
for liquid biopsies	

PROBLEM Sample transport and storage might be required which can change the nucleic acids' quality/quantity.

SOLUTION: Stabilization of DNA and RNA (this can include all different types of DNA and RNA depending on the source and target situation)

70. Is your organization interested in further development of such an approach?

- O Yes
- No
- Comment

71. PROBLEM Sample pre-analytical processing can influence the final result of NGS.

SOLUTION: The project participants have compiled a list of key challenges in sample and library preparation that could be improved within the next 2 - 3 years.

Please weight each of them on the basis of their importance from 1 (lower) to 5 (higher).

	0	1	2	3	4	5
Increase in sample stability	0	0	0	0	0	0
Increase in sample quantity	0	0	0	0	\odot	0
Increase target concentration in the sample	0	0	0	\odot	\odot	0
Direct or long range sequencing	\odot	0	\bigcirc	0	0	0
Alternative innovative stabilization methods	\odot	0	0	0	0	0
Compatibility of sample stabilizers with analytical test procedure that need to be performed or have been performed on the same sample (e.g. histology/cytology)	0	Õ	Q	Q	Q	\bigcirc
Efficiency of the analyte isolation/extraction procedure	0	0	0	0	\odot	0
Quality assessment of the isolated analyte	0	0	0	0	0	0
Sample storage	\odot	\odot	\odot	\odot	\odot	\odot
Analyte storage	0	\odot	\odot	\bigcirc	\odot	\bigcirc
Automation of the isolation procedures	\bigcirc	0	\odot	0	0	0
Standardization	0	\odot	0	\odot	0	\odot
Other	0	0	0	0	0	0
Other (please specify)						

72. Is your organization interested in further development in one or more of these approaches?

- O Yes
- O No

73. Please give the numbers with the highest priority first, down to the lowest priority you are interested in:
Increase in sample stability
·
Increase in sample quantity
Increase target concentration in the sample
Direct or long range sequencing
,
Alternative innevetive stabilization methods
Compatibility of sample stabilizers with analytical test procedure that need to be performed or have been performed on the same
sample (e.g. histology/cytology)
Efficiency of the analyte isolation/extraction procedure
·
Quality assessment of the isolated analyte
Sample storage
Analyte storage

Automation of the isolation procedures	Automation of the isolation procedures , Standardization	
Automation of the isolation procedures	Automation of the isolation procedures , Standardization	
. Standardization 4. Comment and explain your choice for the highest and second highest priority.	Standardization	
Standardization 4. Comment and explain your choice for the highest and second highest priority. ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing with small size DLUTION: Simultaneous isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach ? 76. Yes 70. 7. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rige stocks that might expire before they can be used up. DLUTION: Compatibility of NA isolation methods with multiple source target situations	Standardization	
Standardization 4. Comment and explain your choice for the highest and second highest priority. ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing with small size imples. OLUTION: Simultaneous isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach? Yes No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) NA 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. DLUTION: Compatibility of NA isolation methods with multiple source target situations	Standardization	
A. Comment and explain your choice for the highest and second highest priority. ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing with small size amples. OLUTION: Simultaneous Isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach? Yes No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) N/A 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different Isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		
4. Comment and explain your choice for the highest and second highest priority. ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing with small size amples. OLUTION: Simultaneous isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach? Yes No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) N/A 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		
ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing with small size amples. OLUTION: Simultaneous Isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach? Ves No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) NA T. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations	4. Comment and explain your choice for the highest and second highest priority.	
ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing with small size amples. OLUTION: Simultaneous Isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach? Ves No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) NA T. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		
ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing with small size amples. OLUTION: Simultaneous Isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach? Ves No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) N/A 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. CLUTION: Compatibility of NA isolation methods with multiple source target situations		
ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing with small size amples. OLUTION: Simultaneous Isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach? Ves No 5. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) NA 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		
OLUTION: Simultaneous Isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 75. Is your organization interested for further development in this approach? Ves No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) NA 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) NA 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. CUUTION: Compatibility of NA isolation methods with multiple source target situations	ROBLEM During nucleic acid isolation, it is often impossible to obtain more than one NA target especially when dealing w amples.	vith small size
75. Is your organization interested for further development in this approach? Yes No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) N/A SSSSS O 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations	OLUTION: Simultaneous Isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run.	
/5. Is your organization interested for further development in this approach? Yes No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) N/A 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		
Yes No 6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) N/A S S S S S O 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations	75. Is your organization interested for further development in this approach?	
O O O	⊖ Yes	
6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) N/A S S S S S S S O 7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations	No	
6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) N/A SSSSSO		
N/A Š Š Š Š Š Š Š J 7. Please let us know if you have any comment	6. Please weight the importance of the further development of this approach from 1 (lower) to 5 (h	igher)
7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		N/A
7. Please let us know if you have any comment ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		\odot
7. Please let us know if you have any comment		
ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations	7. Please let us know if you have any comment	
ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		
ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and rge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		
ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and irge stocks that might expire before they can be used up. OLUTION: Compatibility of NA isolation methods with multiple source target situations		
OLUTION: Compatibility of NA isolation methods with multiple source target situations	ROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to cho arge stocks that might expire before they can be used up.	oose from and
OLUTION: Compatibility of NA isolation methods with multiple source target situations		
	OLUTION: Compatibility of NA isolation methods with multiple source target situations	

) Yes	
No	
Comment	
DBLEM Panels are lated before use. T inalysed.	constantly in need of extension for addition of new diagnostic marker sequences and need to be verified and his complicates library preparation due to optimal selection of changing panels to diagnostically cover the case to
UTION: Whole Ge	nome Sequencing (WGS) and Whole Exome Sequencing (WES) instead of targeted NGS approach
79. Is your orga	nization interested in such an approach?
) Yes	
No	
Comment	
80. In case of F	&D involvement, would financial support available in the Instand-NGS4P project be
80. In case of F	&D involvement, would financial support available in the Instand-NGS4P project be
80. In case of F onot needed helpful crucial to de	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach
80. In case of F onot needed helpful crucial to de Please comment a	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach nd specify if your this answer is confidential or it can be shared
80. In case of F	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach nd specify if your this answer is confidential or it can be shared
80. In case of F	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach nd specify if your this answer is confidential or it can be shared
80. In case of F onot needed helpful crucial to de Please comment an DBLEM The ligation ecially for low input	2&D involvement, would financial support available in the Instand-NGS4P project be velop this approach nd specify if your this answer is confidential or it can be shared
80. In case of F onot needed helpful crucial to de Please comment a DBLEM The ligatior ecially for low input 81. Which appr	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach nd specify if your this answer is confidential or it can be shared of adapters is a critical step in library preparation. Insufficient adapter ligation leads to low library complexity, samples
80. In case of F onot needed helpful crucial to de Please comment a DBLEM The ligatior ecially for low input 81. Which appr amplicon-ba	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach ad specify if your this answer is confidential or it can be shared of adapters is a critical step in library preparation. Insufficient adapter ligation leads to low library complexity, samples bach(es) are you currently using: sed library preparation
80. In case of F onot needed ohelpful ocrucial to de Please comment a DBLEM The ligatior ecially for low input 81. Which appr onumber amplicon-ba onumber full length ac	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach nd specify if your this answer is confidential or it can be shared of adapters is a critical step in library preparation. Insufficient adapter ligation leads to low library complexity, samples bach(es) are you currently using: sed library preparation lapters
80. In case of F onot needed ohelpful ocrucial to de Please comment a DBLEM The ligatior ecially for low input 81. Which appr onumber of the stubby adap	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach nd specify if your this answer is confidential or it can be shared of adapters is a critical step in library preparation. Insufficient adapter ligation leads to low library complexity, samples bach(es) are you currently using: sed library preparation lapters ters
80. In case of F not needed helpful crucial to de Please comment an DBLEM The ligation ecially for low input 81. Which appr amplicon-ba full length ac stubby adap Other (please) 	A&D involvement, would financial support available in the Instand-NGS4P project be velop this approach ad specify if your this answer is confidential or it can be shared of adapters is a critical step in library preparation. Insufficient adapter ligation leads to low library complexity, samples bach(es) are you currently using: sed library preparation lapters ters e specify)
80. In case of F onot needed ohelpful ocrucial to de Please comment a DBLEM The ligatior ecially for low input 81. Which appr onumber of the stubby adap other (please)	&D involvement, would financial support available in the Instand-NGS4P project be velop this approach ad specify if your this answer is confidential or it can be shared of adapters is a critical step in library preparation. Insufficient adapter ligation leads to low library complexity, samples bach(es) are you currently using: sed library preparation lapters ters e specify)

			noguirou		
82. What is the	e guaranteed ligati	ion efficacy you can	acquire.		
0 70-80%					
80-90%					
90-95%					
○ >95%					
_UTION: Improve	the adapter design an	d library preparation proto	col.		
use of alternativ	ves to adapter- and an	nplicon-based approaches	i		
improve adapte	r design				
improve ligatior	ı process				
83. Is your org	anization intereste	ed in such an approa	ch?		
Yes					
No					
Sunday Street					
If yes, please give	e the number of the so	lution your organization is	interested in		
If yes, please give	e the number of the so t the importance o	lution your organization is	interested in ment of approache	es from 1 (lower) to	5 (higher) N/A
If yes, please give Please weight	t the importance o	lution your organization is of the further develop	interested in ment of approache Š	es from 1 (lower) to Š	o 5 (higher) N/A
If yes, please give Please weigh Š	e the number of the so t the importance o Š WES are difficult with	Iution your organization is of the further develop Š	interested in ment of approache	es from 1 (lower) to	o 5 (higher) N/A
If yes, please give Please weigh OBLEM WGS and	e the number of the so t the importance o Š WES are difficult with	Iution your organization is of the further develop Š	interested in ment of approache Š	es from 1 (lower) to	o 5 (higher) N/A
If yes, please give Please weight OBLEM WGS and JUTION: Use alternative	e the number of the so t the importance o Š WES are difficult with s for FFPE tissue, like	lution your organization is of the further develop Š FFPE tissue.	interested in ment of approache	es from 1 (lower) to	o 5 (higher) N/A
If yes, please give Please weight OBLEM WGS and JUTION: Use alternatives Use other fixation	e the number of the so t the importance o Š WES are difficult with s for FFPE tissue, like on/ stabilization metho	Iution your organization is of the further develop S FFPE tissue. frozen tissue ods that preserve the histo	interested in ment of approache Š	es from 1 (lower) to	o 5 (higher) N/A
If yes, please give Please weigh OBLEM WGS and JUTION: Use alternative Use other fixation 85. Is your org	e the number of the so t the importance o WES are difficult with s for FFPE tissue, like on/ stabilization metho anization intereste	Iution your organization is of the further develop S FFPE tissue. frozen tissue ods that preserve the histo ed or involved in R&E	interested in ment of approache Š	es from 1 (lower) to Š bilizes DNA and RNA fo	o 5 (higher) N/A
If yes, please give Please weigh OBLEM WGS and UTION: Use alternative Use other fixation 85. Is your org	e the number of the so t the importance o WES are difficult with s for FFPE tissue, like on/ stabilization metho anization intereste	Iution your organization is of the further develops S FFPE tissue. frozen tissue ods that preserve the histo ed or involved in R&E	interested in ment of approache Š	es from 1 (lower) to Š bilizes DNA and RNA fo pach?	o 5 (higher) N/A
If yes, please give Please weigh OBLEM WGS and UTION: Use alternative Use other fixation 85. Is your org	e the number of the so t the importance o S WES are difficult with s for FFPE tissue, like on/ stabilization metho anization intereste	Iution your organization is of the further develops S FFPE tissue. frozen tissue ods that preserve the histo ed or involved in R&E	interested in ment of approache Š	es from 1 (lower) to Š	o 5 (higher) N/A
If yes, please give Please weigh OBLEM WGS and UTION: Use alternative: Use other fixation 85. Is your org Yes No If yes, please give	e the number of the so t the importance o S WES are difficult with s for FFPE tissue, like on/ stabilization metho anization intereste	Iution your organization is of the further develops S FFPE tissue. frozen tissue ods that preserve the histo ed or involved in R&E	interested in ment of approache Š	es from 1 (lower) to S bilizes DNA and RNA fo bach?	o 5 (higher) N/A
If yes, please give Please weight DBLEM WGS and UTION: Use alternative: Use other fixation 85. Is your org Yes No If yes, please give	e the number of the so t the importance o Š WES are difficult with s for FFPE tissue, like on/ stabilization metho anization intereste	Iution your organization is of the further develops S FFPE tissue. frozen tissue ods that preserve the histo ed or involved in R&E	interested in ment of approache S logic qualities and stat D for such an appro interested in (highest p	es from 1 (lower) to S bilizes DNA and RNA for bach?	o 5 (higher) N/A
If yes, please give Please weight OBLEM WGS and UTION: Use alternative Use other fixation 85. Is your org Yes No If yes, please give	e the number of the so t the importance o S WES are difficult with s for FFPE tissue, like on/ stabilization metho anization intereste	Iution your organization is of the further develops S FFPE tissue. frozen tissue ods that preserve the histo ed or involved in R&E lution your organization is	interested in ment of approache S logic qualities and state o for such an appro-	es from 1 (lower) to Š bilizes DNA and RNA fo bach?	o 5 (higher) N/A

	36. In case of R&D invo	lvement, wou	uld financial supp	ort available in th	e Instand-NGS4P p	project be
	O not needed					
	helpful					
	Crucial to develop this	approach				
	_et us know if you have any	comments and if	these are confidentia	al		
87.	Please weight the impo	ortance of the	further developm	nent of this appro	ach from 1 (lower)	to 5 (higher)
						N/A
						0
88.	Comment					
PRO	BI FM WGS and WFS diffic	ult for Liquid Bior	osv (cfDNA and cfRN	A exosome and CT((s)	
			,			
SOL	UTION:					
1.	Maximise the yield and qua	ality of isolated ta	argets			
2.	Increase sequence sensitiv	vity and precisior	n			
	39. Is your organization	interested or	involved in R&D	for such an appr	oach?	
) Yes					
	No					
	f yes, please give the numbe	er of the solution	your organization is i	nterested in (highest	priority first):	
	90. In case of R&D invo	lvement, wou	uld financial supp	ort available in th	e Instand-NGS4P p	project be
) not needed					
	 helpful 					
	crucial to develop this	approach				
	_et us know if you have any	comments and if	these are confidentia	al		

5. Thease weight each e	or them on tr	ie basis of their	importance fr	om 1 (lower) to	5 (higher).	
	1	2	3	4	5	N/A
Universal approach for different sample types and targets	0	\odot	\odot	\odot	\odot	0
Automation of library preparation	\odot	\odot	\odot	\bigcirc	\bigcirc	\bigcirc
Automation of nucleic acid isolation alone	0	0	0	0	0	\odot
Automation from nucleic acid isolation (different targets and DNA and RNA, low and high yield) to sequencing (closed system)	0	0	0	0	0	0
Direct or long range sequencing	\odot	\odot	\odot	\bigcirc	0	0
Reduced number of steps needed	\odot	\odot	\odot	0	\odot	\odot
UMIs mandatory (for mutation detection down to 0.01-0.1% allele frequency)	0	0	0	0	0	0
Improvement of library preparation success rate	0	\odot	0	0	0	\odot
Improvement of library yield	Q	0	\bigcirc	\bigcirc	0	0
Minimal quality and quantity requirements for the input material	0	0	0	\odot	0	0
					\sim	

94. Is your organization interested in one or more of these approaches?



95. In case of R&D involvement, would financial support available in the Instand-NGS4P project be

O not needed

) helpful

 $(\)$ crucial to develop this approach

Let us know if you have any comments and if these are confidential

,	
Universal ap	proach for different sample types and targets
,	
Automation of	of library preparation
,	
Automation of	of nucleic acid isolation alone
,	
Automation 1	rom nucleic acid isolation (different targets and DNA and RNA, low and high yield) to sequencing (closed system)
,	
Direct or long	g range sequencing
,	
Reduced nu	mber of steps needed
,	
Reduced ne	ed for pre-amplification
,	
UMIs manda	tory (for mutation detection down to 0.01-0.1% allele frequency)
,	
Improvemen	t of library preparation success rate
,	
Minimal qual	ity and quantity requirements for the input material
. Commer	nt

PROBLEM Waste of material due to package sizes and stability of reagents
SOLUTION:

Increased long term stability of reagents and simplified storage requirements
Reduce and recycle packaging and consumables
Scalability of reagents

98. Is your organization interested in further development in one or more of these approaches?

Yes
No

Please give the numbers of interest or involvement with the highest priority first, down to the last priority:

99. In case of R&D involvement, would financial support available in the Instand-NGS4P project be

) not needed

helpful

crucial to develop this approach

Let us know if you have any comments and if these are confidential

CHALLENGE 2:

Integrating pre-analytical, analytical processes and data analytics into a standardized workflow

PROBLEM WGS and WES are difficult for FFPE material because of the multitude of chemical changes present in the sample.

SOLUTION: Alternative isolation/stabilisation methods suitable for WGS and WES need to be compatible with other diagnostic methods, like histology and cytology.

100. Is your organization interested in further development of such an approach?



No

Comment

101. In case of R&D involvement, would financial support available in the Instand-NGS4P project be
O not needed
O helpful
 crucial to develop this approach
Let us know if you have any comments and if these are confidential
PROBLEM WGS and WES are difficult for low yield samples (e.g. plasma cfDNA, cfRNA).
SOLUTION:
1. Alternative isolation/stabilisation methods giving highest yield possible
2. Increased sequencing sensitivity
102. Is your organization interested in the further development of such an approach?
O Yes
UN6
Please give the numbers of interest or involvement with the highest priority first, down to the lowest priority:
103. In case of R&D involvement, would financial support available in the Instand-NGS4P project be o not needed
O helpful
C) crucial to develop this approach
Let us know if you have any comments and if these are confidential
1. PROBLEM Isolated nucleic acid preparations can contain interfering substances. This can impair the reliability of the results and the compatibility of isolation methods with multiple sequencing methods.
SOLUTION:
1. The isolates are tested for inhibitory substances for various sequencing methods
2. Development of alternative procedures to avoid known interfering substances
104. Is your organization interested or involved in R&D for such an approach?
Yes
No
Comment

105. In case of R&D involvement, would financial support available in the Instand-NGS4P project be

not needed

helpful

crucial to develop this approach

Let us know if you have any comments and if these are confidential

PROBLEM Long turnaround times (for pre-analytics and library preparation).

SOLUTION: Below you can find a list of possible approaches to solve the problem.

106. Please weigh each of them on the basis of their importance from 1 (lower) to 5 (higher).

	1	2	3	4	5	N/A
Faster nucleic acid isolation methods	\bigcirc	0	0	0	0	\bigcirc
Faster library preparation procedures	\odot	0	0	0	0	\bigcirc
Integration of multiple steps in nucleic acid isolation procedure	\bigcirc	0	0	\odot	\bigcirc	\bigcirc
Integration of multiple steps in library preparation	0	0	0	0	0	\odot
Integration of multiple steps in isolation and library preparation	0	0	0	0	0	\odot
Automation	\bigcirc	0	\odot	0	\odot	0
Other	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	\odot
Other (please specify)						

107. Comment

CHALLENGE 3:

Defining genetic variants with established medical implications for common and rare, adult and pediatric, cancers including pharmacogenetic variants relevant for therapy selection in cancer care

SOLUTION: 1. Increase the efficiency of nucleic acid isolation methods 2. Increased sequencing sensitivity. 108. Is your organization interested or involved in R&D for such an approach?
 Increase the efficiency of nucleic acid isolation methods Increased sequencing sensitivity. 108. Is your organization interested or involved in R&D for such an approach? Yes No Please give the numbers with the highest priority first, down to the lowest priority 109. In case of R&D involvement, would financial support available in the Instand-NGS4P project be not needed helpful crucial to develop this approach Let us know if you have any comments and if these are confidential
 Increased sequencing sensitivity. 108. Is your organization interested or involved in R&D for such an approach? Yes No Please give the numbers with the highest priority first, down to the lowest priority 109. In case of R&D involvement, would financial support available in the Instand-NGS4P project be not needed helpful crucial to develop this approach Let us know if you have any comments and if these are confidential
108. Is your organization interested or involved in R&D for such an approach? Yes No Please give the numbers with the highest priority first, down to the lowest priority 109. In case of R&D involvement, would financial support available in the Instand-NGS4P project be ont needed helpful crucial to develop this approach Let us know if you have any comments and if these are confidential
 Yes No Please give the numbers with the highest priority first, down to the lowest priority 109. In case of R&D involvement, would financial support available in the Instand-NGS4P project be not needed helpful crucial to develop this approach Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs, although it is not known if the drugs are metabolized normally
No Please give the numbers with the highest priority first, down to the lowest priority 109. In case of R&D involvement, would financial support available in the Instand-NGS4P project be not needed helpful crucial to develop this approach Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs, although it is not known if the drugs are metabolized normally
Please give the numbers with the highest priority first, down to the lowest priority 109. In case of R&D involvement, would financial support available in the Instand-NGS4P project be not needed helpful crucial to develop this approach Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs although it is not known if the drugs are metabolized normally.
109. In case of R&D involvement, would financial support available in the Instand-NGS4P project be not needed helpful crucial to develop this approach Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and bazardous drugs, although it is not known if the drugs are metabolized normally.
109. In case of R&D involvement, would financial support available in the Instand-NGS4P project be not needed helpful crucial to develop this approach Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs, although it is not known if the drugs are metabolized normally.
 helpful crucial to develop this approach Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and bazardous drugs although it is not known if the drugs are metabolized normally
 Crucial to develop this approach Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs, although it is not known if the drugs are metabolized normally.
Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs, although it is not known if the drugs are metabolized normally.
Let us know if you have any comments and if these are confidential PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs, although it is not known if the drugs are metabolized normally.
PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs, although it is not known if the drugs are metabolized normally.
PROBLEM The NGS diagnostic result is used for determination of the treatment for patients. This involves the administration of complex and hazardous drugs, although it is not known if the drugs are metabolized normally.
SOLUTION: Including Pharmacogenomic Analysis in panels targeted on somatic mutations
110. Is your organization interested in further development of such an approach?
Yes
No
Comment
111. In case of R&D involvement, would financial support available in the Instand-NGS4P project be
() not needed
) helpful
Crucial to develop this approach
Let us know if you have any comments and if these are confidential

CHALLENGE 4:							
Developing reference material for quality control							
PROBLEM Availability of suitable internal quality control (QC) approaches and reference materials to monitor the NGS workflow as a whole.							
SOLUTION: Multiple QC steps for each phase of the workflow to act as stop/go criteria to avoid wasting time and resources.							
112. Is your organization interested in further development of such an approach?							
No							
Comment							
L							
113. In case of R&D involvement, we	ould financial support available in the	e Instand-NGS4P project be					
not needed							
helpful							
() crucial to develop this approach							
Let us know if you have any comments and if	these are confidential						
PROBLEM Availability of well-designed NGS performance testing (PT), External Quality Assessment (EQA) and reference material.							
SOLUTION: Use of PT, EQA and reference material for NGS workflows							
114 Please select the box if your organization is using or wants to use it.							
		Wants to use					
PT							
EQA							
Reference material							
Please specify what kind and what is your source							
	Yes	No					
---	--	---					
introduced into the orkflow during pre- nalytic processing	\odot	0					
atches authentic ample matrix	0	\odot					
overs diagnostically elevant variants	0	\bigcirc					
cludes RNA	0	0					
ontains 'difficult' ariants Of interest	0	\odot					
mment							
OBLEM Patient identity of patier	t material is sometimes swapped with that of a a lot of extra time.	nother patient. In doubt the material must be re-					
OBLEM Patient identity of patier ntified with ID SNP. This requires ution: Inclusion of patient ID SNI	t material is sometimes swapped with that of a a lot of extra time. P's in the sequence panel or result.	nother patient. In doubt the material must be re-					
OBLEM Patient identity of patier ntified with ID SNP. This requires lution: Inclusion of patient ID SNI 116. Is your organization	t material is sometimes swapped with that of a a lot of extra time. P's in the sequence panel or result. involved in R&D for such an approach	nother patient. In doubt the material must be re-					
OBLEM Patient identity of patier ntified with ID SNP. This requires lution: Inclusion of patient ID SNI 116. Is your organization Yes No	t material is sometimes swapped with that of a a lot of extra time. P's in the sequence panel or result. involved in R&D for such an approach	nother patient. In doubt the material must be re-					
COBLEM Patient identity of patien intified with ID SNP. This requires lution: Inclusion of patient ID SNI 116. Is your organization Yes No Comment	t material is sometimes swapped with that of a a lot of extra time. P's in the sequence panel or result. involved in R&D for such an approach	nother patient. In doubt the material must be re-					
COBLEM Patient identity of patien entified with ID SNP. This requires lution: Inclusion of patient ID SNI 116. Is your organization Yes No Comment	t material is sometimes swapped with that of a a lot of extra time. P's in the sequence panel or result. involved in R&D for such an approach	nother patient. In doubt the material must be re-					
COBLEM Patient identity of patien entified with ID SNP. This requires lution: Inclusion of patient ID SNI 116. Is your organization Yes No Comment	t material is sometimes swapped with that of a a lot of extra time. P's in the sequence panel or result. involved in R&D for such an approach	nother patient. In doubt the material must be re-					
ROBLEM Patient identity of patien entified with ID SNP. This requires lution: Inclusion of patient ID SNI 116. Is your organization Yes No Comment [t material is sometimes swapped with that of a a lot of extra time. P's in the sequence panel or result. involved in R&D for such an approach	nother patient. In doubt the material must be re-					
ROBLEM Patient identity of patier entified with ID SNP. This requires plution: Inclusion of patient ID SNI 116. Is your organization Yes No Comment 117. Is your organization Yes	t material is sometimes swapped with that of a a lot of extra time. 2's in the sequence panel or result. involved in R&D for such an approach	nother patient. In doubt the material must be re-					

118. In case of R&D involvement, would financial support available in the Instand-NGS4P project be

O not needed

) helpful

() crucial to develop this approach

Comment and let us know if your comments are confidential

119. If you have any further comments in the context of pre-analytics and library preparation, please let us know and specify if these comments are confidential

Introduction Lot 2

* 120. Would you like to answer questions related to Lot 2 (Sequencing)?

. 2						
1. How impor	tant is sequenci	ng time for you?	? (0- not importa	int at all, 5-very i	mportant)	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
2. What is you lls?	ur maximum acc	ceptable time fo	r a routine seque	encing run in ho	urs, from library	to variant
3. How impor	tant are over-we	ekend runs for	you? (0- not imp	portant at all, 5-v	ery important)	
0	1	2	3	4	5	N/A
0	0	0	0	0	0	\odot
O Per reage	ents structure					
Per reagoPer infrasFor infras	ents structure tant is reduced h	nands-on time f	or you? (0- not ir	mportant at all, 5	-very important)
 Per reage Per infras How import 0 	ents structure tant is reduced h 1	nands-on time f	or you? (0- not ir 3	mportant at all, 5 4	5-very important 5) N/A
 Per reage Per infrast How import 0 	ents structure tant is reduced h 1	nands-on time f 2	or you? (0- not in 3	mportant at all, 5 4	5-very important 5) N/A
 Per reage Per infras 5. How impor 0 6. How much nds-on time? 	ents structure tant is reduced h 1 0 would you prefe (0- not at all, 5-v	nands-on time f 2 er higher costs f very much)	or you? (0- not in 3 Or reagents and	mportant at all, 5 4 automated solu	5-very important 5 tions in exchanç) N/A O ge for reduce
 Per reage Per infras 5. How impor 0 6. How much nds-on time? 0 	ents structure tant is reduced h 1 0 would you prefe (0- not at all, 5-v 1	nands-on time f 2 er higher costs f very much) 2	or you? (0- not in 3 or reagents and 3	mportant at all, 5 4 automated solu	5-very important 5 tions in exchanç 5) N/A ge for reduce N/A
 Per reage Per infras 6. How much nds-on time? 0 	ents structure tant is reduced h 1 would you prefe (0- not at all, 5-v 1	nands-on time f 2 er higher costs f very much) 2	or you? (0- not in 3 or reagents and 3	mportant at all, 5 4 automated solu 4	5-very important 5 tions in exchang 5) N/A O ge for reduce N/A

,							
⊖ Yes							
No							
If yes, please specify	У						
20 How important	would longer	reade (>600br) he for you?	(0 not import	ant at all 6	verv impo	rtant)
0	1	2	3 3	4	anit at an, c	5	N/A
0	0	0	0	0		0	0
2		5	U	0			U
30. How you impo t all, 5-very importa	rtant is it for yo ant)	ou to read bioc	hemical infor	mation (ex. me	thylation s	tatus)? (0-	not importan
0	1	2	3	4		5	N/A
\odot	0	0	0	0		0	\odot
32. Do you consid	er sequencing	noise a proble	em? (0- not al	t all, 5-very mu	ch)	5	N/A
32. Do you consid	er sequencing 1	noise a proble 2	em? (0- not al 3	t all, 5-very mu 4	ch)	5	N/A
32. Do you conside 0	er sequencing 1	noise a proble 2	em? (0- not al 3	t all, 5-very mu 4	ch)	5	N/A
32. Do you consid 0 33. How big is the	er sequencing 1	noise a proble 2 O emic sequenci	em? (0- not at 3 O	t all, 5-very mu 4 on your work?	ch) (0-no impa	5 O act, 5-very b	N/A O
32. Do you consid 0 33. How big is the	er sequencing 1 impact of syste	noise a proble 2 o emic sequenc	em? (0- not at 3 O ing artefacts o 2	t all, 5-very mu 4 on your work? 3	ch) (0-no impa 4	5 Act, 5-very b 5	N/A O Dig) N/A
 32. Do you consident of the second second	er sequencing 1 impact of syste	noise a proble 2 emic sequenc 1	em? (0- not at 3 ing artefacts o 2	t all, 5-very mu 4 on your work? 3	ch) (0-no impa 4	5 act, 5-very k 5	N/A O Dig) N/A
 32. Do you consident of the second second	er sequencing 1 impact of syste 0	noise a proble 2 emic sequenc 1	em? (0- not at 3 ing artefacts o 2	t all, 5-very mu 4 on your work? 3	ch) (0-no impa 4	5 act, 5-very k 5	N/A Dig) N/A
 32. Do you conside 0 33. How big is the Homopolymers GC rich regions Low complexity regions 	er sequencing 1 impact of syste 0 s	noise a proble 2 emic sequenc 1	em? (0- not at 3 ing artefacts o 2 0	all, 5-very mu 4 on your work? 3	ch) (0-no impa 4	5 act, 5-very k 5	N/A Dig) N/A
 32. Do you conside 0 33. How big is the Homopolymers GC rich regions Low complexity regions 	er sequencing 1 impact of syste 0 s	noise a proble 2 emic sequence 1	em? (0- not at 3 ing artefacts o 2 0	all, 5-very mu 4 on your work? 3	ch) (0-no impa 4	5 act, 5-very k 5	N/A Oig) N/A
 32. Do you conside 0 33. How big is the Homopolymers GC rich regions Low complexity regions 34. How important 	er sequencing 1 impact of syste 0 s is paired-end	noise a proble 2 emic sequence 1 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	em? (0- not at 3 ing artefacts o 2 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	t all, 5-very mu 4 on your work? 3 0 0	ch) (0-no impa 4 0 0 0 0 1 0 1 0 1 0 1 0 1 0 1 0 1 0 1	5 act, 5-very k 5 0 0	N/A)))))))))))))
 32. Do you conside 0 33. How big is the Homopolymers GC rich regions Low complexity regions 34. How important 0 	er sequencing 1 impact of syste 0 s is paired-end 1	noise a proble 2 emic sequence 1 0 0 0 sequencing fo 2	em? (0- not at 3 ing artefacts o 2 0 0 0 0 1 0 1 0 0 0 0 0 0 0 0 0 0 0 0	all, 5-very mu 4 on your work? 3 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	ch) (0-no impa 4 0 0 1 0 1 1 1, 5-very	5 act, 5-very k 5 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	N/A)))))))))))))
32. Do you conside 0 33. How big is the Homopolymers GC rich regions Low complexity regions 34. How important 0	er sequencing 1 impact of syste 0 s is paired-end 1	noise a proble 2 emic sequence 1 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	em? (0- not al 3 ing artefacts o 2 0 0 1 0 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	all, 5-very mu 4 on your work? 3 ot important at 4	ch) (0-no impa 4 0 0 all, 5-very	5 act, 5-very k 5 important) 5	N/A)))))))))))))
 32. Do you conside 0 33. How big is the Homopolymers GC rich regions Low complexity regions 34. How important 0 35. How important 	er sequencing 1 impact of syste 0 s is paired-end 1 0 is the size of t	noise a proble 2 emic sequence 1 0 0 sequencing fo 2 0 the instrument	em? (0- not al 3 ing artefacts o 2 0 0 r you? (0- no 3 for you? (0- no	t all, 5-very mu 4 on your work? 3 ot important at 4 ot important at	ch) (0-no impa 4 0 all, 5-very	5 act, 5-very b 5 important) 5 y important	N/A) N/A N/A N/A)
 32. Do you conside 0 33. How big is the Homopolymers GC rich regions Low complexity regions 34. How important 0 35. How important 0 	er sequencing 1 impact of syste 0 s is paired-end 1 is the size of t 1	noise a proble 2 emic sequence 1 0 0 sequencing fo 2 0 the instrument 2	em? (0- not at 3 ing artefacts o 2 0 0 1 7 you? (0- no 3 for you? (0- no 3 0 1 3	all, 5-very mu 4 on your work? 3 ot important at 4 not important at 4	ch) (0-no impa 4 0 all, 5-very t all, 5-ver	5 act, 5-very k 5 important) 5 y important 5	N/A Dig) N/A N/A N/A N/A

136. How importa important)	ant is flexibility	of the platform f	or scalable thro	ughput for you?	(0- not importan	t at all, 5-very
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
137. How importa	ant is it for you	to pool various l	ibraries in one r	un? (0- not impc	ortant at all, 5-ve	ry important)
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
138. Do you p O ^{Yes}	erform sequen	cing as a lab-de	veloped test?			
No						0
39. How importa	ant are IVDR-C	E certified tests	for you? 0- not	important at all,	5-very importan	t)
U	1	2	3	4	5	N/A
0	0	\cup	\cup	0	0	U
140. How importa	ant are IVDR-C	E instruments fo	or you? 0- not in	nportant at all, 5-	very important)	N/A
U	1	2	3	4	5	N/A
U	0	\cup	\cup	0	0	\cup
PROBLEM High nucl always able to deal w SOLUTION: 1. Increase the ef 2. Increased sequ	eic acid yields are vith difficult genomi ficiency of nucleic uencing sensitivity.	needed as input for c regions, low-yield acid isolation metho	NGS whereas ofte materials and soma	n only small sample: atic mutations at low	s are available and variant allele freque	NGS is not ency.
141. Is your of Yes No 142. Please give	rganization inte the numbers w	erested or involv	ed in R&D for s priority first, dow	uch an approach n to the lowest p	? riority.	

144. If you have any further comments in the context of sequencing, please let us know.

145. Is the information above confidential?

Introduction Lot 3

* 146. Would you like to answer questions related to Lot 3 (Bioinformatics)?

ot 3						
	ranization / or		positioned in the	field of Next (Concretion Sec	
Personalized Thera	by? (Please s	elect all relevar	nt points). Are y	ou a?	Selleration Sec	Juencing (NGS)
Sequencing facil	ty					
Computational fac	ility					
Bioinformatics sof	tware developer					
Software service p	provider					
Diagnostic facility						
Research facility						
Commercial comp	any					
Other (please spe	cify)					
48. Are you currently v Please choose all relev 0-60%, 4 = 60-80%, 5	using results f vant points an = more than	rom NGS bioin nd estimate per 80%)	formatics analy centage) (0 = 0	sis for /not at all, 1 =	up to 20%, 2 =	20-40%, 3 =
48. Are you currently o Please choose all relev 0-60%, 4 = 60-80%, 5	using results f vant points an = more than 0	from NGS bioin nd estimate per 80%) 1	formatics analy centage) (0 = 0 2	sis for /not at all, 1 = 3	up to 20%, 2 = 4	20-40%, 3 = 5
48. Are you currently o Please choose all relev 0-60%, 4 = 60-80%, 5 Diagnostic	using results f vant points an = more than 0	from NGS bioin nd estimate per 80%) 1	formatics analy centage) (0 = 0 2	sis for /not at all, 1 = 	up to 20%, 2 = 4	20-40%, 3 = 5
48. Are you currently o Please choose all relev 0-60%, 4 = 60-80%, 5 Diagnostic Research	using results f vant points an = more than 0	from NGS bioin nd estimate per 80%) 1	formatics analy centage) (0 = 0 2	sis for /not at all, 1 = 	up to 20%, 2 = 4	20-40%, 3 = 5
48. Are you currently of Please choose all relev 0-60%, 4 = 60-80%, 5 Diagnostic Research Software development	using results f vant points an = more than 0	From NGS bioin ad estimate per 80%) 1 	formatics analy centage) (0 = 0 2 	sis for /not at all, 1 = 3 	up to 20%, 2 = 4	20-40%, 3 = 5
48. Are you currently of Please choose all relev 0-60%, 4 = 60-80%, 5 Diagnostic Research Software development ther (please specify)	using results f vant points an = more than 0	From NGS bioin ad estimate per 80%) 1	formatics analy centage) (0 = 0 2	sis for /not at all, 1 = 3 	up to 20%, 2 = 4	20-40%, 3 = 5
48. Are you currently of Please choose all relev 0-60%, 4 = 60-80%, 5 Diagnostic Research Software development ther (please specify)	using results f vant points an = more than 0	From NGS bioin ad estimate per 80%) 1 	formatics analy centage) (0 = 0 2	sis for /not at all, 1 = 	up to 20%, 2 =	20-40%, 3 = 5
48. Are you currently of Please choose all relev 0-60%, 4 = 60-80%, 5 Diagnostic Research Software development ther (please specify)	using results f vant points an = more than 0	irom NGS bioin nd estimate per 80%) 1 	formatics analy centage) (0 = 0 2 	sis for /not at all, 1 = 3	up to 20%, 2 =	20-40%, 3 = 5
48. Are you currently of Please choose all relev 0-60%, 4 = 60-80%, 5 Diagnostic Research Software development ther (please specify) 149. From which typ	using results f vant points an = more than 0 0	is NGS data da	formatics analy centage) (0 = 0 2 	sis for /not at all, 1 = 3 	up to 20%, 2 = 4	20-40%, 3 =
48. Are you currently of Please choose all relevel 0-60%, 4 = 60-80%, 5 Diagnostic Research Software development ther (please specify) 149. From which typ I have already rep	using results f vant points an = more than 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	is NGS data da	formatics analy centage) (0 = 0 2 	sis for /not at all, 1 = 3 	up to 20%, 2 = 4	20-40%, 3 =
 48. Are you currently of Please choose all relevent of the origination of the or	using results f vant points an = more than 0 0	is NGS data da	formatics analy centage) (0 = 0 2 	sis for /not at all, 1 = 3 	up to 20%, 2 = 4	20-40%, 3 =
 48. Are you currently of Please choose all relevent of the observation of the ob	using results f vant points an = more than 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	from NGS bioin ad estimate per 80%) 1 3 3 3 4 3 5 5 6 6 7 6 7 7 7 7 7 7 7 7 7 7 7 7 7 7	formatics analy centage) (0 = 0 2 	sis for /not at all, 1 = 3 	up to 20%, 2 = 4 	20-40%, 3 = 5
48. Are you currently of Please choose all releve 0-60%, 4 = 60-80%, 5 Diagnostic Research Software development ther (please specify) 149. From which typ 149. From which typ 50. FFPE	using results f vant points an = more than 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	From NGS bioin ad estimate per 80%) 1 3 3 4 5 5 5 6 6 7 6 7 7 7 7 7 7 7 7 7 7 7 7 7	formatics analy centage) (0 = 0 2 	sis for /not at all, 1 = 3 	up to 20%, 2 = 4 	20-40%, 3 = 5
48. Are you currently of Please choose all relevent of the second	using results f vant points an = more than 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	irom NGS bioin ad estimate per 80%) 1 is NGS data da ing questions in Lo	formatics analy centage) (0 = 0 2 	sis for /not at all, 1 = 3 	up to 20%, 2 =	20-40%, 3 = 5

151. Frozen tissues						
	1	2	3	4	5	N/A
DNA						
RNA						
152 Blood/plasma						
	1	2	3	4	5	N/A
DNA	[
RNA						
cfDNA	[
cfRNA						
153. Fine needle aspira	tes					
	1	2	3	4	5	N/A
BNA						
154. Circulating Tumor	Cells					
	1	2	3	4	5	N/A
DNA	[
RNA						
155. Extracellular vesicl	es	_			_	
DNA	1	2	3	4	5	N/A
RNA						
156. Saliva						
	1	2	3	4	5	N/A
DNA						

RNA			

157. Other						
	1	2	3	4	5	N/A
DNA						
RNA						
158. If other, pleas	e specify					
150 What is the	a indication that you	u ara looking f	ior a colution fo	rO		
		u are looking i		1 (
	reer					
Rare disease	25					
Other (pleas	e specify)					
O Other (pieas	e specify)					
I						
160. Which type	e of NGS data do y	ou process?				
Targeted see	quencing (Panels)					
Whole Exor	ne Sequencing (WES)					
Whole Gene	ome Sequencing (WGS)				
Gene Expres	ssion (RNA-Seq)					
O ChIP-seq						
Methylation						
Other (pleas	e specify)					
161 Whore are	the raw NCS (upp	recessed) dat	a produced for	the biginformat	ice analysis?	
TOT. Where are					103 di la 19313 :	
(Please choose	all relevant points))				
Inhouse						
Another dep	artment					
External no	n-commercial organizat	ion				
Commercial	provider					
Other (pleas	e specify)					

162. \	Where is the NGS bioinformatics analysis performed?
(Plea	se choose all relevant points)
	At the place of NGS data generation
	Inhouse
	Another department
	External non-commercial organization
	Commercial provider
	Other (please specify)
163. \	Who assesses the results of the NGS bioinformatics analysis for actionable items?
(Plea	se choose all relevant points)
	Inhouse
	Another department
	External non-commercial organization
	Commercial provider
	Other (please specify)
164. I	How do you currently access the raw NGS data for the bioinformatics analysis?
(Pleas	se choose all relevant points)
	No access required to raw data
	Raw data not available
	Available Inhouse
	Available within organization
	Download from (internal / external) sequencing provider
	Other (please specify)

(Piea	ase choose all relevant points)
	By downloading from a server
	By running our solution locally
	Other (please specify)
166.	Which type of software is used for the bioinformatics analysis?
(Plea	ase choose all relevant points)
	Inhouse software (not available)
	open-source software (Non-commercial)
	open-source software (Commercial)
	Commercial software (source code not available)
	Not known
	Other (please specify)
167.	Do you use any workflow manager / workflow description standard? (multiple answer possible)
	Do you use any workflow manager / workflow description standard? (multiple answer possible) No, I only use command line Yes, Galaxy Yes, Common Workflow Language (CWL) Yes, Taverna Yes, Orange Yes, KNIME Yes, Workflow Definition Language (WDL)
	Do you use any workflow manager / workflow description standard? (multiple answer possible) No, I only use command line Yes, Galaxy Yes, Common Workflow Language (CWL) Yes, Taverna Yes, Orange Yes, KNIME Yes, KNIME Yes, Next Flow Yes, Snake Make
	Do you use any workflow manager / workflow description standard? (multiple answer possible) No, I only use command line Yes, Galaxy Yes, Common Workflow Language (CWL) Yes, Taverna Yes, Orange Yes, KNIME Yes, Workflow Definition Language (WDL) Yes, Next Flow Yes, Snake Make Other (please specify)
	Do you use any workflow manager / workflow description standard? (multiple answer possible) No, I only use command line Yes, Galaxy Yes, Common Workflow Language (CWL) Yes, Taverna Yes, Orange Yes, KNIME Yes, Workflow Definition Language (WDL) Yes, Next Flow Yes, Snake Make Other (please specify)
	Do you use any workflow manager / workflow description standard? (multiple answer possible) No, I only use command line Yes, Galaxy Yes, Galaxy Yes, Common Workflow Language (CWL) Yes, Taverna Yes, Orange Yes, KNIME Yes, Workflow Definition Language (WDL) Yes, Next Flow Yes, Snake Make Other (please specify)
	Do you use any workflow manager / workflow description standard? (multiple answer possible) No, I only use command line Yes, Galaxy Yes, Common Workflow Language (CWL) Yes, Taverna Yes, Orange Yes, KNIME Yes, Workflow Definition Language (WDL) Yes, Next Flow Yes, Snake Make Other (please specify)

168. Which file formats are used to exchange data?
◯ FASTQ
BAM / CRAM
○ VCF
⊖ csv
OJSON
Other (please specify)
169. Please indicate the software license (if known)
(Please choose all relevant points)
GNU General Public License
MIT License
Apache License
BSD License
Commercial products
Not sure
Other (please specify)
170. What type of apportations do you currently get?
Clinical actionable items
Drug-gene interactions
Dosage sensitivity
Other (please specify)

171	. What type of annotations do you need? (Please choose all relevant points)
	Variant classification
	Variant population frequency
	Variant Effect prediction
	Disease gene panel
	Disease-gene association
	Clinically actionable items
	Drug-gene interactions
	Dosage sensitivity
	Other (please specify)
172	. Is the annotation information from
(Ple	ase choose all relevant points)
	Inhouse resources
	Findable Accessible Interoperable Reusable (FAIR) data providers
	Free available data
	Commercial data
	Other (please specify)
173	. What kind of QC do you perform for your bioinformatics pipeline?
6	Validation using criteria for accredited labs
0	Regular proficiency testing (benchmarking yourself with other labs, normally required for accredited labs)
	None. This is a task of our service/software provider
C	Other (please specify)

ISO27001:2013 (int	formation secur	ity standard)				
IEC 62304 (Softwar	re Lifecycle)					
🔵 ISO 14155:2011 (C	linical investiga	tion of medical dev	/ices for human su	bjects)		
🔵 ISO 14971 (Risk ma	anagement)					
O ISO13485:2016 (M	edical devices)					
Not sure						
Other (please spec	ify)					
lely applied yet.				,		
75. Which challenges a	are the highe	st immediate p	riority? (scoring	j 0-5)		
75. Which challenges a	are the highe	st immediate p	riority? (scoring 2	3 (0-5)	4	5
75. Which challenges a Automation / Reduced nanual work	are the highe 0	st immediate pi 1 O	riority? (scoring 2	3 3	4	5
75. Which challenges a Automation / Reduced nanual work Reproducibility	are the highe 0 0	st immediate pi	riority? (scoring 2 O	3 3 0	4	5
75. Which challenges a Automation / Reduced nanual work Reproducibility Packaging (e.g. containers)	are the highe	est immediate provide the provide the provide the providence of the providence of the providence of the provide the providence of the prov	riority? (scoring 2 0	3 3 0 0	4	5
75. Which challenges a Automation / Reduced nanual work Reproducibility Packaging (e.g. containers)	are the highe	est immediate provide the provide the provide the providence of the providence of the providence of the provide the providence of the prov	riority? (scoring	3 3 0 0 0	4	5
75. Which challenges a Automation / Reduced nanual work Reproducibility Packaging (e.g. containers) Portability Long-term availability	are the highe	est immediate provide the provided the p	riority? (scoring	3 3 0 0 0 0	4	5
75. Which challenges a Automation / Reduced nanual work Reproducibility Packaging (e.g. containers) Portability Long-term availability	are the highe	est immediate provide the providet the pro	riority? (scoring	3 3 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	4 0 0 0 0 0 0 0 0 0	5 () () () () () () ()
25. Which challenges a Automation / Reduced nanual work Reproducibility Packaging (e.g. containers) Portability cong-term availability cong-term naintainability Standardized data exchange	are the highe	est immediate province of the second se	riority? (scoring 2 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	3 () () () () () () () () () () () () ()	4 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	5 () () () () () () () () ()
 25. Which challenges a Automation / Reduced nanual work Reproducibility Packaging (e.g. containers) Portability cong-term availability cong-term naintainability Standardized data exchange Standardized data storage formats 	are the highe	est immediate pi	riority? (scoring 2 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	3 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	4 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	5 00 00 00 00 00 00 00 00
25. Which challenges a Automation / Reduced nanual work Reproducibility Packaging (e.g. containers) Portability ong-term availability ong-term availability cong-term naintainability Standardized data exchange Standardized data torage formats	are the highe		riority? (scoring 2 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	3 () () () () () () () () () () () () ()	4 0 0 0 0 0 0 0 0 0 0 0 0 0	5 () () () () () () () () () () () () ()

176. Which challenges are the highest long-term priority? (scoring 0-5)						
	0	1	2	3	4	5
Automation / Reduced manual work	\bigcirc	0	\bigcirc	\bigcirc	0	0
Reproducibility	\odot	\odot	\odot	\bigcirc	\odot	\bigcirc
Packaging (e.g. containers)	\odot	0	0	\bigcirc	0	0
Portability	\odot	\odot	\odot	\bigcirc	\odot	\bigcirc
Long-term availability	\odot	\odot	\odot	\odot	\bigcirc	0
Long-term maintainability	\odot	0	0	\odot	\odot	\bigcirc
Standardized data exchange	\bigcirc	0	\bigcirc	0	0	0
Standardized data storage formats	\odot	\odot	0	0	0	\odot
Encryption / data security	\odot	0	\odot	\odot	0	0
Other (please specify)						

177. Which challenges are the highest long-term priority? (scoring 0-5)

	0	1	2	3	4	5
Automation / Reduced manual work	\odot	\odot	\bigcirc	\bigcirc	0	0
Reproducibility	\odot	\odot	\odot	0	\odot	0
Packaging (e.g. containers)	0	\odot	\odot	\odot	0	0
Portability	\odot	\odot	\odot	\bigcirc	0	\bigcirc
Long-term availability	\odot	0	0	\odot	\odot	\odot
Long-term maintainability	\odot	\odot	0	0	\bigcirc	0
Standardized data exchange	\odot	\odot	\odot	\odot	\odot	0
Standardized data storage formats	\odot	\odot	\odot	\odot	0	0
Encryption / data security	\odot	\odot	\odot	\odot	0	0
Other (please specify)						

178. Are there specific challenges to apply NGS bioinformatics analysis in your diagnostics service?

0	No
0	Yes, lack of bioinformatics staff
0	Yes, lack of bioinformatics training
0	Yes, lack of computational processing
0	Yes, lack of long-term storage
0	Yes, unsure which solutions fits our needs best
0	Yes, other (please specify)

179. What kind of training / capacity building activities in the field of bioinformatics do you consider most suitable? (score each 0-5)

	0	1	2	3	4	5
face-to-face	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc
online training courses	\bigcirc	\bigcirc	\odot	\odot	\odot	\bigcirc
online training materials (asynchronous tutorials, videos, presentations).	0	0	0	0	0	\odot
books and/or scientific papers about the subject	0	0	0	\odot	\odot	0

Software and pipelines

Automation offers the execution of different software products at different steps, which allows a standardized and reproducible analysis at different places. This can include different software solutions but ultimately allows harmonizing the detection of genomic events.

180. What do you believe are the key challenges to overcome in relation to introducing standardized pipelines and software to detect actionable items for diagnostics purposes? Score each (0-5)

	0	1	2	3	4	5
getting commercial support for open source products	0	\odot	0	0	0	0
long-time support for commercial products	\odot	\odot	\odot	0	0	0
troubleshooting problems with products	0	\bigcirc	0	\bigcirc	0	0
usability	\odot	\odot	\odot	\odot	\odot	\bigcirc
reproducibility	\odot	\bigcirc	\odot	\odot	\odot	\odot
complying with standard data formats	\odot	\odot	\odot	0	\odot	0
adjusting workflows	\odot	\odot	\odot	\odot	\odot	\bigcirc
integration in open- source pipeline systems	\odot	\odot	0	\bigcirc	\bigcirc	0
testing of pipelines with standardized samples	0	\odot	0	\bigcirc	0	0
scalability to whole genome sequencing	0	\odot	0	0	0	0
organisation of shared work on the data	\bigcirc	\bigcirc	0	\bigcirc	\bigcirc	0
lack of training opportunities	0	\odot	\odot	\bigcirc	\bigcirc	0
Other (please specify)						

Which are in your opinion the relevant standards on this topic and how ready are they to be used in diagnostics? (please rate the relevant standards for readiness 0-5)

181. Software file form	nats and secure	e data exchang	e protocols			
	0	1	2	3	4	5
BAM/SAM/FASTQ format	0	0	\bigcirc	\bigcirc	\bigcirc	\bigcirc
VCF format	\odot	\odot	\odot	\odot	\odot	\bigcirc
gVCF format	\odot	\odot	\odot	\odot	\bigcirc	\bigcirc
GA4GH HTSGet	\odot	\odot	\odot	\odot	\odot	\odot
Encryption (Crypt4GH)	\odot	\odot	\odot	\odot	\odot	\odot
CRAM format	\odot	\odot	\odot	\odot	\odot	\bigcirc
Other (please specify)						

ISO standards

ISO 20397-2:2021 "Biotechnology - Massively parallel sequencing - Part 2: Quality evaluation of sequencing data"

182. Please score the relevance of software license model regarding their use in diagnostics? (score each 0-5)

	0	1	2	3	4	5
Open Source (https://docs.github.com/en/github/creating- cloning-and-archiving- repositories/licensing-a- repository#choosing-the-right-license)	0	0	0	0	0	0
Closed source	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
BSD 3	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Apache	\odot	\bigcirc	\odot	\odot	\odot	\odot
CC	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot
GNU GPL (v1,v2, v3)	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot
OSL	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
MIT	\odot	\odot	\bigcirc	\odot	\odot	\bigcirc
Other (please specify)						

Annotation resources

The annotation of variants or genomic events is fundamental to identify actionable items.

diagnostics purposes? Updating external resources Standardized list of approved annotation resources Quality of resources (CE marking, ISO standard) Lifetime of resource Size of resource Licensing of resources Other (please specify)	183. What do you believe are the key challenges to overcome the standardized use of annotation resources fo
 Updating external resources Standardized list of approved annotation resources Quality of resources (CE marking, ISO standard) Lifetime of resource Size of resource Licensing of resources Other (please specify) 	diagnostics purposes?
 Standardized list of approved annotation resources Quality of resources (CE marking, ISO standard) Lifetime of resource Size of resource Licensing of resources Other (please specify) 	Updating external resources
Quality of resources (CE marking, ISO standard) Lifetime of resource Size of resource Licensing of resources Other (please specify)	Standardized list of approved annotation resources
Lifetime of resource Size of resource Licensing of resources Other (please specify)	Quality of resources (CE marking, ISO standard)
Size of resource Licensing of resources Other (please specify)	Lifetime of resource
Licensing of resources Other (please specify)	Size of resource
Other (please specify)	Licensing of resources
	Other (please specify)

Data storage and sharing

The storing and sharing of data is an integral part of a bioinformatics pipeline. The types of data include raw NGS data, result files from the pipeline as well as metadata.

Finally, it needs to be decided which data should be stored, where should they be stored, for how long and who should have access to?

184. What do you believe are the key challenges to overcome in relation to storing & sharing of relevant data for diagnostics purposes? score each (0-5)

	0	1	2	3	4	5
Location of storage (GDPR)	\bigcirc	0	0	0	0	0
GA4GH standards	\odot	\odot	\odot	\odot	\odot	\bigcirc
encryption	\bigcirc	\odot	\odot	\bigcirc	0	0
longtime storage	\odot	\bigcirc	\odot	\odot	\bigcirc	0
Patient access	\odot	\bigcirc	0	\bigcirc	0	0
Data size	0	\odot	0	0	0	0

Other (please specify)

185. How long is your legal obligation to store data for patient care?

186. Do you have a solution in place to store the data to address this legal requirement of data storage?

) Yes

No

187. How big is your expected data volume generated in the next 5 years?

○ < 1 PB</p>
○ 1-5 PB

- 🔵 5-10 PB
-) >10 PB

Security

The acquired NGS data in diagnostics are very sensitivity and can identify patients.

188. What do you believe are the key challenges to overcome in relation to the secure handling of NGS data in diagnostics? score each (0-5)

	0	1	2	3	4	5
standardization	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	\odot
awareness of personnel	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot	\odot
training of personnel	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
quality of solutions (CE marked)	\bigcirc	\odot	\odot	\odot	\odot	0
security updates	\bigcirc	0	\odot	\bigcirc	\odot	\odot
GDPR requirements properly addressed to national implementation	0	\odot	\odot	0	\odot	\bigcirc
Other (please specify)						

189. If you have any further comments in the context of bioinformatics analysis, please let us know.

190. Is this information confidential?

-) Yes
- O No

Introduction Lot 4

* 191. Would you like to answer questions related to Lot 4 (Integrated reporting)?

Lot 4

The aim of Lot 4 is to provide innovative solutions for translating NGS results into medical decision-making reports. This should be achieved by integrating NGS results with pharmacogenomics panels and existing e-medication tools containing information on dosing and drug interactions. Since this information has to be made available to healthcare professionals and patients at the bedside for rapid interpretation, it will be important to determine the optimal method to clearly present NGS results and their medical relevance. The relevant clinical information should be reported in a concise and clear way, reporting only data with validated evidence for clinical decisions and in a form minimizing the risk of data misinterpretation. The questionnaire should help defining the specifications of the integrated reporting tool.

192. Do you use tools for integrated reporting?



193. Are you interested in implementing tools for integrated reporting?

O Yes

194. Are you a solution provider for integrated reporting tools?

195. Do you have a solution for integrated reporting of NGS results that integrates the following data and information (multiple answers may apply)

Cancer–related variants	\odot
Actionable items	\odot
Pharmacogenomic O	\odot
Level of evidence for cancer-related variants (level of evidence could be companion- diagnostics, drug-label, guidelines, databases, scientific literature)	\odot
Level of evidence for pharmacogenomic variants (level of evidence could be companion-diagnostics, drug-label, guidelines, databases, scientific literature)	\odot
Information on clinical O	\odot
Other (please specify)	

196. How important is the integration of the following data and information in reporting of NGS results in order to support medical decision making (please select score; 0=not relevant; 5=highly relevant).

	0	1	2	3	4	5	N/A
Information on informed consent	\bigcirc	0	\bigcirc	\bigcirc	0	\odot	0
Information on the sample analyzed	\bigcirc	\odot	\odot	\odot	\odot	\odot	\odot
Information on the analytical method	\bigcirc	\odot	\bigcirc	\bigcirc	0	\bigcirc	0
Information on the quality of the analysis	\odot	\odot	\odot	\odot	\odot	\odot	\odot
Results on cancer – related variants	\bigcirc	\odot	\odot	\bigcirc	\odot	\bigcirc	\odot
Information on actionable items	\bigcirc	\odot	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc
Information on pharmacogenomics variants	\bigcirc	0	\bigcirc	\bigcirc	0	\bigcirc	0
Information on the level of evidence for cancer- related variants	\bigcirc	0	0	0	0	0	0
Information on the level of evidence for pharmacogenomics variants	0	0	\bigcirc	\bigcirc	0	\odot	0
Information on drug-drug interaction, dosing, side effects and contra indications (e.g., e-medication)	0	0	0	0	0	\odot	0
Information on relevant clinical data (e.g., heart, liver, kidney function)	0	0	0	0	0	0	0
Information on running clinical trials	0	0	0	0	0	\bigcirc	0
Information on possible compassionate use	0	0	0	0	0	0	0
Other (please specify)							

197. Please indicate how important is the following information on clinical evidence for decision making. Evidence based on the following facts (please select score; 0=not relevant; 5=highly relevant).

	0	1	2	3	4	5	N/A
Companion-diagnostics	\odot	\odot	\bigcirc	\bigcirc	\bigcirc	\odot	\odot
Drug-label	\bigcirc	\odot	\bigcirc	\bigcirc	\odot	\odot	\bigcirc
Guidelines of medical societies	\bigcirc	0	\bigcirc	\bigcirc	\odot	\bigcirc	0
Curated databases	\bigcirc	\odot	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc
Scientific literature	\bigcirc	\odot	\bigcirc	\bigcirc	\odot	\bigcirc	\odot
Information on open clinical trials – national	\bigcirc	0	0	\bigcirc	0	\bigcirc	\bigcirc
Information on open clinical trials – Europe	\bigcirc	0	0	\bigcirc	0	\bigcirc	\odot
Information on open clinical trials – international	\bigcirc	0	\bigcirc	\bigcirc	\odot	\odot	\bigcirc

What is your preferred reference for assessing the clinical evidence? Please describe

198. How important are the following features of integrated reporting for decision support

(please select score; 0=not relevant; 5=highly relevant)

	0	1	2	3	4	5	N/A
Desktop solution	\odot	\odot	\bigcirc	\bigcirc	\odot	\odot	\odot
Mobile device (e.g., tablet 12')	\bigcirc	\odot	0	\bigcirc	\odot	\odot	\odot
Touch screen	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	\odot
WLAN	\odot	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Off-line mode	0	\odot	\odot	\odot	\odot	\odot	0
Other (please specify)							

199. How important is graphical presentation (data visualization) of results for decision support? (please select score; 0=not relevant; 5=highly relevant)

0	1	2	3	4	5	N/A
\bigcirc	0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0

If yes, please describe the solution 1. How important is it that the results of integrated reporting are also integrated in electronic health records HR) and the hospital information systems (HIS)? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A elevance of integration EHR elevance of integration HIS 2. What hospital information system are you using? ease describe: 203. Have you tested different products for integrated reporting and decision support? Yes No I f yes please describe which products 4. How are you satisfied with current products on the market for integrated reporting and decision support? 5. How important is it that the integrated reporting and decision support system generates a special report patients? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A	No							
1. How important is it that the results of integrated reporting are also integrated in electronic health records HR) and the hospital information systems (HIS)? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A 203. Have you tested different products for integrated reporting and decision support? Yes No 1	If yes, please describ	e the solution						
1. How important is it that the results of integrated reporting are also integrated in electronic health records HR) and the hospital information systems (HIS)? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A Velevance of integration 0 1 2 3 4 5 N/A Velevance of integration 0 1 2 3 4 5 N/A Velevance of integration 0 1 2 3 4 5 N/A Velevance of integration 0								
1. How important is it that the results of integrated reporting are also integrated in electronic health records HR) and the hospital information systems (HIS)? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A Relevance of integration EHR 0 1 2 3 4 5 N/A Relevance of integration HIS 0								
Image: constraint of the products 0 1 2 3 4 5 N/A Image: constraint of integration 0 1 2 3 4 5 N/A Image: constraint of integration 0 1 2 3 4 5 N/A Image: constraint of integration 0 1 2 3 4 5 N/A Image: constraint of integration 0	1. How important	is it that the res	sults of integ	rated reporting	g are also integ	rated in el	lectronic he	alth records
0 1 2 3 4 5 N/A Relevance of integration BHR 0 0 0 0 0 0 2. What hospital information system are you using? 2. A have you tested different products for integrated reporting and decision support? Yes No 1 2 3 4 5 1 2 3 4 5 1 2 3 4 5 1 2 3 4 5 5 1 2 3 4 5 1 2	HR) and the hospi	tal information	systems (HI	5)? (please se	elect score; U=r	not relevar	nt; 5=nigniy	relevant)
televance of integration n EHR televance of integration HIS 2. What hospital information system are you using? telease describe: 203. Have you tested different products for integrated reporting and decision support? Yes No If yes please describe which products If yes please describe which products on the market for integrated reporting and decision support? 4. How are you satisfied with current products on the market for integrated reporting and decision support? ease select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5< NvA 5. How important is it that the integrated reporting and decision support system generates a special report repatients? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5< NVA 0 1 2 3 4 5< NVA 6 1 2 3 4 5< NVA 6 1 2 3 4 5 No 1 2 3 4 5 NVA 6 7 7 7 8 7 7 8 7 7 8 7 8 7 8 7 7 8 7 8 7 8 8 8 8 9 9 9 9 9 9 9 10		0	1	2	3	4	5	N/A
elevance of integration hHIS 2. What hospital information system are you using? ease describe: 203. Have you tested different products for integrated reporting and decision support? Yes No Yes No Hoy are you satisfied with current products on the market for integrated reporting and decision support? 4. How are you satisfied with current products on the market for integrated reporting and decision support? 4. How are you satisfied with current products on the market for integrated reporting and decision support? 5. How important is it that the integrated reporting and decision support system generates a special report repatients? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A 0 1 2 3 4 5 N/A	televance of integration EHR		0	0	0	0	0	Ō
 2. What hospital information system are you using? lease describe: 203. Have you tested different products for integrated reporting and decision support? Yes No If yes please describe which products 4. How are you satisfied with current products on the market for integrated reporting and decision support? ease select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A 5 How important is it that the integrated reporting and decision support system generates a special report patients? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A 	elevance of integration HIS	n O	\bigcirc	0	0	0	\bigcirc	\odot
No If yes please describe which products 4. How are you satisfied with current products on the market for integrated reporting and decision support? ease select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A 5. How important is it that the integrated reporting and decision support system generates a special report relevant; 5=highly relevant) 0 1 2 3 4 5 N/A 5. How important is it that the integrated reporting and decision support system generates a special report relevant; 5=highly relevant) 0 1 2 3 4 5 N/A	lease describe:							
4. How are you satisfied with current products on the market for integrated reporting and decision support? 0 1 2 3 4 5 N/A 0 1 2 3 4 5 O 5. How important is it that the integrated reporting and decision support system generates a special report relevant; 5=highly relevant) 0 1 2 3 4 5 N/A	lease describe: 203. Have you te	sted different p	products for in	ntegrated repo	orting and decis	sion suppo	ort?	
4. How are you satisfied with current products on the market for integrated reporting and decision support? lease select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A 0 1 2 3 4 5 O 0 1 2 3 4 5 O 0 1 2 3 4 5 O 0 1 2 3 4 5 N/A	lease describe: 203. Have you te Ves No	sted different p	products for in	ntegrated repo	orting and decis	sion suppo	ort?	
4. How are you satisfied with current products on the market for integrated reporting and decision support? 0 1 2 3 4 5 N/A 0 1 2 3 4 5 O 5. How important is it that the integrated reporting and decision support system generates a special report relevant; 5=highly relevant; 0 1 2 3 4 5 N/A	lease describe: 203. Have you te Ves No If yes please d	sted different p	products for in	ntegrated repo	orting and decis	sion suppo	ort?	
 4. How are you satisfied with current products on the market for integrated reporting and decision support? a 1 2 3 4 5 N/A b 1 2 3 4 5 O/A c 1 2 3 4 5 O/A c 2 3 4 5 O/A c 2 3 4 5 O/A 	lease describe: 203. Have you te Ves No	sted different p	products for in	ntegrated repo	orting and decis	sion suppo	ort?	
ease select score; 0=not relevant; 5=highly relevant)012345N/A0000000005. How important is it that the integrated reporting and decision support system generates a special report r patients? (please select score; 0=not relevant; 5=highly relevant)012345N/A012345N/A00000	203. Have you ter Yes No	sted different p	broducts for in	ntegrated repo	orting and decis	sion suppo	ort?	
012345N/A0000000005. How important is it that the integrated reporting and decision support system generates a special report r patients? (please select score; 0=not relevant; 5=highly relevant)012345N/A012345N/A00000	ease describe: 203. Have you ter Ves No If yes please d	sted different p escribe which prod	oroducts for in ducts	ntegrated repo	orting and decis	sion suppo	ort? g and decisi	on support?
5. How important is it that the integrated reporting and decision support system generates a special report patients? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A	ease describe: 203. Have you te: Ves No If yes please d	sted different p escribe which prod atisfied with cur ; 0=not relevan	oroducts for ir ducts rrent products t; 5=highly re	ntegrated repo s on the mark elevant)	orting and decis	sion suppo	ort? g and decisi	on support?
5. How important is it that the integrated reporting and decision support system generates a special report relevant; 5=highly relevant; 0 1 2 3 4 5 N/A 0 0 0 0 0 0 0 0 0	203. Have you ter Yes No If yes please d 4. How are you sa ease select score; 0	sted different p lescribe which prod atisfied with cur ; 0=not relevan 1	oroducts for in ducts t; 5=highly re 2	ntegrated repo s on the mark elevant) 3	orting and decis et for integrated	sion suppo	ort? 9 and decisi 5	on support? N/A
b. How important is it that the integrated reporting and decision support system generates a special report patients? (please select score; 0=not relevant; 5=highly relevant) 0 1 2 3 4 5 N/A 0 0 0 0 0 0 0 0 0 0 0	lease describe: 203. Have you ter Yes No If yes please d 4. How are you sa ease select score; 0	sted different p lescribe which prod atisfied with cur ; 0=not relevan 1	oroducts for in ducts t; 5=highly re 2	ntegrated repo s on the mark elevant) 3	et for integrated	sion suppo	ort? g and decisi 5	on support? N/A
0 1 2 3 4 5 N/A 0	lease describe: 203. Have you tes Yes No If yes please d 4. How are you sa lease select score; 0	sted different p lescribe which prod atisfied with cur ; 0=not relevan 1	oroducts for in ducts t; 5=highly re 2	ntegrated repo s on the mark elevant) 3	et for integrated	sion suppo d reporting	ort? g and decisi 5	on support? N/A
	 lease describe: 203. Have you teating of the second se	sted different p lescribe which prod atisfied with cur ; 0=not relevan 1 is it that the int	egrated repo	ntegrated reports s on the mark elevant) 3 orting and dec	et for integrated 4 ision support sy	sion suppo d reporting ystem gen	ort? g and decisi 5 erates a spe	on support? N/A
	203. Have you ter Yes No If yes please d 4. How are you sa ease select score; 0 5. How important patients? (please 0	sted different p escribe which prod attisfied with cur ; 0=not relevan 1 1 is it that the int select score; (egrated repo D=not relevar	ntegrated reports s on the mark elevant) 3 orting and decint; 5=highly re	et for integrated 4 ision support sy levant)	sion suppo d reporting	ort? g and decisi 5 erates a sp	on support? N/A ecial report
	 lease describe: 203. Have you test of Yes of No If yes please describe: 4. How are you sate ase select score; 0 5. How important patients? (please 0 	sted different p escribe which prod atisfied with cur ; 0=not relevan 1 is it that the int select score; (1	egrated repo D=not relevan 2	ntegrated repo s on the mark elevant) 3 orting and dec nt; 5=highly re 3	et for integrated 4 sion support sy levant) 4	sion suppo d reporting	ort? g and decisi 5 erates a spo 5	on support? N/A ecial report N/A

206. How important is it that the data analysis for integrated reporting is performed locally or via a webservice? (please select score; 0=not relevant; 5=highly relevant)							
	0	1	2	3	4	5	N/A
Local data analysis	\odot	\odot	\bigcirc	\bigcirc	0	\odot	\odot
Data analysis via webservice	\bigcirc	\odot	\bigcirc	\bigcirc	0	0	\bigcirc

207. If you have any further comments in the context of integrated reporting and e-medication, please let us know.

208. Is this information confidential?

O Yes O No

Last question

209. Would you like to share with us any additional information related to any section of the questionnaire? If yes please use the free field to do so.

210. Is the provided information confidential?

O Yes

O No

PATIENTS' NEEDS QUESTIONNAIRE Instand-NGS4P

Welcome to My Survey

The questionnaire arises from an EU funded project called INSTAND-NGS4P. The aim of the questionnaire is to investigate the current standard of knowledge on Next Generation Sequencing (NGS), which is one piece of the puzzle to enable the development of individual treatments for every patient, often known as personalized medicine or precision medicine.

Personalized medicine aims to tailor therapies for every patient to allow optimized therapies to ensure better patient care. One step in this direction is the application of NGS, which allows the detailed analysis and interpretation of the human genome.

But what does NGS mean for a patient? NGS is a new technology to sequence the genome of any individual. Therefore, the DNA (deoxyribonucleic acid) of a person is extracted from cells, like blood cells or tissue samples. The DNA is made of so-called nucleotides, which are A's (adenosine), T's (thymine), C's (cytosine) and G's (guanine). Thereby, it encodes all our hereditary information and around 22,000 genes, which make us to the person we are. Sequencing is applied to determine all these single nucleotides of a person. This information is then used to be compared to DNA of other individuals (also named reference DNA). This allows the identification of so-called mutations, which are positions where a human subject does not have the same nucleotide as the reference DNA. Such mutations can be meaningless, but they can also provide highly valuable information for physicians and geneticists. Mutations can cause diseases, but they can also give a hint as to how well a person might respond to a therapy or how well a certain drug works. Therefore, these sequencing data also help to gain such knowledge. Imagine having plenty of such DNA sequencing information of patients with certain cancers, but also healthy individuals. Comparing and interpreting their genomes can lead to new discoveries on mutations they share. This can be further used to understand diseases. But patients might also share mutations that help to learn whether a medication will work or fail in a patient, which is the research area of pharmacogenetics – the interplay of genetics and pharmacology. When clinicians use pharmacogenetic data for therapy decision making it means that for some mutations there is information about how well cancer cells with the specific mutation respond to different therapies, and they use it when deciding for a treatment option. If positive or negative response is predicted based on the mutations in the patient at diagnosis, the therapy of choice can offer the patient better chances for response to therapy and spare the patient from side effects of therapies which would not kill the cancer cells.

The personal information collected through the questionnaire will be exclusively used for the purposes of the project, will not be shared with third parties, and will be deleted after the closure of the project.

The following questions aim to address the current level of knowledge on what NGS means, what advantages it has, but also which concerns might arise among patients.

The questionnaires also allow disclosure of confidential information to the project consortium, which will be treated with high confidentiality. Aggregated data from the results of the survey will potentially be published in an anonymous way, excluding the information labelled as confidential.

Filling in the questionnaire will take less than 20 minutes of your time. Please submit the questionnaire by May 31st, 2021. Thank you in advance for contributing.

PATIENTS' NEEDS QUESTIONNAIRE Instand-NGS4P

Organisation profile

* 1. Organisation profile

Organization name	
Address	
City	
Country	
* 2. Contact person	
First name	

Family name	
Current position in the organisation	
E-mail	
Telephone	

- * 3. Does your organization represent adult or childhood cancer patients?
 - Childhood
 - Adults

Adult and childhood

		HILDHOOD PATIENTS QUESTIONNAIRE						
General questions about NGS – addressed to your organization								
lease answer the	following questions,	to the best of your	knowledge, on beha	lf of your organizatio	on.			
* 4. Do you ł	now what Next	Generation Sec	uencing (NGS)	means?				
⊖ Yes								
O No								
 Not sure 								
5. Does your a	association have	knowledge abo	out NGS?					
Please answer	on a scale from	0 to 5 (0 - not at	t all: 5 - a lot: N//	A - not sure)				
0	1	2	3	4	5	N/A		
0	0	0	0	0	0	0		
	on a scale from	0 10 3 10 - HOLAI	i all: 5 - a lot: N/A	A - nousure).				
0	n a scale from	2 נט ט ט ט ט ט ט ט ט ט ט ט ט ט ט ט ט ט ט	ali; 5 - a lot; N/A 3	4 - not sure).	5	N/A		
0	1	2	3	4	5	N/A		
0 O	1		all; 5 - a lot; N/A 3	4 4	5	N/A		
0 7. How much	1 O experience does	2 your organizati	t all; 5 - a lot; N/A 3 O	4 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	5	N/A		
0 7. How much Please answer	1 experience does	2 your organizati 0 to 5 (0 - not at	ion have with NC	4 4 GS? A - not sure).	5	N/A		
0 7. How much Please answer 0	1 experience does on a scale from 1	2 your organizati 0 to 5 (0 - not at 2	t all; 5 - a lot; N/A 3 ion have with NC t all; 5 - a lot; N/A 3	4 GS? A - not sure). 4	5	N/A O N/A		
0 7. How much Please answer 0	1 experience does on a scale from 1	2 your organizati 0 to 5 (0 - not at 2	ion have with NC 3 ion have with NC t all; 5 - a lot; N/A 3	4 GSS? A - not sure). 4	5 5	N/A O N/A		
0 7. How much Please answer 0 8. Is your orga	1 experience does on a scale from 1 1 unization prepare	2 your organizati 0 to 5 (0 - not at 2 ed to advice and	ion have with NC all; 5 - a lot; N/A t all; 5 - a lot; N/A 3 I help patients ar	4 GS? A - not sure). 4 d/or their paren	5 5 ts to answer qu	N/A O N/A estions		
0 7. How much Please answer 0 8. Is your orga	1 experience does on a scale from 1 unization prepare	2 your organizati 0 to 5 (0 - not at 2 ed to advice and	ion have with NC all; 5 - a lot; N/A t all; 5 - a lot; N/A 3 I help patients ar	4 GS? A - not sure). 4 nd/or their paren	5 5 ts to answer qu	N/A N/A estions		
0 7. How much Please answer 0 8. Is your orga egarding NGS	1 experience does on a scale from 1 unization prepare	2 2 3 your organizati 0 to 5 (0 - not at 2 ed to advice and 0 to 5 (0 - not at	ion have with NC all; 5 - a lot; N/A all; 5 - a lot; N/A a l help patients ar t all; 5 - a lot; N/A	4 GS? A - not sure). 4 hd/or their paren A - not sure).	5 5 ts to answer qu	N/A N/A estions		
0 7. How much Please answer 0 8. Is your orga egarding NGS Please answer 0	1 experience does on a scale from 1 unization prepare	2 2 3 your organizati 0 to 5 (0 - not at 2 ed to advice and 0 to 5 (0 - not at 2	t all; 5 - a lot; N/A 3 ion have with NC t all; 5 - a lot; N/A 3 I help patients ar t all; 5 - a lot; N/A 3	4 GS? A - not sure). 4 nd/or their paren A - not sure). 4	5 5 ts to answer qu	N/A N/A estions		

* 9. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients and/or their parents?

Yes No Not sure

* 10. Has any patient and/or parent asked for information about NGS, pharmacogenetics, or personalized medicine?

0	Yes	
0	No,	never

Not sure

11. Please share with us any other thoughts on NGS in the context of your organization:

II. General questions about NGS - addressed to patients and parents

Please answer the following questions, to the best of your knowledge, on behalf of patients and/or parents.

* 12. Do patients and/or parents know what NGS means?

- ⊖ Yes
- O No
- Not sure

* 13. How likely is it that patients and/or parents have heard about NGS in the context of childhood cancer therapy and treatment already?

Please answer on a scale from 0 to 5 (0 – not at all; 5 – very likely; N/A – not sure) 0 1 2 3 4 5 N/A * 14. How well do patients and/or parents know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 - not at all; 5 - very well; N/A - not sure). 0 1 2 3 4 5 N/A
* 15. How well d	o patients and/c	or parents under	stand the benef	it of using NGS i	in cancer diagno	ostics?
Please answer o	on a scale from (0 to 5 (0 – not a	t all; 5 – very we	ell; N/A – not sur	e).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc	\bigcirc
* 16. How well d Please answer o	o you think patie	ents and/or pare 0 to 5 (0 – not a	ents know why N t all; 5 – very we	IGS is an import ell; N/A – not sure	ant diagnostic n e).	nethod?
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	0	\odot	\odot	\bigcirc

No, it needs further clarification

O Not sure

* 18. Do patients and/or parents understand the benefit of selecting a therapy based on the genetic constitution of the patient?

O Yes

) No, it needs further clarification

) Not sure

* 19. Do patients and/or parents know that the individual genetic constitution may affect the efficacy and possible side (and late) effects of certain drugs (also known as pharmacogenetics)?

O Yes

No, it needs further clarification

Not sure

* 20. How likely is it that patients and/or parents understand the consequences regarding the information they get about their genome and how to handle this?

Please answer on a scale from 0 to 5 (0 - not at all; 5 - very well; N/A - not sure).

0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc

* 21. How likely is it and data security?	that patients	and/or parents	understand the	consequences r	egarding the ge	nerated data
Please answer on a	scale from (0 to 5 (0 – not at	all; 5 – very we	ll; N/A – not sure	e).	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 22. Do you thin NGS? Yes No. Patients of Not sure	k patients ar	nd/or parents will it will strictly be use	l have any ethic d for diagnostic pur	al concerns rega poses.	rding the data p	produced by
23. If your answer This informati Possible relev Other (please	er is "yes", pl on is used agai rance for family specify)	ease share: whit	ch ethical conce	erns are you thinl	king of?	
24. What level of e on NGS results? Select all that apply very important; N/A	evidence wor (multiple an – not sure).	uld patients and/ swers possible)	or parents like t and provide a ra	o have in order to anking of importa	o agree on a tre ince from 0-5 (0	eatment based) – not at all; 5-
	0	1	2	3	4	5
NGS is approved for therapeutic decision- making by regulatory authorities	\bigcirc	0	0	0	0	0
Recommended by the medical society	0	0	0	0	0	0
Evidence is published a scientific journal	in O	0	\bigcirc	0	0	0
Experience from other patients or patient	\odot	0	\bigcirc	\odot	advocacy groups	
II. Evaluating the educ	ational require <i>v</i> ing questions f	ements on NGS for rom a patient's pers	patients and/or pa	arents of your knowledge.		

* 25. How importa and/or parents?	ant is thorough	information abo	ut the diagnost	ic procedure, bef	ore sequencing,	, for patients
Please answer or	n a scale from () to 5 (0 – not a	t all; 5 – very in	portant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 26. How importa explanations abo	ant is it for patie ut the procedur	ents and/or pare e?	nts to receive a	a document befor	e sequencing, w	vith detailed
Please answer or	n a scale from () to 5 (0 – not a	t all; 5 – very in	iportant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0
In a docum In a docum * 28. How importa analyses/projects Please answer or	nent separate from nent separate from ant is it for patie the NGS data n a scale from (the informed conse the informed conse ents and/or pare will be used for) to 5 (0 – not a	ent, in a printed for ent, in a digital form ents to receive a ? t all; 5 – very in	^{nat} at document that e nportant; N/A – no	explains for whic ot sure).	h
0	1	2	3	4	5	N/A
\odot	\bigcirc	\odot	\odot	\odot	\odot	\odot
29. If you think As part of the As part of the As part of the As part of the Adocument In a document * 30. How imported a constraint the Adocument * 30. How imported a constraint the Adocument Please answer or 0	t it is important, the informed conse- ment separate from ant is it for patie ntages, but als concerning the I n a scale from (1	what is the preent the informed conse the informed conse ents and/or pare o the impact NC NGS data produ 0 to 5 (0 – not at 2	ferred option ent, in a printed for ent, in a digital form nts to receive of SS might have iced? t all; 5 – very in 3	nat at letailed informatio on their and their nportant; N/A – no 4	on, like a brochu families' lives, a ot sure). 5	rre, that as well as the N/A

on nyou un			eleffed option			
O As part o	of the informed conse	ent				
In a doc	ument separate from	the informed cons	ent, in a printed for	mat		
) In a doci	ument separate from	the informed cons	ent, in a digital form	lat		
32. How impo	rtant is it for patie	ents and/or pare	ents to receive a	a document on ho	ow data security	/ is guaranteed
nd how they ca	an withdraw that	data any time b	based on the Ge	eneral Data Prote	ction Regulatio	n?
ease answer	on a scale from	0 to 5 (0 – not a	ıt all; 5 – very in	nportant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Evaluating the	rights of the patien	ts and parents to	receive the results	s in an appropriate f	format	
e an	following quantions f	from a nationt's nor	anactive to the best			
ease answer the	tollowing questions t	from a patient's per	spective to the best	t of your knowledge.		
Suits ?						
ease answer	on a scale from	0 to 5 (0 – not a	it all; 5 – very in	nportant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
0	1	2	3	4	5	N/A
0	1	2	3	4	5	N/A
0 * 34. Should	1 O patients and/or	2 O parents have th	3 O e choice to rece	4 eive the results?	5	N/A
0 * 34. Should Ves No	1 O patients and/or	2 Darents have th	3 e choice to rece	4 eive the results?	5	N/A
0 * 34. Should O Yes O No O Not sure	1 patients and/or	2 parents have th	3 e choice to rece	4 eive the results?	5	N/A
0 * 34. Should Yes No Not sure * 35. Should	1 patients and/or	2 parents have th	3 e choice to rece	4 eive the results?	5 O different option	N/A
0 * 34. Should Ves No Not sure * 35. Should comprehens	1 patients and/or patients and/or ive the returned	2 parents have th parents have th results should	3 e choice to rece he possibility to be? The compre	4 eive the results? choose between ehensiveness co	5 different option uld reach from	N/A
 a a a a b a b a b a b a a b a a b a a a b a a<	1 patients and/or patients and/or ive the returned regarding the cur	2 parents have the parents have the results should prrent treatment,	3 e choice to rece he possibility to be? The compre to all the detaile	4 eive the results? choose between ehensiveness co ed results of the N	5 different option uld reach from NGS analysis.	N/A
0 * 34. Should Yes No Not sure * 35. Should comprehens results only n Yes	1 patients and/or patients and/or ive the returned regarding the cur	2 parents have the parents have the results should rrent treatment,	3 e choice to rece he possibility to be? The compre to all the detaile	4 eive the results? choose between ehensiveness co ed results of the N	5 different option uld reach from NGS analysis.	N/A
0 * 34. Should Yes No * 35. Should comprehens results only n Yes No No	1 patients and/or patients and/or ive the returned regarding the cur	2 parents have the parents have the results should rrent treatment,	3 e choice to rece he possibility to be? The compre to all the detaile	4 eive the results? choose between ehensiveness co ed results of the N	5 different option uld reach from NGS analysis.	N/A
0 * 34. Should Yes No Not sure * 35. Should comprehens results only r Yes No No Not sure	1 patients and/or patients and/or ive the returned regarding the cur	2 parents have the parents have the results should a rrent treatment,	3 e choice to rece he possibility to be? The compre to all the detaile	4 eive the results? choose between ehensiveness co ed results of the N	5 different option uld reach from NGS analysis.	N/A
0 * 34. Should Yes No Not sure * 35. Should comprehens results only r Yes No No Not sure	1 patients and/or patients and/or ive the returned regarding the cur	2 parents have the parents have the results should a rrent treatment,	3 e choice to rece he possibility to be? The compre to all the detaile	4 eive the results? choose between ehensiveness co ed results of the N	5 different option uld reach from NGS analysis.	N/A
0 * 34. Should Yes No Not sure * 35. Should comprehens results only n Yes No Not sure	1 patients and/or patients and/or ive the returned regarding the cur	2 parents have the parents have the results should the rrent treatment,	3 e choice to rece he possibility to be? The compre to all the detaile	4 eive the results? choose between ehensiveness co ed results of the N	5 different option uld reach from NGS analysis.	N/A
0 * 34. Should Yes No Not sure * 35. Should comprehens results only r Yes No Not sure	1 patients and/or ive the returned regarding the cur	2 parents have the parents have the results should for rrent treatment,	3 e choice to rece he possibility to be? The compre to all the detaile	4 eive the results? choose between ehensiveness co ed results of the N	5 different option uld reach from NGS analysis.	N/A
0 * 34. Should Yes No Not sure * 35. Should comprehens results only n Yes No Not sure	1 patients and/or ive the returned regarding the cur	2 parents have the parents have the results should for rrent treatment,	3 e choice to rece he possibility to be? The compre to all the detaile	4 eive the results? choose between ehensiveness co ed results of the N	5 different option uld reach from NGS analysis.	N/A

* 36. How importations of the	ant is it for the p NGS analysis	oatients and/or p that affect fami	parents that the ly members?	final report con	tains informatior	about those
Please answer or	n a scale from () to 5 (0 – not a	t all; 5 – very im	portant; N/A – r	not sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 37. How importate the discussion of	ant is it for patie the results?	ents and/or pare	ents to have ger	etic consultatio	n assistance dui	ring and after
Please answer or	n a scale from () to 5 (0 – not a	t all; 5 – very im	portant; N/A – r	not sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
 Only in a d Only in a p Only in a p On a mobil * 39. If provide good I Yes No Not sure V. Evaluating the rig with a well-informed * 40. Should for they turn 18? Yes No Not sure 	igital form on a mo rinted form e device and in a p ed in a digital fo evel of data sec hts of a former ch physician, and th ormer childhood	bile device brinted form rm on a mobile curity confidenc hildhood cancer pa heir right to recons d cancer patient	device, does a e to the patient atient to be inform sider	security level si and/or parents?	milar to that use or NGS analysis, a at NGS was app	ed for e-banking second meeting lied, as soon as

41. If yes, how should they be informed?
Per email
Per post
Face to face
Phone call
Other (please specify)

* 42. Should this notification include the offer for an appointment with a well-informed physician, geneticist, and psychologist?

\bigcirc	Yes
\bigcirc	No
0	Not sure

* 43. Should former childhood cancer patients also receive detailed information to have the option of withdrawing their data based on the General Data Protection Regulation, in case they wish to do so?

Yes
 No
 Not sure

* 44. Should the patient have the pharmacogenetic results available on a mobile device or in a printed form?

- Only in a digital form on a mobile device
- Only in a printed form
- On a mobile device and in a printed form

* 45. If provided in a digital form on a mobile device, does a security level similar to that used for e-banking provide a good level of data security confidence to the patient?

) Yes No Not sure

PATIENTS INCLIPSIONNAIRE Instand-NGS4P ADULT PATIENTS QUESTIONNAIRE V General questions about NGS - addressed to your organization Please answer the following questions, to the best of your knowledge, on behalf of your organization. * 46. Does your association have knowledge about NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? 2 3 4 5 N/A * 40. O 1 2									
ADULT PATIENTS QUESTIONNAIRE V. General questions about NGS - addressed to your organization Please answer the following questions, to the best of your knowledge, on behalf of your organization. * 46. Does your association have knowledge about NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? 1 2 3 4 5 N/A * 49. Does your organization have any initiatives and/or material, like brochures, to explain NGS to patients? <td>PATIENTS' I</td> <td>NEEDS QUE</td> <td>STIONNAIRE</td> <td>Instand-NGS</td> <td>S4P</td> <td></td> <td></td>	PATIENTS' I	NEEDS QUE	STIONNAIRE	Instand-NGS	S4P				
V. General questions about NGS - addressed to your organization Please answer the following questions, to the best of your knowledge, on behalf of your organization. * 46. Does your association have knowledge about NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? 1 2 3 4 5 N/A * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes	ADULT PATIEN	ITS QUESTIC	ONNAIRE						
Please answer the following questions, to the best of your knowledge, on behalf of your organization. * 46. Does your association have knowledge about NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A 4 7. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A 6 4 7. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A 6 4 8 4 8. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A 6 7 8 * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A 6 7 * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A 7 * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A 7 * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? 7 9 9 9 9 9 9 9 9 9 9 9 9 9 9 9 9 9 9	VI. General question	s about NGS – ac	Idressed to your c	organization					
 * 46. Does your association have knowledge about NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A 6 1 2 3 4 5 N/A 6 1 2 3 4 5 N/A 6 1 7 8 No ualready have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No Not sure	Please answer the foll	lowing questions, t	o the best of your k	nowledge, on beh	alf of your organization	n.			
Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 NA * 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? 1 2 3 4 5 N/A * 60. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No No No No No No No No No	* 46. Does your a	ssociation have	e knowledge ab	out NGS?					
0 1 2 3 4 5 N/A * 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? * * 4 5 N/A 0 1 2 3 4 5 N/A 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? * * N/A • <t< td=""><td colspan="9">Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure).</td></t<>	Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure).								
 * 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A 6 7 as No No sure 	0	1	2	3	4	5	N/A		
* 47. How well does your organization know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 NA 4 4 8. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 NA 4 5 NA 4 5 NA 4 5 NA 5 NA 5 NA 6 6 7 8 5 NA 6 7 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8 8	\bigcirc	\odot	\odot	\odot	\bigcirc	\odot	\bigcirc		
Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A 5 N/A 5 N/A 6 4 5 N/A 6 6 7 8 5 0 1 2 3 4 5 N/A 6 7 8 7 8 5 0 1 2 3 4 5 N/A 6 7 8 7 8 7 8 7 8 7 8 7 8 7 8 7 8 7 8 7	* 47. How well do	es your organiz	zation know that	t NGS can be ι	ised in cancer dia	gnostics?			
0 1 2 3 4 5 N/A * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No Not sure	Please answer on	a scale from C	to 5 (0 – not at	all; 5 – a lot; N	I/A – not sure).				
 * 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure). 0 1 2 3 4 5 N/A * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No Not sure 	0	1	2	3	4	5	N/A		
* 48. How much experience does your organization have with NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardinn NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No Not sure	\bigcirc	\bigcirc	\bigcirc	-O	\odot	\odot	\bigcirc		
Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No Not sure	* 48. How much e	experience does	s your organizat	ion have with I	NGS?				
0 1 2 3 4 5 N/A * 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A • 0 0 0 0 0 0 0 * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes • No No Not sure	Please answer on	a scale from C	to 5 (0 – not at	all; 5 – a lot; N	I/A – not sure).				
* 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No Not sure	0	1	2	3	4	5	N/A		
* 49. Does your organization have the knowledge to advice and help patients to answer questions regardin NGS? Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A • 5 N/A • 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? • Yes • No • Not sure	\bigcirc	\cup	\bigcirc	\bigcirc	\bigcirc	0	U		
Please answer on a scale from 0 to 5 (0 – not at all; 5 – a lot; N/A – not sure). 0 1 2 3 4 5 N/A • 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? • Yes • No • Not sure	* 49. Does your of NGS?	rganization hav	e the knowledg	e to advice and	d help patients to a	answer questio	ns regarding		
012345N/A••• <td< td=""><td>Please answer on</td><td>a scale from 0</td><td>to 5 (0 – not at</td><td>all; 5 – a lot; N</td><td>I/A – not sure).</td><td></td><td></td></td<>	Please answer on	a scale from 0	to 5 (0 – not at	all; 5 – a lot; N	I/A – not sure).				
 * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No Not sure 	0	1	2	3	4	5	N/A		
 * 50. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients? Yes No Not sure 	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc		
	* 50. Do you al Yes No Not sure	lready have an	y initiatives and	/or material, lik	e brochures, to ex	plain NGS to p	atients?		

* 51. Is your asso	ociation familiar	with biological	therapies?			
Please answer o members).	n a scale from () to 5 (0 – not fa	amiliar at all; 5-v	very well informed	and we educa	te our
0	1	2	3	4	5	N/A
0	0	0	0	\bigcirc	\bigcirc	\bigcirc
* 52. Is your asso	ociation familiar	with immunoth	erapies?			
Please answer o members).	n a scale from () to 5 (0 – not fa	amiliar at all; 5-v	very well informed	and we educa	te our
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 53. Does your a Please answer o	association hav n a scale from (e knowledge ab) to 5 (0 – not a	oout molecular t t all: 5-verv wel	esting or genetic t l informed and we	esting in clinica educate our m	al practice? nembers).
0	1	2	3	4	5	N/A
0	0	0	0	0	0	0
* 54. Is your asso Please answer o	ociation familiar n a scale from (with biomarker) to 5 (0 – not a	s? t all; 5-very wel	l informed and we	educate our m	nembers).
0	1	2	3	4	5	N/A
\odot	\odot	\odot	\odot	\odot	\odot	0
* 55. Has any Yes No, never	patient asked f	or information a	about the above	mentioned therap	bies?	
56. Please share	with us any oth	ner thoughts on	NGS in the cor	ntext of your organ	ization:	
VII. General questio	ons about NGS – a	ddressed to patie	nts			
Please answer the fo	bllowing questions,	to the best of your	knowledge, on beh	alf of patients.		

* 57. How likely already?	is it that patients	have heard ab	out NGS in the	context of cancer	⁻ therapy and tre	eatment
Please answer c	on a scale from () to 5 (0 – not a	t all; 5 – very lił	cely; N/A – not sur	re).	
0	1	2	3	4	5	N/A
0	\bigcirc	0	0	\bigcirc	\bigcirc	0
* 58. How well d	o patients know	that NGS can I	be used in canc	er diagnostics?		
Please answer o	on a scale from () to 5 (0 – not a	t all; 5 – very lił	cely; N/A – not sur	re).	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0
* 59. How well d	o patients under	stand the bene	fit of using NGS	S in cancer diagno	ostics?	
Please answer c	on a scale from () to 5 (0 – not a	t all; 5-they und	erstand very well).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	0	\bigcirc	\bigcirc	\odot	\bigcirc
* 60. How well d method? Please answer c	o you think patie on a scale from (ents know why l) to 5 (0 – not a	Next Generation t all; 5-they knc	n Sequencing is a w very well).	n important dia	gnostic
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0	\bigcirc
* 61. Do patie making? Yes No, it nee * 62. Do patie	ents know that th eds further clarification ents understand	ne genetic cons	titution of the pa	atient can be used	d in cancer thera	apy decision
🕖 No, it nee	eds further clarification	on				

* 63. Do patients of certain drugs (know that the in the term we use	ndividual geneti e for this is phar	c constitution n macogenetics)	nay affect the effi ?	cacy and possib	ble side effects
Please answer of	n a scale from () to 5 (0 – not a	t all; 5 - they kr	ow very well).		
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 64. How likely is	s it that patients	understand the	e indications re	garding the gener	ated data and c	lata security?
Please answer of	n a scale from () to 5 (0 – not a	t all; 5 – very lił	cely; N/A – not su	re).	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
 No. Patier Not sure 66. If your ans This inform Possible re Other (ple 	swer is "yes", planation is used again elevance for family ase specify)	it will strictly be use ease share: wh nst the patient's inte members	ed for diagnostic pu ich ethical cond erest (eg. by emplo	urposes cerns are you thin overs or insurances)	king of?	
VIL Evaluating the Please answer the for * 67. How import Please answer of	educational requir Moving questions fr ant is thorough n a scale from C	ements on NGS for om a patient's pers information abo) to 5 (0 – not a	or patients spective to the bes out the diagnost t all; 5 – very in	t of your knowledge. ic procedure, bef nportant; N/A – no	ore sequencing ot sure).	, for patients?
0	1	2	3	4	5	N/A
0	0	0	0	\bigcirc	0	\bigcirc

* 68. How importation about the proced	ant is it for patie ure?	ents to receive a	a document bef	ore sequencing, v	with detailed exp	planations		
Please answer of	n a scale from () to 5 (0 – not a	t all; 5 – very in	iportant; N/A – no	ot sure).			
0	1	2	3	4	5	N/A		
0	0	0	0	\bigcirc	0	0		
69. If you think it is important, what is the preferred option								
O As part of	the informed conse	ent						
🔵 In a docun	nent separate from	the informed conse	ent, in a printed for	nat				
O In a docun	nent separate from	the informed conse	ent, in a digital form	at				
* 70. How importa data will be used	ant is it for patie for?	ents to receive a	a document that	explains for whi	ch analyses/pro	jects the NGS		
0	1	2	3	4	5	N/A		
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0		
 In a docun In a docun * 72. How importa advantages, but a especially concer Please answer of 	nent separate from nent separate from ant is it for patie also the impact ning the NGS o n a scale from (the informed const the informed const ents to receive of NGS might hav lata produced?	ent, in a printed for ent, in a digital form detailed informa ve on their and t t all: 5 – verv im	^{nat} tion, like a broch heir families' live	ure, that explain s, as well as the ot sure)	s the e risks,		
0	1	2	3	4	5	N/A		
0	0	0	0	\bigcirc	0	0		
73. If you think As part of In a docum	k it is important the informed conse nent separate from nent separate from	, what is the pre- ent the informed conse the informed conse	eferred option ent, in a printed for ent, in a digital form	nat at				

* 74. How importa can withdraw that	nt is it for patie data any time	nts to receive a based on the G	a document on h eneral Data Pro	now data security stection Regulation	/ is guaranteed, a	and how they
Please answer on	a scale from 0) to 5 (0 – not a	t all; 5 – very im	portant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
K. Evaluating the rig	hts of the patient	s to receive the r	esults in an appro	priate format		
* 75. How importa	nt is it for patie	nts to receive a	a report, after the	e NGS analysis,	containing the re	esults?
Please answer on	a scale from 0	to 5 (0 – not a	t all; 5-very imp	ortant).		
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\odot	\bigcirc	\odot	\bigcirc
 * 76. Should pa Yes No Not sure * 77. Should pa returned result current treatmed Yes No Not sure * 78. How importation 	atients have the atients have the s should be? T ent, to all the de nt is it for the p that affect fami	e choice to rece e possibility to o he comprehens etailed results o patients that the ily members?	eive the results? choose between siveness could r f the NGS analy final report con	different options each from no res vsis. tains information	s on the how con sults, to results o about those imp	nprehensive the nly regarding the plications of
Please answer on	a scale from 0) to 5 (0 – not a	t all; 5 – very im	portant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 79. How importa the results?	nt is it for patie	nts to have ger	netic consultatio	n assistance dur	ing and after the	discussion of
Please answer on	a scale from 0) to 5 (0 – not a	t all; 5 – very im	portant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
0	0	0	0	\bigcirc	0	0

* 80. From a report or sep	patient's perspec arated in two repo	tive, should the orts?	e genetic result	s and pharmaco	genetic results be	e part of one
All result	s should be part of on	e report, in a print	ed format			
All result	s should be part of on	e report, in a digit	al format			
They sho	ould be separated in t	wo reports, in a pri	inted format			
They sho	ould be separated in ty	wo reports, in a diç	gital format			
			-			
* 81. Should	the patient have t	the pharmacog	enetic results a	vailable on a m	obile device or in a	a printed form?
Only in a	digital form on a mot	oile device				
Only in a	printed form					
🕥 On a mo	bile device and in a p	rinted form				
82. If provide provide a go	ed in a digital form od level of data se	on a mobile d curity confider	evice, does a s nce to the patie	ecurity level sim nt?	ilar to that used fo	or e-banking
O No						
◯ Not sure						
* 83. How impor	tant is it for the pa	atient who has	access to the r	esults of the ger	netic testing (inclu	ding mutated
genes and diag	nosis)?			Ũ	C (C
Please answer	on a scale from 0	to 5 (0 – not a [;]	t all; 5-very imp	ortant).		
0	1	2	3	4	5	N/A
\odot	\odot	\odot	\odot	\odot	\odot	\bigcirc
* 84. From a	patient's perspec	tive, how limite	ed should the a	ccess to the res	ults from the gene	tic testing be?
O Very limi	ted (ex. only the patie	nt and the cliniciar	n in cancer care sh	ould have access)		
 Mildly lim 	nited (ex. the patient, t	he clinician in can	cer care and other	clinicians the patier	nt goes to should have	e access)
Limited (have acc	ex. the patient, the cliness)	nician in cancer ca	are and other clinic	ans the patient goe	s to, as well as the ph	armacists should
🕖 Not limite	ed (anyone can have a	access)				

* 85. How importa	nt is it for the pa	tient who has a	ccess to pharma	acogenetic resul	ts?	
Please answer on	a scale from 0 t	o 5 (0 – not at a	all; 5-very import	ant).		
0	1	2	3	4	5	N/A
Q	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 86. From a pa	itient's perspecti	ve, how limited	should the acce	ess to the pharm	nacogenetic res	ults be?
Very limited	(ex. only the patien	t and the clinician ir	n cancer care shoul	d have access)		
Mildly limiter	d (ex. the patient, th	e clinician in cance	r care and other cli	nicians the patient g	oes to should have	access)
Limited (ex. have access	the patient, the clini s)	cian in cancer care	and other clinician	s the patient goes to	o, as well as the ph	armacists should
Not limited (anyone can have a	ccess)				
* 87. What level of	evidence would	a patient like to	o have in order	to be treated ba	sed on NGS res	sults?
Select all that appl very important).	y (multiple answ	ers possible) ai	nd provide rank	ng of importanc	e from 0-5 (0 –	not at all; 5-
	0	1	2	3	4	5
The NGS test is approved for therapy decision by regulator authorities	y O	Ō	0	0	0	0
Recommended by th medical society	e O	0	0	0	0	0
Evidence is publishe a scientific journal	d in	0	0	0	0	0
Experience from other patients or patient	er O	0	0	0	advocacy groups	
* 88. How importal participate in (in the diagnostic proceed)	nt is it that the pa e patient's coun cedure?	atients receive i try or neighborii	nformation aboung countries), a	ut existing clinicand that the inform	al trials the patie mation is genera	ent can ated through
Please answer on	a scale from 0 t	o 5 (1 – not at a	all; 5-very import	ant).		
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0	\bigcirc

* 89. How importar the research or any	nt is it for the p y further incide	atient to be info ental finding rel	ormed of incider evant to her/his	ntal findings relev health as well as	ant to the disea her/his genetic	se object of al relatives?
Please answer on a	a scale from 0	to 5 (1 – not a	t all; 5-very imp	ortant).	_	
0	1	2	3	4	5	N/A
0	0	\bigcirc	\odot	\bigcirc	0	\bigcirc
* 90. How importan the results of the te	nt is it for the p est?	atient to have a	a psychological	assistance during	g/after the comn	nunication of
Please answer on a	a scale from 0	to 5 (1 – not a	t all; 5-very imp	ortant).		
0	1	2	3	4	5	N/A
0	0	0	\bigcirc	\bigcirc	0	0

PATIENTS' I	NEEDS QUE	STIONNAIRE	Instand-NG	54P				
CHILDHOOD P	ATIENTS QU	JESTIONNAII	RE					
I. General questions	about NGS – ado	lressed to your or	ganization					
Please answer the fol patients.	lowing questions, t	to the best of your l	knowledge, on beh	alf of your organization	n, in the context of	childhood		
* 91. Do you k	now what Next	Generation Se	quencing (NGS	6) means?				
⊖ Yes								
O No								
Not sure	Not sure							
* 92 Does your a	ssociation have	a knowledge ab	out NGS2					
92. Does your association have knowledge about NGS?								
Please answer on a scale from 0 to 5 (0 - not at all; 5 - a lot; N/A - not sure).								
0	1	2	3	4	5	N/A		
* 93. How well do	* 93. How well does your organization know that NGS can be used in cancer diagnostics?							
Please answer or	a scale from () to 5 (0 - not at	all; 5 - a lot; N/	′A - not sure).				
0	1	2	3	4	5	N/A		
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc		
* 94. How much e	experience doe	s your organiza	tion have with l	NGS?				
Please answer or	a scale from () to 5 (0 - not at	all; 5 - a lot; N	/A - not sure).				
0	1	2	3	4	5	N/A		
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc		
* 95. Is your orga regarding NGS?	nization prepar	ed to advice an	d help patients	and/or their parer	nts to answer q	uestions		
	a scale from (າ ເວ ວ (U - not at 2	all; 5 - a lot; N/	A - not sure).	5	N/A		
U		2	5	4	5	17/74		

* 96. Do you already have any initiatives and/or material, like brochures, to explain NGS to patients and/or their parents?

Ves No Not sure

* 97. Has any patient and/or parent asked for information about NGS, pharmacogenetics, or personalized medicine?

Ves No, never

98. Please share with us any other thoughts on NGS in the context of your organization:

II. General questions about NGS - addressed to patients and parents

Please answer the following questions, to the best of your knowledge, on behalf of patients and/or parents, in the context of childhood patients.

* 99. Do patients and/or parents know what NGS means?

Ves No Not sure

* 100. How likely is it that patients and/or parents have heard about NGS in the context of childhood cancer therapy and treatment already?

Please answer on a scale from 0 to 5 (0 – not at all; 5 – very likely; N/A – not sure) 0 2 3 5 N/A 1 4 * 101. How well do patients and/or parents know that NGS can be used in cancer diagnostics? Please answer on a scale from 0 to 5 (0 - not at all; 5 - very well; N/A - not sure). 0 2 3 4 5 N/A 1

* 102. How well	do patients and/	or parents unde	erstand the bene	efit of using NGS	in cancer diagr	nostics?
Please answer o	n a scale from 0) to 5 (0 – not at	t all; 5 – very we	ell; N/A – not sure	e).	
0	1	2	3	4	5	N/A
\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 103. How well of Please answer o	do you think pat n a scale from 0	ients and/or par) to 5 (0 – not al	ents know why t all; 5 – very we	NGS is an impor ell; N/A – not sure	tant diagnostic	method?
0	1	2	3	4	5	N/A
Q	\bigcirc	\bigcirc	\bigcirc	0	\bigcirc	\bigcirc
* 104. Do pat therapy decis	ients and/or par ion making?	ents know that t	he genetic cons	stitution of the pa	tient can enhan	ce cancer

O Yes

No, it needs further clarification

Not sure

* 105. Do patients and/or parents understand the benefit of selecting a therapy based on the genetic constitution of the patient?

Yes

) No, it needs further clarification

) Not sure

* 106. Do patients and/or parents know that the individual genetic constitution may affect the efficacy and possible side (and late) effects of certain drugs (also known as pharmacogenetics)?

O Yes

No, it needs further clarification

Not sure

* 107. How likely is it that patients and/or parents understand the consequences regarding the information they get about their genome and how to handle this?

Please answer on a scale from 0 to 5 (0 - not at all; 5 - very well; N/A - not sure).

0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc

* 108. How likely i and data security?	s it that patien	ts and/or parents	s understand the	consequences	regarding the g	enerated data
Please answer on	a scale from (0 to 5 (0 – not at	all; 5 – very well;	N/A – not sure	·).	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 109. Do you f NGS? Yes No. Patient Not sure	think patients a	and/or parents w it will strictly be used	ill have any ethic	al concerns reg oses.	arding the data	produced by
110. If your an This inform Possible rei	swer is "yes", ation is used agai levance for family se specify)	blease share: wh nst the patient's inte members	ich ethical conce	rns are you thins or insurances)	nking of?	
* 111. What level based on NGS res	of evidence we sults?	ould patients and	l/or parents like t	o have in order	to agree on a tr	reatment
very important; N/	A – not sure).	swers possible)				- 110t at all, 5-
	0	1	2	3	4	5
NGS is approved for therapeutic decision making by regulator authorities	y O	0	\bigcirc	0	0	0
Recommended by the medical society	he O	0	0	0	0	0
Evidence is publishe a scientific journal	ed in	0	0	0	0	0
Experience from oth patients or patient	er	Ő	0	\odot	advocacy groups	

III. Evaluating the educational requirements on NGS for patients and/or parents

Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patients.

* 112. How impor and/or parents?	tant is thorough	n information ab	out the diagnos	stic procedure, be	fore sequencin	g, for patients
Please answer or	n a scale from () to 5 (0 – not at	t all; 5 – very in	nportant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 113. How impor explanations abo	tant is it for pat ut the procedur	ients and/or par e?	ents to receive	a document befo	re sequencing,	with detailed
Please answer or	n a scale from () to 5 (0 – not at	t all; 5 – very in	portant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
As part of t As part of t In a docum In a docum * 115. How impor analyses/projects Please answer or	the informed conse nent separate from tent separate from tant is it for pat the NGS data n a scale from (the informed conse the informed conse ients and/or par will be used for) to 5 (0 – not at	ent, in a printed for ent, in a digital form rents to receive ? t all; 5 – very in	^{mat} a document that nportant; N/A – nc	explains for wh t sure).	ich
0	1	2	3	4	5	N/A
\odot	\bigcirc	\bigcirc	\odot	\odot	\odot	\bigcirc
 116. If you thin As part of the second s	hk it is importan the informed conse nent separate from tent separate from tant is it for pat ntages, but als oncerning the I n a scale from (1	it, what is the present the informed conse the informed conse ients and/or par o the impact NC NGS data produ 0 to 5 (0 – not at 2	referred option ent, in a printed for ent, in a digital form rents to receive GS might have uced? t all; 5 – very in 3	mat hat detailed informat on their and their hportant; N/A – no 4	ion, like a broch families' lives, a ot sure). 5	nure, that as well as the N/A

 118. If you think it is important, what is the preferred option As part of the informed consent In a document separate from the informed consent, in a printed format In a document separate from the informed consent, in a digital format * 119. How important is it for patients and/or parents to receive a document on how data security is guaranteed, and how they can withdraw that data any time based on the General Data Protection Regul Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A M. Evaluating the rights of the patients and parents to receive the results in an appropriate format Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patients of the patients from a patient's perspective to the best of your knowledge, in the context of childhood patients and parents to receive the results of your knowledge, in the context of childhood patients and parents to receive the results of your knowledge, in the context of childhood patients and parents to receive the patient of your knowledge, in the context of childhood patients and parents to receive the patient of your knowledge, in the context of childhood patients and parents to receive the patient of your knowledge, in the context of childhood patients and parents to receive the patient of your knowledge, in the context of childhood patients and parents to receive the patients of your knowledge, in the context of childhood patients and parents to receive the patients of your knowledge, in the context of childhood patients and parents to receive the patients of your knowledge, in the context of childhood patients of the patients and parents to receive the patients and parents to receive the patients and parents to patients and parents to patients and patients and patients and patients patients and patie	ation?
 As part of the informed consent In a document separate from the informed consent, in a printed format In a document separate from the informed consent, in a digital format * 119. How important is it for patients and/or parents to receive a document on how data security is guaranteed, and how they can withdraw that data any time based on the General Data Protection Regul Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A M. Evaluating the rights of the patients and parents to receive the results in an appropriate format Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patients of the patients and parents to receive the results in an appropriate format 	ation?
 In a document separate from the informed consent, in a printed format In a document separate from the informed consent, in a digital format * 119. How important is it for patients and/or parents to receive a document on how data security is guaranteed, and how they can withdraw that data any time based on the General Data Protection Regulated and how they can withdraw that data any time based on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed on the General Data Protection Regulated and how they can be accessed and how they can be	ation?
 In a document separate from the informed consent, in a digital format * 119. How important is it for patients and/or parents to receive a document on how data security is guaranteed, and how they can withdraw that data any time based on the General Data Protection Regult Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A M. Evaluating the rights of the patients and parents to receive the results in an appropriate format Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patients of the patients and parents to receive the results of your knowledge, in the context of childhood patients of the patients and parents to receive the result of your knowledge, in the context of childhood patients of the patient's perspective to the best of your knowledge, in the context of childhood patients of the patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the patient's perspecti	ation?
 * 119. How important is it for patients and/or parents to receive a document on how data security is guaranteed, and how they can withdraw that data any time based on the General Data Protection Regulated in the general Data Protecti	ation?
 * 119. How important is it for patients and/or parents to receive a document on how data security is guaranteed, and how they can withdraw that data any time based on the General Data Protection Regul Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A M. Evaluating the rights of the patients and parents to receive the results in an appropriate format Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patients of the patients and parents to receive the results of your knowledge, in the context of childhood patients of the patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the best of your knowledge, in the context of childhood patient's perspective to the patient's perspective to	ation?
guaranteed, and how they can withdraw that data any time based on the General Data Protection Regul Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A O 1 2 3 4 5 N/A O 1 2 3 4 5 N/A O 0 0 0 0 0 0 0 M. Evaluating the rights of the patients and parents to receive the results in an appropriate format Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patients	ation?
0 1 2 3 4 5 N/4 0 <t< td=""><td></td></t<>	
0 1 2 3 4 5 N/4 0 <td></td>	
M. Evaluating the rights of the patients and parents to receive the results in an appropriate format Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patients	
N. Evaluating the rights of the patients and parents to receive the results in an appropriate format Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patient.	
N . Evaluating the rights of the patients and parents to receive the results in an appropriate format Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood patient	
Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood pati	
Please answer the following questions from a patient's perspective to the best of your knowledge, in the context of childhood pat	
	ents.
* 120. How important is it for patients and/or parents to receive a report, after the NGS analysis, containi	ng the
results?	
Please answer on a scale from 0 to 5 (0 – not at all: 5 – very important: N/A – not sure)	
$0 \qquad 1 \qquad 2 \qquad 3 \qquad 4 \qquad 5 \qquad N/4$	
* 121 Should patients and/or parents have the choice to receive the results?	
O No	
O Not sure	
No Not sure	
 No Not sure * 122 Should patients and/or parents have the possibility to choose between different options on the 	how
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, 	how to
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, results only regarding the current treatment, to all the detailed results of the NGS analysis. 	how to
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, results only regarding the current treatment, to all the detailed results of the NGS analysis. 	how to
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No 	how to
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure 	how to
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure 	how to
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure 	how to
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure 	how to
 No Not sure * 122. Should patients and/or parents have the possibility to choose between different options on the comprehensive the returned results should be? The comprehensiveness could reach from no results, results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure 	how to

implications of the	NGS analysis	that affect fami	parents that the parents the parent	e final report cor	itains informatio	n about those
Please answer on	a scale from (0 to 5 (0 – not a	t all; 5 – very im	portant; N/A – n	ot sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 124. How import the discussion of t	ant is it for pat he results?	ients and/or pai	rents to have ge	enetic consultatio	on assistance du	ring and after
Please answer on	a scale from (0 to 5 (0 – not a	t all; 5 – very im	iportant; N/A – n	ot sure).	
0	1	2	3	4	5	N/A
\Box	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc	\bigcirc
 On a mobile * 126. If provide good le Yes No Not sure V. Evaluating the right with a well-informed * 127. Should for they turn 18? Yes No Not sure 	e device and in a ed in a digital f evel of data se nts of a former cl physician, and th former childhoo	printed form form on a mobile curity confidenc hildhood cancer p neir right to recons	e device, does a le to the patient atient to be inform sider nts be automatio	a security level s and/or parents? ned about an earlie cally informed th	imilar to that use	ed for e-banking second meeting

128. If yes, how should they be informed?
Per email
Per post
Face to face
Phone call
Other (please specify)

* 129. Should this notification include the offer for an appointment with a well-informed physician, geneticist, and psychologist?

Ves No Not sure

* 130. Should former childhood cancer patients also receive detailed information to have the option of withdrawing their data based on the General Data Protection Regulation, in case they wish to do so?

Yes
 No
 Not sure

* 131. Should the patient have the pharmacogenetic results available on a mobile device or in a printed form?

Only in a digital form on a mobile device

Only in a printed form

On a mobile device and in a printed form

* 132. If provided in a digital form on a mobile device, does a security level similar to that used for e-banking provide a good level of data security confidence to the patient?

Ves No Not sure

PATIENTS' ADULT PATIEN General guestio	NEEDS QUE	STIONNAIRE ONNAIRE – addressed to	Instand-NGS	34P ation.		
Please answer t in the context of	he following q [:] adult patients	uestions, to the	e best of your	knowledge, on I	behalf of your	organization
V. General question	ns about NGS – a	ddressed to your o	organization			
Please answer the fo	llowing questions,	to the best of your k	nowledge, on beha	alf of your organizatio	n, in the context of	adult patients.
* 133. Does your	association ha	ve knowledge a	bout NGS?			
Please answer or	n a scale from () to 5 (0 – not at	all; 5 – a lot; N	I/A – not sure).		
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 134. How well o Please answer or	loes your orgar n a scale from (nization know the	at NGS can be : all; 5 – a lot; N	used in cancer d I/A – not sure).	iagnostics?	
0	1	2	3	4	5	N/A
0	\bigcirc	\odot	\odot	\bigcirc	\bigcirc	\bigcirc
* 135. How much	experience do	es your organiz	ation have with	NGS?		
Please answer or	n a scale from () to 5 (0 – not at	: all; 5 – a lot; N	I/A – not sure).		
0	1	2	3	4	5	N/A
	U	0	0	0	0	0
* 136. Does your NGS?	organization ha	ave the knowled	ge to advice ar	nd help patients to	o answer quest	ions regarding
Please answer or	n a scale from () to 5 (0 – not at	all; 5 – a lot; N	I/A – not sure).		
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	0	\odot	\odot	\bigcirc	\bigcirc

* 137. Do you	already have a	any initiatives an	d/or material, lik	ke brochures, to	explain NGS to	patients?
O Yes						
🚫 No						
Not sure						
* 138. Is your as	sociation familia	ar with biological	therapies?			
Please answer o members).	n a scale from	0 to 5 (0 – not fa	ımiliar at all; 5-v	ery well informe	d and we educa	te our
0	1	2	3	4	5	N/A
Q	\bigcirc	0	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 139. Is your as	sociation familia	ar with immunoth	nerapies?	erv well informe	d and we educa	te our
members).		0 10 5 (0 – 1101 12	inniai at an, 5-v	ery wen morned	u anu we euuca	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 140. Does your Please answer o	association ha n a scale from 1	ive knowledge a 0 to 5 (0 – not a 2	bout molecular t all; 5-very well 3	informed and w	c testing in clini e educate our m 5	nembers).
		0				
* 141. Is your as	sociation familia	ar with biomarke	rs?	informed and w	e educate our m	vembers)
		2 10 5 (0 – 1101 a	all, 5-very well		5	N/A
Õ	Ö	0	0	- -	0	
* 142. Has an Yes No, never	y patient asked	I for information	about the above	e mentioned ther	anization:	

VII. General questions	about NGS – add	ressed to patien	ts			
Please answer the follo	wing questions, to t	he best of your k	nowledge, on beha	alf of your organisation	on in the context of a	adult patients.
* 144. How likely is already?	it that patients	have heard at	oout NGS in the	e context of canc	er therapy and t	reatment
Please answer on	a scale from 0 to	o 5 (0 – not at	all; 5 – very lik	ely; N/A – not su	re).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot	0	0
* 145. How well do	patients know t	hat NGS can	be used in can	cer diagnostics?		
Please answer on	a scale from 0 to	o 5 (0 – not at	all; 5 – very lik	ely; N/A – not su	re).	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0	0
* 146. How well do	patients unders	stand the bene	fit of using NG	S in cancer diag	nostics?	
Please answer on	a scale from 0 to	o 5 (0 – not at	all; 5-they und	erstand very wel	I).	
0	1	2	3	4	5	N/A
	U	\bigcirc	0	\bigcirc	0	U
* 147. How well do method?	you think patie	nts know why	Next Generatio	on Sequencing is	an important di	agnostic
Please answer on	a scale from 0 to	o 5 (0 – not at	all; 5-they know	w very well).		
0	1	2	3	4	5	N/A
\Box	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0
 * 148. Do patier making? Yes No, it needs * 149. Do patier Yes Yes No, it needs 	nts know that the further clarification nts understand t	e genetic cons	stitution of the p	patient can be us	ed in cancer the	ion of the patient?

* 150. Do patients know that the individual genetic constitution may affect the efficacy and possible side effects of certain drugs (the term we use for this is pharmacogenetics)?						
Please answer or	n a scale from	0 to 5 (0 – not at	all; 5 - they kr	ow very well).		
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
* 151. How likely	is it that patien	ts understand th	e indications r	egarding the gen	erated data and	data security?
Please answer or	n a scale from	0 to 5 (0 – not at	all; 5 – very lil	ely; N/A – not su	ıre).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
No. Patient Not sure Not sure 153. If your an This inform Possible re Other (plea UIL Evaluating the of Please answer the for * 154. How impor	ts understand that nswer is "yes", nation is used agai elevance for family ase specify) educational requi ollowing questions tant is thoroug	it will strictly be use please share: wi nst the patient's inte members rements on NGS fo from a patient's pers h information ab	nich ethical cor erest (eg. by emplo or patients spective to the bes out the diagno	rrposes ncerns are you th nyers or insurances) t of your knowledge stic procedure, b	inking of? in the context of adu efore sequencing	It patients. g, for patients?
Please answer or	n a scale from	0 to 5 (0 – not al	all; 5 – very in	nportant; N/A – n	ot sure).	
0	1	2	3	4	5	N/A
U	U	0	0	0	0	\cup

* 155. How impor about the procedu	* 155. How important is it for patients to receive a document before sequencing, with detailed explanations about the procedure?				xplanations	
Please answer or	a scale from (0 to 5 (0 – not a	t all; 5 – very im	iportant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0
156. If you thir	ık it is importar	nt, what is the pr	eferred option			
As part of t	he informed conse	ent				
O In a docum	ent separate from	the informed conse	ent, in a printed form	nat		
In a docum	ent separate from	the informed conse	ent, in a digital form	at		
* 157. How impor data will be used	tant is it for pat for?	ients to receive	a document tha	at explains for wh	ich analyses/pr	ojects the NGS
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc	\bigcirc
158. If you thir As part of t In a docum	nk it is importar the informed conse tent separate from tent separate from	nt, what is the pr ent the informed conse the informed conse	referred option ent, in a printed forr ent, in a digital form	nat at		
* 159. How impor advantages, but a especially concer	tant is it for pa also the impact ning the NGS o	tients to receive NGS might hav data produced?	detailed inform e on their and	nation, like a broc their families' live	hure, that expla s, as well as th	ains the e risks,
Please answer or	a scale from (0 to 5 (0 – not a	t all; 5 – very im	iportant; N/A – no	ot sure).	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\odot	\bigcirc	\odot	0
* 160. How important is it for patients to receive a document on how data security is guaranteed, and how they can withdraw that data any time based on the General Data Protection Regulation? Please answer on a scale from 0 to 5 (0 – not at all: 5 – very important: N/A – not sure)						
0	1	2	3	4	5	N/A
0	0	0	0	0	0	0
K. Evaluating the rig	ghts of the patien	ts to receive the r	esults in an appro	priate format		

Please answer on a scale from 0 to 5 (0 – not at all; 5-very important). 0 1 2 3 4 5 NA * 162. Should patients have the choice to receive the results? Yes No No Not sure * 163. Should patients have the possibility to choose between different options on the how comprehensive the returned results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No No No in sure * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic results and pharmacogenetic results be part of one report or separated in two reports; in a printed format N/A 5 N/A	* 161. How impor	* 161. How important is it for patients to receive a report, after the NGS analysis, containing the results?					results?
0 1 2 3 4 5 NA * 162. Should patients have the choice to receive the results? Yea No No No sure * 163. Should patients have the possibility to choose between different options on the how comprehensive the results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. Yea No No No sure * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 NA * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 9 3 4 6 5 NA * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a pinted format 10 1 11 1 12 3 13 1 14 1 15 1 16 1 16 1 16 1 16 1 16 1 16 1 <tr< td=""><td>Please answer or</td><td>n a scale from (</td><td>) to 5 (0 – not a</td><td>t all; 5-very imp</td><td>ortant).</td><td></td><td></td></tr<>	Please answer or	n a scale from () to 5 (0 – not a	t all; 5-very imp	ortant).		
 + 162. Should patients have the choice to receive the results? Yes No + 163. Should patients have the possibility to choose between different options on the how comprehensive the returned results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure + 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 NA • 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 NA • 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a printed format All results abould be part of one report, in a printed format All results abould be part of one report, in a printed format They should be separated in two reports, in a printed format They should be separated in two reports, in a digital format 	0	1	2	3	4	5	N/A
 162. Should patients have the choice to receive the results? Yes No Not sure * 163. Should patients have the possibility to choose between different options on the how comprehensive the returned results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. No No sure * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 NA * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 1 2 3 4 5 N/A 6 7 8 8 9 1 2 3 4 5 N/A 6 6 7 8 9 9 1 1<td>\bigcirc</td><td>\bigcirc</td><td>\bigcirc</td><td>\bigcirc</td><td>\bigcirc</td><td>\bigcirc</td><td>\bigcirc</td>	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
 * 162. Should patients have the choice to receive the results? Yes No Not sure * 163. Should patients have the possibility to choose between different options on the how comprehensive the returned results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report, or separated in two reports; All results should be part of one report, in a printed format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format 							
Nes No sure * 163. Should patients have the possibility to choose between different options on the how comprehensive the returned results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No No sure * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A • 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A • 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report, in a printed format All results should be part of one report, in a printed format All results should be separated in two reports, in a printed format They should be separated in two reports, in a digital format	* 162. Should	patients have t	he choice to red	ceive the result	s?		
 No Not sure * 163. Should patients have the possibility to choose between different options on the how comprehensive the returned results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 NA * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 NA * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 NA * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report, in a pinted format All results should be part of one report, in a digital format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format 	O Yes						
Not sure * 163. Should patients have the possibility to choose between different options on the how comprehensive the returned results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No No Not sure * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? It results should be part of one report, in a printed format It results should be part of one report, in a digital format . All results should be parated in two reports, in a digital format They should be se	O No						
 * 163. Should patients have the possibility to choose between different options on the how comprehensive the returned results should be? The comprehensiveness could reach from no results, to results only regarding the current treatment, to all the detailed results of the NGS analysis. Yes No Not sure * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report. All results should be part of one report, in a printed format All results should be part of one report, in a printed format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format 	Not sure						
 * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a printed format All results should be part of one report, in a printed format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format 	* 163. Should returned resul current treatm Yes No No Not sure	patients have t ts should be? T ent, to all the de	he possibility to The comprehen etailed results c	o choose betwe siveness could f the NGS anal	en different optio reach from no re ysis.	ns on the how o sults, to results	comprehensive the only regarding the
 * 164. How important is it for the patients that the final report contains information about those implications of the NGS analysis that affect family members? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a grinted format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format 							
Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a digital format All results should be separated in two reports, in a digital format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format	* 164. How impor the NGS analysis	tant is it for the that affect fam	patients that th ily members?	e final report c	ontains informatio	n about those ir	nplications of
0 1 2 3 4 5 N/A * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? • <t< td=""><td>Please answer or</td><td>n a scale from (</td><td>) to 5 (0 – not a</td><td>t all; 5 – very in</td><td>nportant; N/A – no</td><td>ot sure).</td><td></td></t<>	Please answer or	n a scale from () to 5 (0 – not a	t all; 5 – very in	nportant; N/A – no	ot sure).	
 * 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a printed format All results should be part of one report, in a digital format They should be separated in two reports, in a digital format 	0	1	2	3	4	5	N/A
* 165. How important is it for patients to have genetic consultation assistance during and after the discussion of the results? Please answer on a scale from 0 to 5 (0 – not at all; 5 – very important; N/A – not sure). 0 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a printed format All results should be part of one report, in a printed format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	\odot	0
0 1 2 3 4 5 N/A * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? • All results should be part of one report, in a printed format •	* 165. How impor of the results? Please answer or	tant is it for pat n a scale from (ients to have ge) to 5 (0 – not a	enetic consulta t all; 5 – very ir	ion assistance du nportant; N/A – no	iring and after tl ot sure).	ne discussion
 * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a printed format All results should be separated in two reports, in a printed format They should be separated in two reports, in a digital format They should be separated in two reports, in a digital format 	0	1	2	3	4	5	N/A
 * 166. From a patient's perspective, should the genetic results and pharmacogenetic results be part of one report or separated in two reports? All results should be part of one report, in a printed format All results should be part of one report, in a digital format They should be separated in two reports, in a printed format They should be separated in two reports, in a digital format 	0	0	0	0	0	0	\bigcirc
	* 166. From a report or sepa All results All results They shou	patient's persp rated in two rep should be part of o should be part of o ld be separated in ld be separated in	ective, should t ports? ne report, in a print ne report, in a digit two reports, in a pr two reports, in a di	he genetic resu ted format al format inted format gital format	llts and pharmaco	genetic results	be part of one

* 167. Should	the patient have	e the pharmaco	ogenetic results	available on a mo	obile device or ir	n a printed form?
Only in a c	ligital form on a mo	bile device				
Only in a p	rinted form					
On a mobi	le device and in a p	printed form				
168. If provide provide a goo	d in a digital for level of data s	m on a mobile ecurity confide	device, does a nce to the patier	security level sim nt?	ilar to that used	for e-banking
O Yes						
O No						
O Not sure						
* 169. How impor mutated genes a	tant is it for the nd diagnosis)?	patient who ha	s access to the	results of the ger	netic testing (inc	luding
Please answer or	n a scale from 0	to 5 (0 – not a	t all; 5-very imp	ortant).		
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
 170. From a Very limite Mildly limit Limited (e) have acce Not limited 	patient's perspend d (ex. only the patient ed (ex. the patient, the patient, the cl ss) (anyone can have	ective, how limi ent and the clinicia the clinician in car inician in cancer ca access)	Ited should the a n in cancer care sh ncer care and other are and other clinici	access to the rest ould have access) clinicians the patient ans the patient goes	uits from the ger goes to should have to, as well as the pf	etic testing be? e access) harmacists should
* 171. How impor	tant is it for the	patient who ha	is access to the	pharmacogenetic	c results?	
0	1	2	3	4	5	N/A
0	Ó	0	0	0	0	0
* 172. From a Very limite Mildly limit	patient's perspend d (ex. only the patiend ed (ex. the patient, c. the patient, the cl ss)	ective, how limi ent and the clinicia the clinician in car inician in cancer c	ited should the a n in cancer care sh ncer care and other are and other clinici	access to the pha ould have access) clinicians the patient ans the patient goes	rmacogenetic re goes to should have to, as well as the ph	esults? e access) earmacists should
Limited (ex have acce	a the patient, the cl ss) (anyone can have	inician in cancer c access)	are and other clinici	ans the patient goes	to, as well as the ph	armacists should

* 173. What level of evidence would a patient like to have in order to be treated based on NGS results?

Select all that apply (multiple answers possible) and provide ranking of importance from 0-5 (0 – not at all; 5-very important).

	0	1	2	3	4	5
The NGS test is approved for therapy decision by regulatory authorities	0	0	0	0	0	0
Recommended by the medical society	\odot	0	\bigcirc	0	\bigcirc	\odot
Evidence is published in a scientific journal	0	0	\bigcirc	0	0	0
Experience from other patients or patient	0	0	\odot	Q	dvocacy groups	

* 174. How important is it that the patients receive information about existing clinical trials the patient can participate in (in the patient's country or neighboring countries), and that the information is generated through the diagnostic procedure?

Please answer on a scale from 0 to 5 (1 - not at all; 5-very important).

0	1	2	3	4	5	N/A
\odot						

* 175. How important is it for the patient to be informed of incidental findings relevant to the disease object of the research or any further incidental finding relevant to her/his health as well as her/his genetical relatives?

Please answer on a scale from 0 to 5 (1 – not at all; 5-very important).

0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\odot	\odot	\odot	\bigcirc

* 176. How important is it for the patient to have a psychological assistance during/after the communication of the results of the test?

Please answer on a scale from 0 to 5 (1 – not at all; 5-very important).

0	1	2	3	4	5	N/A
0	\bigcirc	\odot	\odot	\odot	\odot	0

PATIENTS' NEEDS QUESTIONNAIRE Instand-NGS4P

Last question

177. Would you like to share with us any additional information related to any section of the questionnaire? If yes please use the free field to do so.

178. Is the provided information confidential?

O Yes

SOLUTION PROVIDERS Questionnaire

PROJECT INTRODUCTION

INSTAND-NGS4P is an EU-funded Pre-Commercial Procurement (PCP) project for improving cancer patient's benefit from Next Generation Sequencing (NGS). Driven by patient and clinical needs, two innovative NGS workflows from sample pre-analytics to medical decision-making will be developed for routine diagnostics of common and rare adult and paediatric cancers complying with the IVDR. The developed workflows will compile information from cancer gene testing, pharmacogenetics testing and e-medication in proper presentation to medical doctors for supporting therapy decision-making at bedside widely applicable in health systems.

The EU-co-funded PCP project provides funding for a public consortium to define unmet medical and technical needs based on an Open Market Consultation, which lays the foundation for a call for tenders addressing solution providers (companies) to develop their products to better meet user needs. At three cut-off periods, companies responding to this call will be evaluated regarding their ability to answer these users' needs from design perspective until the product phase. The total funding allocated to companies for product development (in total 8.55 M€) will finally lead to two integrated standardized NGS workflows, including decision support.

The different lots of this project cover the entire workflow for integration and standardization of targeted and whole genome DNA sequencing and decision making at the bedside:

lot 1: Pre-sequencing (specimen collection, nucleic acid isolation, library preparation)

lot 2: Sequencing

lot 3: Bioinformatics analysis

lot 4: Integrated reporting

The procurement will take the form of a pre-commercial procurement (PCP) under which R&D service contracts will be awarded to a number of solution providers in parallel in a phased approach. This will make it possible to compare competing alternative solutions.

Each selected operator will be awarded a framework agreement that covers 3 R&D phases.

The 3 phases are:

Design of lots

Prototype of lots

Fully integrated NGS workflow.

Each of the 3 phases will address all 4 lots.

SOLUTION PROVIDERS Questionnaire

PURPOSE OF THE QUESTIONNAIRE

This questionnaire is part of the Open Market Consultation (OMC) which aims at refining the clinical, patient and technical needs defined by the project consortium, as well as the emerging solutions available or under development to address these needs. For this purpose, three questionnaires were developed for three different stakeholder groups – users, solution providers and patient associations. The feedback collected from the questionnaires will support the subsequent preparation of the call for tenders. The questionnaires also allow disclosure of confidential information to the project consortium, which will be treated with high confidentiality. Aggregated data from the results of the survey will potentially be published in an anonymous way, excluding the information labelled as confidential.

The personal information collected through the questionnaires will be exclusively used for the purposes of the project, will not be shared with third parties, and will be deleted after the closure of the project.

GUIDANCE

The questionnaire is addressed to all solution providers in the field of NGS, and is divided into the following sections: General questions, Lot 1, Lot 2, Lot 3, Lot 4. After answering the general questions, you will be asked for each section whether you want to fill it in or not. We would highly appreciate if you would take the time to fill in the section(s) related to your expertise. However, if you do not find the question relevant, feel free to select N/A (not applicable) as an answer or skip the question and to move on to the next one..

Please submit the questionnaire by May 31st, 2021. Thank you in advance for contributing.

* 1. Organisation profile	e
Organisation name	
Address	
City/Town	
Country	
* 2. Contact person	
First name	
Family name	
Email Address	
Phone Number	


General questions

* 3. How is your organization positioned in the NGS scene (more than one answer possible):

P	Provider
N	<i>N</i> anufacturer
lr	ncludes Research & Development
H	lospital/medical centre
A	Academic hospital
	Diagnostic laboratory
F	Patient organization
C	Other (please specify)

* 4. What is your current position in your organization?

* 5. Are you currently performing production/diagnostic activities for products used or diagnostics in the NGS workflow in (please specify the percentage in the textbox):

Europe	
North America	
Asia	
South America	
Australia	
N/A	

urope		
orth America		
Asia		
South America		
Nustralia		
J/A		
* 7. Are you u	ising NGS sequencing for research or for diagnostics?	
O Yes		
O No		
N/A		
* 0 5 1		
" δ. Do you ha	ave access to sequencing facilities?	
* 9. Do you pe	erform sequencing in house or externally?	
O In house		
Externally		
N/A		
* 10. What pla	atform do you use?	
MGI		
O Pac Bio		
N/A		
Other (ple	ease specify)	
Other (pie		

	* 11. How many	samples are you	typically sequer	ncing per week?			
	O Less than 1						
	1-10						
	0 10-50						
	50-100						
	More than 100)					
	○ N/A						
* 12	2. How interested	l are you in whol	e exome sequer	icing? (0-not inte	erested in all, 5 ·	- very interested	d)
	0	1	2	3	4	5	N/A
	\bigcirc	0	0	\bigcirc	\bigcirc	0	\bigcirc
+ 40					-		
13	3. How interested	are you in gene	e panels? (0-not i	interested in all,	5 - very interest	ed)	N1/A
	U	1	2	3	4	5	N/A
	U	U	0	\cup	0	0	0
* 1/	1 How interested	t are you in whol	e denome seque	ancing? (0-not in	terested in all F	. verv interest	ed)
'-	0		2	3		5	N/A
	\sim	\bigcirc	\sim	\cup	\cup		\cup

Introduction lot 1

* 15. Would you like to answer questions related to Lot 1 (Pre-Analytics and Library preparation)?

⊖ Yes

O No

ot 1 page 1/2		_	_	_	_	
CHALLENGE 1:						
moroving the analy	tical porformanco by s	tandardizing and	or simplifying the	nro analytical n	20205505	
	lical performance by s	stanuaruizing anu	or simplifying the	e pre-analytical pi	ocesses	
16. Most comr	nonly used/produce	ed samples for	NGS analysis			
Which are the	types of specimen:	s most included	l in the test pro	cedure kit prod	uced or is prod	uced as a kit by
your organizat	ion?			·	·	,
FFPE						
Frozen tiss	sues					
Blood						
FNAs						
Plasma						
Other						
Other (plea	ase specify)					
17. Is the infor	mation above confi	idential?				
⊖ Yes, it is co	onfidential					
◯ No, it can b	be shared					
Among the following,	please indicate the spe	cimen(s) you consi	ider most challengi	ng in terms of defir	ning detailed pre-a	nalytical
processing standards.	. Please weight each of	them on the basis	of their importance	from 1 (lower) to 5	5 (higher).	
18 FEDE						
	1	2	3	Δ	5	N/A
DNA		2	Ű	-	3	N/A
RNA			0	0	0	0
	\cup	\cup			\cup	
19 Frozen tissue	s					
		2	3	4	5	N/A
	1					
DNA	1	-				

٦

Γ

20. Blood/plasma						
	1	2	3	4	5	N/A
DNA						\bigcirc
RNA	\bigcirc	\odot	\odot	\bigcirc	\odot	\bigcirc
cfDNA						\odot
cfRNA	\odot	\bigcirc	\odot	0	0	0
21. Fine Needle Aspirates						
	1	2	3	4	5	N/A
DNA						0
RNA	0	0	0	0	0	0
22. Circulating Tumor Cells	3					
	1	2	3	4	5	N/A
DNA						\odot
RNA	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
23. Extracellular vesicles						
	1	2	3	4	5	N/A
DNA						0
RNA	0	0	0	0	0	0
24. Saliva						
	1	2	3	4	5	N/A
DNA						\odot
25. Other						
	1	2	3	4	5	N/A
DNA						\bigcirc
RNA	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	\odot
26. If you selected other, p	lease spec	cify				

Yes, it is confidential

No, it can be shared

PROBLEM: Preanalytical processing procedures can cause variations in the test result.

SOLUTION: Standardisation according to ISO and CEN documents

Is your organization aware of the following standards? Please select the first box if there is awareness and the second if your organization works according to or applies the standard:

28. ISO 15189 Medical Laboratories - Requirements for quality and competence

O Aware

Applies

29. ISO 20166-1:2018 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for formalin-fixed and paraffin-embedded (FFPE) tissue - Part 1: isolated RNA

Aware

30. ISO 20166-3:2018 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for formalin-fixed and paraffin-embedded (FFPE) tissue - Part 3: isolated DNA

Aware Applies

31. ISO 20184-1:2018 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for frozen tissue - Part 1: isolated RNA

Aware

32. ISO 20186-1:2019 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for venous whole blood - Part 1: isolated cellular RNA

Aware Applies

33. ISO 20186-2:2019 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for venous whole blood - Part 2: isolated genomic DNA

Aware

Applies

34. ISO 20186-3:2019 Molecular in vitro diagnostic examinations - Specifications for pre-examination processes for venous whole blood - Part 3: isolated circulating cell free DNA from plasma

Aware

35. CEN/TS 17390-1:2020 (WI=00140123) Molecular in vitro diagnostic examinations - Specifications for preexamination processes for circulating tumor cells (CTCs) in venous whole blood - Part 1: Isolated RNA

Aware Applies

36. CEN/TS 17390-2:2020 (WI=00140125) Molecular in vitro diagnostic examinations - Specifications for preexamination processes for circulating tumor cells (CTCs) in venous whole blood - Part 2: Isolated DNA

Aware

37. Do you adhere to other ISO standards for NGS pre-analytics or library preparation? If yes, please specify

38. Is the information above confidential?

Yes, it is confidential

No, it can be shared

39. Does your organization see ISO standardization as a good and useful approach?

O Yes

O No

Comment

40. Is the information above confidential?

Yes, it is confidential

) No, it can be shared

PROBLEM Many quality issues in pre-analytics arise even before nucleic acid extraction, during the whole process starting from sample collection, processing and storage.

SOLUTION: An entry-level quality check to confirm the sample is fit for purpose.

41. In your opinion, this would be:

Not needed

Helpful

Crucial to develop

42. Which parameters beyond adherence to standards would be needed to be checked

for fixed, paraffin- embedded samples	
for fresh frozen tissue	
for white blood cells (PBMC)	
for liquid biopsies	

43. Is the information above confidential?

Yes, it is confidential

No, it can be shared

PROBLEM Sample transport and storage might be required which can change the nucleic acids' quality/quantity.

SOLUTION: Stabilization of DNA and RNA (this can include all different types of DNA and RNA depending on the source and target situation)

44. Is your organization involved in R&D for such an approach?

O Yes

45. In case of R&D involvement, would financial support be:



Helpful

Crucial to develop this approach

Comment

46. Is the information above confidential?

Yes, it is confidential

No, it can be shared

47. PROBLEM Sample pre-analytical processing can influence the final result of NGS.

SOLUTION: The project participants have compiled a list of key challenges in sample and library preparation that could be improved within the next 2 - 3 years.

Please weight each of them on the basis of their importance from 1 (lower) to 5 (higher).

						IN/A
Increase in sample stability	0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0
Increase in sample quantity	0	\bigcirc	\bigcirc	0	0	0
Increase target concentration in the sample	\odot	\odot	\bigcirc	\bigcirc	\odot	\odot
Direct or long range sequencing	\bigcirc	\odot	\odot	\odot	0	0
Alternative innovative stabilization methods	0	\odot	\bigcirc	\odot	\bigcirc	0
Compatibility of sample stabilizers with analytical test procedure that need to be performed or have same sample (e.g. histology/cytology)	0	0	0	be	en performed on th	ne
Efficiency of the analyte						
isolation/extraction procedure	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
isolation/extraction procedure Quality assessment of the isolated analyte	0	0	0	0	0	0
isolation/extraction procedure Quality assessment of the isolated analyte Sample storage	0	0	0			0
Initial context and state and state isolation/extraction procedure Quality assessment of the isolated analyte Sample storage Analyte storage						
Lindency of the analyteisolation/extractionprocedureQuality assessment of the isolated analyteSample storageAnalyte storageAutomation of the isolation procedures						
Lindency of the analyteisolation/extractionprocedureQuality assessment of the isolated analyteSample storageAnalyte storageAutomation of the isolation proceduresStandardization						
Lindency of the analyteisolation/extractionprocedureQuality assessment of the isolated analyteSample storageAnalyte storageAutomation of the isolation proceduresStandardizationOther						

48. Is your organization involved in R&D in one or more of these approaches?

O Yes

50. Is the informa	tion above confide	ntial?			
─ Yes, it is confid	lential				
◯ No, it can be s	hared				
51. In case of R8	D involvement, wo	uld financial supp	ort be		
Not needed					
helpful for					
crucial to deve	lop this approach				
Comment					
52 Is the informa	tion above confide	ntial?			
		indi:			
Yes, it is confi	lential				
Yes, it is confi No, it can be s	lential hared	11001:			
Yes, it is confi No, it can be s	lential hared	11001:			
Ves, it is confi No, it can be s	lential hared riority please expla	ain why?			
Yes, it is confi No, it can be s	lential hared riority please expla	ain why?			
Yes, it is confi No, it can be s	lential hared riority please expla	ain why?			
Yes, it is confi No, it can be s	lential hared	ain why?			
 Yes, it is confident of the first of the highest p 54. Is the information of the first of the highest p 	lential hared riority please expla	ain why?			
54. Is the information of the fight of the f	lential hared riority please expla tion above confide	ain why? ntial?			
54. Is the information of the fight of the f	lential hared tion above confide lential hared	ain why? ntial?			
54. Is the information of the fightest point	lential hared tion above confide lential hared	ain why? ntial?			
 Yes, it is confident of the highest p 54. Is the information of the highest p 54. Is the highest p 55. For the highest p 56. For the second h 	lential hared tion above confide lential hared	ain why? ntial? se explain why?			
 Yes, it is confident of the highest p For the highest p 54. Is the information of the highest p Yes, it is confident of the highest p The highest p For the second highest p 	lential hared tion above confide lential hared ighest priority plea	ain why? ntial? se explain why?			
 Yes, it is confident of the highest p For the highest p 54. Is the information of the highest p Yes, it is confident of the highest p No, it can be s For the second h 	lential hared tion above confide lential hared ighest priority plea	ain why? ntial? ise explain why?			
 Yes, it is confident of the highest p For the highest p 54. Is the information of the second h No, it can be second h 	lential hared tion above confide lential hared	ain why? ntial? ise explain why?			
 Yes, it is confident of the highest p 54. Is the information of the highest p 54. Is the information of the second highest p For the second highest p 	In above confide	ain why? Initial?			
 Yes, it is confident of the highest p 54. Is the information of the highest p 54. Is the information of the second highest p 56. Is the information of the second highest p 	tion above confide	ain why? Initial?			
 54. Is the information of the second here. 54. Is the information of the second here. 56. Is the information of the second here. 56. Is the information of the second here. 56. Is the information of the second here. 	tion above confide	ain why? ntial? se explain why?			

SOLUTION: Simultaneous Isolation of multiple nucleic acid types (e.g. DNA and RNA) in the same run. 51. Is your organization interested for further development in this approach? Yes No 58. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) 59. In case of R&D involvement, would financial support be Not needed relipful for organization above confidential? Yes, it is confidential No, it can be shared PROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and arge stocks that might expire before they can be used up. SOLUTION: Compatibility of nucleic acid isolation methods with multiple targets and applications. 61. Is your organization interested in further development in this approach? Yes No 62. In case of R&D involvement, would financial support be No	PROBLEM During nucleic a samples.	cid isolation, it is often impose	sible to obtain more than on	e NA target especially when	dealing with small size
57. Is your organization interested for further development in this approach? Yes No 58. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) 59. In case of R&D involvement, would financial support be Not needed No 90. In case of R&D involvement, would financial support be No No 60. Is the information above confidential? Yes, it is confidential No, it can be shared PROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kills to choose from and large stocks that might expire before they can be used up. SolutTION: Compatibility of nucleic acid isolation methods with multiple targets and applications. 61. Is your organization interested in further development in this approach? 62. In case of R&D involvement, would financial support be No	SOLUTION: Simultaneous I	solation of multiple nucleic ac	id types (e.g. DNA and RNA	A) in the same run.	
58. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher) S S S S S 59. In case of R&D involvement, would financial support be Not needed	57. Is your organiza Yes No	ation interested for furthe	er development in this	approach?	
S S S S 59. In case of R&D involvement, would financial support be Not needed helpful for crucial to develop this approach 60. Is the information above confidential? O Ves, it is confidential No, it can be shared PROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and large stocks that might expire before they can be used up. SOLUTION: Compatibility of nucleic acid isolation methods with multiple targets and applications. 61. Is your organization interested in further development in this approach? Yes No 62. In case of R&D involvement, would financial support be Not needed	58. Please weight the	importance of the furthe	r development of this a	approach from 1 (lower) to 5 (higher)
59. In case of R&D involvement, would financial support be Not needed helpful for crucial to develop this approach 60. Is the information above confidential? O' Yes, it is confidential No, it can be shared PROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and large stocks that might expire before they can be used up. SOLUTION: Compatibility of nucleic acid isolation methods with multiple targets and applications. 61. Is your organization interested in further development in this approach? Yes No 62. In case of R&D involvement, would financial support be Not needed					
 No, it can be shared PROBLEM Each target situation and type of nucleic acid imply different isolation procedures resulting in multiple kits to choose from and large stocks that might expire before they can be used up. SOLUTION: Compatibility of nucleic acid isolation methods with multiple targets and applications. 61. Is your organization interested in further development in this approach? Yes No 62. In case of R&D involvement, would financial support be Not needed 	59. In case of R&D inv Not needed helpful for crucial to develop this approach 60. Is the information Yes, it is confider	olvement, would financi	al support be		
SOLUTION: Compatibility of nucleic acid isolation methods with multiple targets and applications. 61. Is your organization interested in further development in this approach? Yes No 62. In case of R&D involvement, would financial support be Not needed	No, it can be sha PROBLEM Each target situa large stocks that might expir	red ation and type of nucleic acid re before they can be used up	imply different isolation proc	cedures resulting in multiple	kits to choose from and
61. Is your organization interested in further development in this approach? Yes No 62. In case of R&D involvement, would financial support be Not needed	SOLUTION: Compatibility o	f nucleic acid isolation method	ds with multiple targets and	applications.	
62. In case of R&D involvement, would financial support be Not needed	61. Is your organiza	ation interested in furthe	r development in this a	approach?	
Not needed	62. In case of R&D inv	olvement, would financi	al support be		
	Not needed				
helpful for	helpful for				
crucial to develop this approach	crucial to develop this approach				

63.	Is the	information	above	confidential?
•••				

Yes, it is confidential

No, it can be shared

PROBLEM Panels are constantly in need of extension for addition of new diagnostic marker sequences and need to be verified and validated before use. This complicates library preparation due to optimal selection of changing panels to diagnostically cover the case to be analysed.

SOLUTION: Whole Genome Sequencing (WGS) and Whole Exome Sequencing (WES) instead of targeted NGS approach

64. Is your organization interested in further development in this approach?

O Yes

65. In case of R&D involvement, would financial support available in the Instand-NGS4P project be

Not needed	
helpful for	
crucial to develop this approach	

66. Is the information above confidential?

Yes, it is confidential

No, it can be shared

PROBLEM The ligation of adapters is a critical step in library preparation. Insufficient adapter ligation leads to low library complexity, especially for low input samples

67. Which approach(es) are you currently using:

amplicon-based library preparation

full length adapters

stubby adapters

Other (please specify)

68. Is the information above confidential?

Yes, it is confidential

No, it can be shared

69. What is the gu	uaranteed ligation effica	cy you can acquire:					
~ 70%							
70-80%							
80-90%	0 80-90%						
90-95%							
>95%							
	adautau daaine and likuawaan						
SOLUTION: Improve the a	adapter design and library pr	eparation protocol.					
1. use of alternatives t	to adapter- and amplicon-bas	sed approaches					
2. improve adapter de	sign						
3. improve ligation pro	ocess						
70 1		fan mariel an marie a haa 2					
70. Is your organi.	zation involved in R&D	for novel approaches?					
O No							
If ves, give the	number of the solution your	organisation is involved in					
	······································						
71. Is the information	tion above confidential?)					
Yes, it is confid	ential						
No, it can be sl	hared						
72. Please weight the	e importance of the furt	her development of appr	roaches from 1 (lower) to	o 5 (higher):			
73. In case of R&D ir	nvolvement, would finar	ncial support be					
Not needed							
helpful for							
crucial to develop this							
approach							

	BLEM WGS and WES	S are difficult with FFPE tiss	ue.			
SOL	UTION:					
1.	1. Use alternatives for FFPE tissue, like frozen tissue					
2.	Use other fixation/ st	tabilization methods that pre	eserve the histologic qualities a	and stabilizes DNA and RN/	A for use in NGS	
	74 le vour organiz	ration interacted or inv	alved in P&D for such a	approach?		
	O No					
	If yes, please gi	ve the number of the solution	on your organization is interest	ted in (highest priority first)		
	75. Is the informati	ion above confidential?	?			
	Yes, it is confide	ential				
	○ No, it can be sh	ared				
76.	76. Please weight the importance of the further development of this approach from 1 (lower) to 5 (higher)					
77.	In case of R&D in	volvement, would finar	ncial support available in	the Instand-NGS4P p	roject be	
77. Not	In case of R&D in	volvement, would finar	ncial support available in	S the Instand-NGS4P p	roject be	
77. Not help	In case of R&D in needed ful for	volvement, would finar	ncial support available in	S the Instand-NGS4P p	roject be	
77. Not help cruc	In case of R&D in needed ful for ial to develop this	volvement, would finar	ncial support available in	S the Instand-NGS4P p	roject be	
77. Not help cruc appr	In case of R&D in needed ful for ial to develop this roach	volvement, would finar	ncial support available in	the Instand-NGS4P p	roject be	
77. Not help cruc appr	In case of R&D in needed ful for ial to develop this roach	Volvement, would finar	cfDNA and cfRNA, exosome a	the Instand-NGS4P p	roject be	
77. Not help cruc appr PRC SOL	In case of R&D in needed ful for ial to develop this oach DBLEM WGS and WES UTION:	Volvement, would finar	cfDNA and cfRNA, exosome a	the Instand-NGS4P p	roject be	
77. Not help cruc appr PRC SOL	In case of R&D in needed ful for ial to develop this roach DBLEM WGS and WES UTION: Maximise the yield a	volvement, would finar	ncial support available in	the Instand-NGS4P p	roject be	
77. Not help cruc appr PRC SOL 1.	In case of R&D in needed ful for ial to develop this roach DBLEM WGS and WES UTION: Maximise the yield a	volvement, would finar	ncial support available in	the Instand-NGS4P p	s roject be	
77. Not help cruc appr PRC SOL 1.	In case of R&D in needed ful for ial to develop this oach DBLEM WGS and WES UTION: Maximise the yield a Increase sequence s	volvement, would finar	cfDNA and cfRNA, exosome a	the Instand-NGS4P p	s roject be	
77. Not help cruc appr PRC SOL 1. 2.	In case of R&D in needed ful for ial to develop this oach DBLEM WGS and WES UTION: Maximise the yield a Increase sequence s	volvement, would finar	ncial support available in	the Instand-NGS4P p	roject be	
77. Not help cruc appr PRC SOL 1. 2.	In case of R&D in needed ful for ial to develop this oach DBLEM WGS and WES UTION: Maximise the yield a Increase sequence s	volvement, would finar	cfDNA and cfRNA, exosome a	the Instand-NGS4P p	roject be	
77. Not help cruc appr PRC SOL 1. 2.	In case of R&D in needed ful for ial to develop this oach DBLEM WGS and WES UTION: Maximise the yield a Increase sequence s	volvement, would finar	ncial support available in	the Instand-NGS4P p	roject be	

78. Is your organi	zation interested or inv	olved in R&D for such an	approach?			
O Yes						
🚫 No						
If yes, please give the	If yes, please give the number of the solution your organization is interested in (highest priority first):					
79. Is the informa	tion above confidential	?				
Yes, it is confic	lential					
O No, it can be s	hared					
80. Please weight th	e importance of the furt	her development of this a	approach from 1 (lowe	r) to 5 (higher)		
81. In case of R&D i	nvolvement, would finar	ncial support available in	the Instand-NGS4P p	roject be		
Not needed	,		- I]		
Not needed]		
helpful for						
crucial to develop this approach						
82. Is the informa	tion above confidential	2				
Yes, it is confic	lential					
No, it can be shared						
PROBLEM Complexity ar	nd the large number of steps	needed for the complete librar	y preparation, which can als	so differ in various		
specific situations, increas	ses the risk of error causing fa	allure or variations in the end re	esult.			
SOLUTION: Below you ca	an find a list of possible appr	paches to solve the problem.				

83. Please weight each o	of them on th	ne basis of their	importance fro	om 1 (lower) to	5 (higher).	
	1	2	3	4	5	N/A
Universal approach for different sample types and targets	0	\bigcirc	\odot	0	0	\bigcirc
Automation of library preparation	\bigcirc	0	\bigcirc	\odot	\bigcirc	0
Automation of nucleic acid isolation alone	\bigcirc	\bigcirc	\bigcirc	0	0	0
Automation from nucleic acid isolation (different targets and DNA and RNA, low and high yield) to sequencing (closed system)	0	0	0	Q	Ö	0
Direct or long range sequencing	0	0	0	0	\bigcirc	0
Reduced number of steps needed	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	0
UMIs mandatory (for mutation detection down to 0.01-0.1% allele frequency)	0	0	0	0	0	0
Improvement of library preparation success rate	\odot	\odot	\odot	\odot	\bigcirc	0
Improvement of library yield	\bigcirc	0	0	0	\bigcirc	0
Minimal quality and quantity requirements for	\bigcirc	0	0	O	e input material	
Other	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Other (please specify)						

O Yes, it is confidential

◯ No, it can be shared

Yes No Please give the n s the informatio	mbers of interest or inv	volvement with the l	nighest priority first, de	own to the last priori	tr <i>e</i>
No Please give the n	mbers of interest or inv	volvement with the I	nighest priority first, d	own to the last priori	tv.
Please give the n	mbers of interest or inv	volvement with the I	nighest priority first, d	own to the last priori	tv
s the informatio					ty.
s the informatio					
s the informatio					
	n above confidentia	al?			
Yes, it is confiden	al				
No, it can be shar	ed				
ase of R&D inv	lvement would fin	ancial support h			
r					
aevelop this					
nment					
s the informatio	n above confidentia	al?			
Yes, it is confiden	al				
No, it can be shar	ed				
s the informatio	n above confidentia	al?			
Yes, it is confiden	al				
No, it can be shar	ed				
M Waste of materia	due to package sizes	and stability of reag	ents		
DN:					
reased long term s	ability of reagents and	simplified storage re	equirements		
duce and recycle p	ckaging and consuma	bles			
alability of reagents					
	ase of R&D invo	ase of R&D involvement, would fin	ase of R&D involvement, would financial support b	ase of R&D involvement, would financial support be d d d d d d d d d d d d d d d d d d	ase of R&D involvement, would financial support be id id id id id id id id id i

91. Is your organiz	ization interested in further development in one or more of these approaches?	
⊖ Yes		
O No		
Please give the	e numbers of interest or involvement with the highest priority first, down to the last priority:	
92. Is the informat	ation above confidential?	
Yes, it is confide	dential	
◯ No, it can be sh	hared	
93. In case of R&D in	nvolvement, would financial support available in the Instand-NGS4P project be	
Not needed		
helpful for		
crucial to develop this		
approach		
94. Comment		
<u> </u>		

Lot 1, Page 2/2

CHALLENGE 2:

Integrating pre-analytical, analytical processes and data analytics into a standardized workflow

PROBLEM WGS and WES are difficult for FFPE material because of the multitude of chemical changes present in the sample.

SOLUTION: Alternative isolation/stabilisation methods suitable for WGS and WES need to be compatible with other diagnostic methods, like histology and cytology.

95. Is your organization interested in further development of such an approach?

O Yes

96. In case of R&D involvement, would financial support be

Not needed	
helpful for	
crucial to develop this approach	

97. Comment

98. Is the information above confidential?

Yes, it is confidential

No, it can be shared

PROBLEM WGS and WES are difficult for low yield samples (e.g. plasma cfDNA, cfRNA).

SOLUTION:

1. Alternative isolation/stabilisation methods giving highest yield possible

2. Increased sequencing sensitivity

99. Is your organ	99. Is your organization interested in the further development of such an approach?					
Yes						
No						
Please give th	Please give the numbers of interest or involvement with the highest priority first, down to the lowest priority:					
100. Is the inforn	nation above confidential?					
Yes, it is confi	dential					
◯ No, it can be s	shared					
101. In case of R&D) involvement, would financial support be					
Not needed						
helpful for						
crucial to develop this						
approach						
102. Comment						
103. Is the more						
Yes, it is confi	dential					
O No, it can be s	shared					
PPORI EM Isolatod nucl	nic acid proparations can contain interforing substances. This can impair the reliability of the results and the					
compatibility of isolation	nethods with multiple sequencing methods.					
SOLUTION:						
1. The isolates are tested for inhibitory substances for various sequencing methods						
2. Development of alternative procedures to avoid known interfering substances						
104. Is your orga	nization interested or involved in R&D for such an approach?					
Yes						
No						
\smile						

06. Comment 107. Is the information above confidential? Yes, it is confidential No, it can be shared ROBLEM Long turnaround times (for pre-analytics and library OLUTION: Below you can find a list of possible approaches f 08. Please weigh each of them on the basis of th 1 2 Faster nucleic acid isolation methods	y preparation). to solve the problem neir importance f 3	n. From 1 (lower) to	5 (higher) .	
107. Is the information above confidential? Yes, it is confidential No, it can be shared ROBLEM Long turnaround times (for pre-analytics and library OLUTION: Below you can find a list of possible approaches to 08. Please weigh each of them on the basis of the 1 2 Faster nucleic acid isolation methods	y preparation). to solve the problen neir importance f 3	n. From 1 (lower) to	5 (higher) .	
107. Is the information above confidential? Yes, it is confidential No, it can be shared ROBLEM Long turnaround times (for pre-analytics and library DLUTION: Below you can find a list of possible approaches to 08. Please weigh each of them on the basis of the 1 2 Faster nucleic acid solation methods	y preparation). to solve the problen neir importance f 3	n. Trom 1 (lower) to 4	5 (higher) .	
Yes, it is confidential No, it can be shared ROBLEM Long turnaround times (for pre-analytics and library DLUTION: Below you can find a list of possible approaches 08. Please weigh each of them on the basis of th 1 2 Faster nucleic acid solation methods	y preparation). to solve the problen neir importance f 3	^{n.} rom 1 (lower) to 4	5 (higher) .	
No, it can be shared ROBLEM Long turnaround times (for pre-analytics and library DLUTION: Below you can find a list of possible approaches No. Please weigh each of them on the basis of the 1 2 Faster nucleic acid solation methods	y preparation). to solve the problen neir importance f 3	n. Trom 1 (lower) to 4	5 (higher) .	
ROBLEM Long turnaround times (for pre-analytics and library DLUTION: Below you can find a list of possible approaches 08. Please weigh each of them on the basis of th 1 2 Faster nucleic acid solation methods	y preparation). to solve the problen neir importance f 3	n. Trom 1 (lower) to 4	5 (higher) .	
DLUTION: Below you can find a list of possible approaches 08. Please weigh each of them on the basis of th 1 2 Faster nucleic acid solation methods	to solve the problem neir importance f 3	n. Trom 1 (lower) to 4	5 (higher) .	
 Please weigh each of them on the basis of th 1 2 Faster nucleic acid solation methods 	neir importance f	rom 1 (lower) to	5 (higher) .	
1 2 Faster nucleic acid solation methods	neir importance f 3	rom 1 (lower) to 4	5 (higher) .	
1 2 Faster nucleic acid solation methods 0	3	4		
solation methods			5	N/A
	0	\odot	0	\odot
reparation procedures	0	0	0	\bigcirc
ntegration of multiple steps in nucleic acid O O	0	\odot	0	0
ntegration of multiple teps in library	0	\odot	preparation	
ntegration of multiple teps in isolation and O O O O O O O O O O O O O O O O O O O	0	\odot	0	0
Automation O	0	\bigcirc	0	0
Other O	0	0	\odot	0
her (please describe and specify if this information is confid-	lential)			

110. In case of R&D involvement, would financial support be not needed helpful crucial to develop this approach Comment 111. Is the information above confidential Yes O No **CHALLENGE 3:** Defining genetic variants with established medical implications for common and rare, adult and pediatric, cancers including pharmacogenetic variants relevant for therapy selection in cancer care PROBLEM High nucleic acid yields are needed as input for NGS whereas often only small samples are available and NGS is not always able to deal with difficult genomic regions, low-yield materials and somatic mutations at low variant allele frequency. SOLUTION: 1. Increase the efficiency of nucleic acid isolation methods 2. Increased sequencing sensitivity 112. Is your organization interested or involved in R&D for such an approach? O Yes No No Please give the numbers with the highest priority first, down to the lowest priority 113. Is the information above confidential? Yes, it is confidential No, it can be shared 114. In case of R&D involvement, would financial support be Not needed helpful for crucial to develop this approach

115. Comment	
116. Is the inform	ation above confidential?
Yes, it is confid	lential
◯ No, it can be s	hared
"lead"	
PROBLEM The NGS diag and hazardous drugs, alth	nostic result is used for determination of the treatment for patients. This involves the administration of complex nough it is not known if the drugs are metabolized normally.
SOLUTION: Including Pha	armacogenomic Analysis in panels targeted on somatic mutations
117. Is your orgai	nization interested in further development of such an approach?
⊖ Yes	
O No	
118. In case of R&D	involvement, would financial support available in the Instand-NGS4P project be
Not needed	
helpful for	
crucial to develop this	
approach	
119. Comment	
120. Is the inform	ation above confidential?
Yes, it is confid	lential
O No, it can be s	hared
CHALLENGE 4:	
Developing reference m	aterial for quality control
PROBLEM Lack of availa as a whole.	bility of suitable internal quality control (QC) approaches and reference materials to monitor the NGS workflow
SOLUTION Multiple OC	steps for each phase of the workflow to act as stop/go criteria to avoid wasting time and resources

121. Is your organi	zation interested in further development o	f such an approach?			
⊖ Yes					
O No					
122. In case of R&D II	ivolvement, would financial support be				
Not needed					
helpful for					
crucial to develop this					
approach					
123. Comment					
124. Is the informa	tion above confidential?				
Yes, it is confide	ntial				
No, it can be sha	ared				
<u> </u>					
PROBLEM Availability of w	ell-designed NGS performance testing (PT), Externa	al Quality Assessment (EQA) and reference material.			
SOLUTION: Use of PT, EQ	A and reference material for NGS workflows				
125. Please select the	box if your organization is using or wants	s to use it:			
	Develops	Produces			
PT					
EQA					
Reference material					
Please specify what kind and what is your source					
126. Is the informa	tion above confidential?				
Yes, it is confide	ntial				
⊖ No, it can be sha	ared				

127. The reference material:		
	Yes	Νο
is introduced into the workflow during pre- analytic processing	\bigcirc	\odot
matches authentic sample matrix	\bigcirc	\odot
covers diagnostically relevant variants	0	\odot
includes RNA	0	0
contains 'difficult' Variants Of interest	\bigcirc	\odot
Comment		

Yes, it is confidential

No, it can be shared

PROBLEM Patient identity of patient material is sometimes swapped with that of another patient. In doubt the material must be reidentified with ID SNP. This requires a lot of extra time.

Solution: Inclusion of patient ID SNP's in the sequence panel or result.

129. Is your organization involved in R&D for such an approach?

O Yes

O No

130. In case of R&D involvement, would financial support be

Not needed

helpful for

crucial to develop this approach

131. Comment

Yes, it is confidential

No, it can be shared

133. If you have any further comments in the context of pre-analytics and library preparation, please let us know.

134. Is the information above confidential?

Yes, it is confidential

No, it can be shared

Introduction Lot 2

* 135. Would you like to answer questions related to Lot 2 (Sequencing)?

⊖ Yes

O No

t 2						
6. How impor	tant is sequenci	ng time for you?	(0- not importa	nt at all, 5-very i	mportant)	
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\odot	\odot	\odot	0
7. What is yo ls?	ur maximum acc	ceptable time for	a routine seque	encing run in ho	urs, from library	to variant
8. How impor	tant are over-we	eekend runs for	you? (0- not imp	oortant at all, 5-v	ery important)	
0	1	2	3	4	5	N/A
		0	0	\bigcirc	\bigcirc	0
139. What is Per analy Per reag	your preferred p /sis ents structure	pricing model?				
139. What is Per analy Per reage Per infra: 0. How impor	your preferred p /sis ents structure tant is reduced l	pricing model? hands-on time fo	pr you? (0- not ir	mportant at all, 5	i-very important)
139. What is Per analy Per reage Per infra: 0. How impor	your preferred p ysis ents structure tant is reduced l 1	pricing model? hands-on time for 2	or you? (0- not ir 3	mportant at all, 5 4	i-very important <u>;</u> 5) N/A
139. What is Per analy Per reag Per infras	your preferred p /sis ents structure tant is reduced l 1	pricing model? hands-on time for 2	or you? (0- not ir 3	mportant at all, 5 4	5-very important) 5) N/A
139. What is Per analy Per reage Per infras 0. How impor 0 1. How much nds-on time?	your preferred p ysis ents structure tant is reduced l 1 0 would you prefe (0- not at all, 5-v	pricing model? hands-on time fo 2 er higher costs fo very much)	or you? (0- not ir 3 or reagents and	mportant at all, 5 4 automated solu	5-very important) 5 tions in exchang) N/A ge for reduced
139. What is Per analy Per reage Per infras 0. How import 0 1. How much nds-on time? 0	your preferred p ysis ents structure tant is reduced l 1 0 would you prefe (0- not at all, 5-v 1	pricing model? hands-on time fo 2 er higher costs fo very much) 2	or you? (0- not ir 3 or reagents and 3	mportant at all, 5 4 automated solu 4	5-very important) 5 tions in exchang 5) N/A ge for reduced N/A
139. What is Per analy Per reage Per infras 0. How impor 0 1. How much nds-on time? 0	your preferred p ysis ents structure tant is reduced f 1 0 would you prefe (0- not at all, 5-v 1	pricing model? hands-on time for 2 er higher costs for very much) 2	or you? (0- not ir 3 or reagents and 3	mportant at all, 5 4 automated solu 4	5-very important 5 tions in exchang 5) N/A ge for reduced N/A
139. What is Per analy Per reag Per infras 0. How impor 0 1. How much nds-on time? 0 142. Do you	your preferred p ysis ents structure tant is reduced l 1 would you prefe (0- not at all, 5-v 1 use reference m	pricing model? hands-on time fo 2 er higher costs fo very much) 2 naterial (inter-/in	or you? (0- not in 3 or reagents and 3 trarun control)?	mportant at all, 5 4 automated solu 4	5-very important) 5 tions in exchang 5) N/A ge for reduced N/A
139. What is Per analy Per reag Per infras 0. How impor 0 1. How much nds-on time? 0 142. Do you	your preferred p ysis ents structure tant is reduced f 1 would you prefe (0- not at all, 5-v 1 use reference m	pricing model? hands-on time fo 2 er higher costs fo very much) 2 naterial (inter-/in	or you? (0- not in 3 or reagents and 3 trarun control)?	mportant at all, 5 4 automated solu 4	5-very important) 5 tions in exchang 5) N/A ge for reduced N/A
139. What is Per analy Per reage Per infras 0. How import 0 1. How much nds-on time? 0 142. Do you Yes No	your preferred p ysis ents structure tant is reduced l 1 would you prefe (0- not at all, 5-v 1 use reference m	pricing model? hands-on time fo 2 er higher costs fo very much) 2 naterial (inter-/in	or you? (0- not ir 3 or reagents and 3 trarun control)?	mportant at all, 5 4 automated solu 4	5-very important) 5 tions in exchang 5) N/A ge for reduced N/A

🕥 No						
If yes, please spec	ify					
14. How importar	nt would longer	reads (>600bp) be for you?	(0- not importa	nt at all, 5-very im	iportant)
0	1	2	3	4	5	N/A
\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
15. How importar	nt is it for you to	read biochem	ical informatio	on (ex. methylat	tion status)? (0- no	ot important at
l, 5-very importai	nt)	0	0		F	N1/A
U	1	2	3	4	5	N/A
\cup			\cup	\bigcirc	0	\cup
17. Do you consi	der sequencing	noise a proble	em? (0- not at	all, 5-very muc	h)	
17. Do you consi 0	der sequencing	noise a proble 2	em? (0- not at 3	all, 5-very muc 4	h) 5	N/A
17. Do you consi 0	der sequencing 1	noise a proble 2	em? (0- not at 3	all, 5-very muc 4	h) 5	N/A
47. Do you consi 0	der sequencing 1	noise a proble 2	em? (0- not at 3	all, 5-very muc 4	h) 5	N/A
17. Do you consi 0 0 18. How big is th	der sequencing 1 O e impact of syst	noise a proble 2 O emic sequenci	em? (0- not at 3 O	all, 5-very muc 4 on your work? ((h) 5 O-no impact, 5-vei	N/A O ry big)
17. Do you consi 0 18. How big is the	der sequencing 1 e impact of syst	noise a proble 2 emic sequenci	em? (0- not at 3 O ing artefacts o 2	all, 5-very muc 4 on your work? ((3	h) 5 O-no impact, 5-vei 4 5	N/A O ry big) N/A
17. Do you consi 0 18. How big is the Homopolymers	der sequencing 1 e impact of syst	noise a proble 2 emic sequence 1	em? (0- not at 3 ing artefacts o 2	all, 5-very muc 4 on your work? (1 3	h) 5 0-no impact, 5-vei 4 5	N/A Pry big) N/A
 47. Do you consi 0 48. How big is the Homopolymers GC rich regions 	der sequencing 1 e impact of syst 0	noise a proble 2 emic sequenci 1	em? (0- not at 3 ing artefacts o 2	all, 5-very muc 4 on your work? (1 3	h) 5 0-no impact, 5-vei 4 5	N/A C Try big) N/A
47. Do you consi 0 48. How big is the Homopolymers GC rich regions	der sequencing 1 e impact of syst 0	noise a proble 2 emic sequence 1	em? (0- not at 3 ing artefacts o 2	all, 5-very muc 4 on your work? ((3	h) 5 0-no impact, 5-ver 4 5 0 0	N/A C Ty big) N/A
 47. Do you consi 0 48. How big is the 48. How big is the 40 on opolymers GC rich regions 40 on opolexity regions 	der sequencing 1 e impact of syst 0	noise a proble 2 emic sequence 1	em? (0- not at 3 ing artefacts o 2 0	all, 5-very muc 4 n your work? (1 3	h) 5 0-no impact, 5-ver 4 5 0 0	N/A C ry big) N/A C C C C C C C C C C C C C
 47. Do you consi 0 48. How big is the Homopolymers GC rich regions Low complexity regions 49. How important 	der sequencing 1 e impact of syst 0 ons	noise a proble 2 emic sequenci 1 0 0	em? (0- not at 3 ing artefacts o 2 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	all, 5-very muc 4 n your work? ((3 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	h) 5 0-no impact, 5-ver 4 5 0 0 0 0	N/A Pry big) N/A N/A N/A N/A N/A
 47. Do you consi 0 48. How big is the Homopolymers GC rich regions Low complexity regions 49. How important 0 	der sequencing 1 e impact of syst 0 ons) t is paired-end 1	noise a proble 2 emic sequence 1 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	em? (0- not at 3 ing artefacts o 2 0 0 1 0 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	all, 5-very muc 4 n your work? (1 3 0 0 0 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	h) 5 0-no impact, 5-ver 4 5 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	N/A N/A N/A N/A N/A t)
 47. Do you consi 0 48. How big is the Homopolymers GC rich regions Low complexity regions 49. How important 0 	der sequencing 1 e impact of syst 0 ins 0 ins 0	noise a proble 2 emic sequenci 1 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	em? (0- not at 3 ing artefacts o 2 0 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	all, 5-very muc 4 all, 5 all, 5-very muc 4 all, 5 all, 6 all, 6 al	h) 5 0-no impact, 5-ver 4 5 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	N/A N/A N/A N/A N/A N/A
 47. Do you consi 0 48. How big is the Homopolymers GC rich regions Low complexity regions 49. How important 0 	der sequencing 1 e impact of syst 0 ons 1 0 0 0 0 0 0 0 0 0 0 0 0 0	noise a proble 2 emic sequence 1 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	em? (0- not at 3 ing artefacts o 2 0 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	all, 5-very muc 4 n your work? (f 3 important at al 4	h) 5 0-no impact, 5-ver 4 5 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	N/A N/A N/A N/A N/A N/A
 47. Do you consi 0 48. How big is the 48. How big is the 49. How important 0 49. How important 0 50. How important 	der sequencing 1 e impact of syst 0 ins ins int is paired-end 1 int is the size of t	noise a proble 2 emic sequenci 1 sequencing fo 2 the instrument	em? (0- not at 3 ing artefacts o 2 0 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1 1	all, 5-very muc 4 an your work? (1 3 a important at al 4 ot important at	h) 5 0-no impact, 5-ver 4 5 0 0 0 0 1, 5-very importan 5 all, 5-very importa	N/A Pry big) N/A N/A N/A N/A N/A N/A
 47. Do you consi 0 48. How big is the Homopolymers GC rich regions Low complexity regions 49. How important 0 50. How important 0 	der sequencing 1 e impact of syst 0 ns 1 1 1 1 1 1 1 1 1 1 1	noise a proble 2 emic sequencia 1 sequencing fo 2 the instrument 2	em? (0- not at 3 ing artefacts o 2 0 1 1 1 1 1 1 1 1 1 1 1 1 1	all, 5-very muc 4 an your work? (1 3 a important at al 4 ot important at al 4	h) 5 0-no impact, 5-ver 4 5 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0 0	N/A N/A N/A N/A N/A N/A N/A

•		or the platform in	or scalable throu	ighput for you?	(U- not importan	it at all, 5-very
0	1	2	3	4	5	N/A
0	0	\bigcirc	\bigcirc	\bigcirc	0	\bigcirc
152. How import	ant is it for you	to pool various l	ibraries in one r	un? (0- not impo	ortant at all, 5-ve	ery important)
0	1	2	3	4	5	N/A
0	\bigcirc	\bigcirc	\odot	\bigcirc	\odot	\odot
153. Do you j	perform sequen	cing as a lab-de	veloped test?			
⊖ Yes						
O No						
154. How import	ant are IVDR-C	E certified tests	for you? 0- not i	important at all,	5-very importan	t)
0	1	2	3	4	5	N/A
\bigcirc	0	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
EE How import	tant are IVDP C	E instrumente fo	rvou20 notim	portont of all E	vory important)	
		2	3 3	1001tant at all, 5		N/A
0		-	0		0	
		~				~
1						
PROBLEM High nuc	leic acid yields are with difficult genomi	needed as input for ic regions, low-yield	⁻ NGS whereas ofter materials and soma	n only small sample tic mutations at low	s are available and variant allele freque	NGS is not ency.
PROBLEM High nuc lways able to deal v	cleic acid yields are with difficult genomi	needed as input for c regions, low-yield	[.] NGS whereas ofter materials and soma	n only small sample tic mutations at low	s are available and variant allele freque	NGS is not ency.
PROBLEM High nuc Navays able to deal v SOLUTION:	eleic acid yields are with difficult genomi fficiency of nucleic	needed as input for ic regions, low-yield acid isolation metho	NGS whereas often materials and soma	n only small sample tic mutations at low	s are available and variant allele freque	NGS is not ency.
PROBLEM High nuc always able to deal v SOLUTION: I. Increase the e	sleic acid yields are with difficult genomi sfficiency of nucleic uencing sensitivity.	needed as input for ic regions, low-yield acid isolation metho	[•] NGS whereas ofter materials and soma	n only small sample tic mutations at low	s are available and variant allele freque	NGS is not ency.
PROBLEM High nuc always able to deal v SOLUTION: I. Increase the e 2. Increased seq	cleic acid yields are with difficult genomi efficiency of nucleic uencing sensitivity.	needed as input for ic regions, low-yield acid isolation metho	NGS whereas often materials and soma	n only small sample tic mutations at low	s are available and variant allele freque	NGS is not ency.
PROBLEM High nuc Navays able to deal v SOLUTION: I. Increase the e 2. Increased seq 156. Is your c	cleic acid yields are with difficult genomi officiency of nucleic uencing sensitivity.	needed as input for ic regions, low-yield acid isolation metho erested or involv	NGS whereas often materials and soma ods ed in R&D for su	n only small sample tic mutations at low uch an approach	s are available and variant allele freque	NGS is not ency.
PROBLEM High nuc always able to deal v SOLUTION: I. Increase the e 2. Increased seq 156. Is your c Ves	cleic acid yields are with difficult genomi efficiency of nucleic uencing sensitivity.	needed as input for ic regions, low-yield acid isolation metho erested or involv	NGS whereas often materials and soma ods ed in R&D for su	n only small sample tic mutations at low uch an approach	s are available and variant allele freque	NGS is not ency.
PROBLEM High nuc always able to deal v SOLUTION: I. Increase the e 2. Increased seq 156. Is your c Ves	cleic acid yields are with difficult genomi efficiency of nucleic uencing sensitivity.	needed as input for ic regions, low-yield acid isolation metho erested or involv	NGS whereas often materials and soma	n only small sample tic mutations at low uch an approach	s are available and variant allele freque	NGS is not ency.
PROBLEM High nuc always able to deal v SOLUTION: I. Increase the e 156. Is your c Yes No 157. Please give	cleic acid yields are with difficult genomi efficiency of nucleic puencing sensitivity. organization inte	needed as input for ic regions, low-yield acid isolation metho erested or involv	NGS whereas often materials and soma ods ed in R&D for su priority first, down	n only small sample tic mutations at low uch an approach n to the lowest p	s are available and variant allele freque 1? oriority.	NGS is not ency.
PROBLEM High nuc always able to deal v SOLUTION: I. Increase the e 2. Increased seq 156. Is your c Yes No 157. Please give	cleic acid yields are with difficult genomi- efficiency of nucleic puencing sensitivity. organization inte	needed as input for ic regions, low-yield acid isolation metho erested or involv	NGS whereas ofter materials and soma ods ed in R&D for su	n only small sample tic mutations at low uch an approach n to the lowest p	s are available and variant allele freque n? oriority.	NGS is not
PROBLEM High nuc always able to deal v SOLUTION: I. Increase the e 2. Increased seq 156. Is your c Yes No 157. Please give	cleic acid yields are with difficult genomi efficiency of nucleic puencing sensitivity. organization inte	needed as input for ic regions, low-yield acid isolation metho erested or involv	NGS whereas ofter materials and soma ods ed in R&D for su	n only small sample tic mutations at low uch an approach n to the lowest p	s are available and variant allele freque n?	NGS is not ency.

Vac H	confidential					
	in be shared					
59 Comment						
160. Are the	comments confi	dential?				
Yes, the	y are confidential					
O No, they	r can be shared					
S1 If you have	any further com	ments in the o	ntext of secure	ncing please	let us know	
on. In you hav			Silexi of Seque	filling, please	iet us know.	
162. Is the i	nformation above	confidential?				
Yes						
Send Send						
O No						
⊖ No						
No No						
No No						
No No						
No No						
Νο						
Νο						
No						
No						
No						
No						
No						
No						
No						
Νο						

Introduction Lot 3

* 163. Would you like to answer questions related to Lot 3 (Bioinformatics)?

O Yes

O No

ot 3						
164. How is your or Personalized Thera	ganization / sp py field? (Plea	ecialized unit p se select all re	bositioned in the levant points).	e Next Generat Are you a?	ion Sequencin	g (NGS) for
Sequencing fac	lity					
Computational fa	cility					
Bioinformatics so	ftware developer					
Software service	provider					
Diagnostic facility	,					
Research facility						
Commercial com	pany					
Other (please sp	ecify)					
165. Does your ana (please check all th	alysis solution s at applies). plied to this questi	support bioinfo	rmatics data fro	om		
165. Does your ana (please check all th I have already re	alysis solution s at applies). plied to this questi 1	support bioinfor on in the context of	rmatics data fro of Lot 1. 3	om 4	5	N/A
165. Does your ana (please check all th I have already re 66. FFPE	alysis solution s at applies). plied to this questi 1	support bioinfor on in the context of 2	rmatics data fro	2 4	5	N/A
165. Does your ana (please check all th I have already re 66. FFPE	alysis solution s at applies). plied to this questing 1	2	rmatics data fro	4	5	N/A
165. Does your and (please check all th I have already re 66. FFPE	alysis solution s at applies). plied to this questi	2	rmatics data fro	4	5	N/A
165. Does your ana (please check all th I have already re 66. FFPE	alysis solution s at applies). plied to this questing 1	2 2 2 2	rmatics data fro of Lot 1.	2 4 	5	N/A
165. Does your and (please check all th I have already re 66. FFPE DNA RNA 57. Frozen tissues	alysis solution s at applies). plied to this questing 1	2 2 2 2 2 1 1 2 1 1 1 1 1 1 1 1 1 1 1 1	rmatics data fro	4 	5	N/A
165. Does your ana (please check all th I have already re 66. FFPE DNA RNA 67. Frozen tissues	alysis solution s at applies). plied to this questing 1 1 1 1	2 2 2 2 2 1 1 2 1 1 1 1 1 1 1 1 1 1 1 1	rmatics data fro	A	5	N/A
165. Does your and (please check all th I have already re 66. FFPE DNA RNA 67. Frozen tissues	alysis solution s at applies). plied to this questi 1	2 2 2 2 2 1 1 2 1 1 1 1 1 1 1 1 1 1 1 1	rmatics data fro	A	5 	N/A
165. Does your ana (please check all th ☐ I have already re 36. FFPE DNA RNA 37. Frozen tissues	alysis solution s at applies). plied to this questi 1	2 2 2 2 1 1 2 1 1 1 1 1 1 1 1 1 1 1 1 1	rmatics data fro	4 	5 	N/A
165. Does your ana (please check all th I have already re 6. FFPE NA NA 7. Frozen tissues	alysis solution s at applies). plied to this questi 1	support bioinfor	rmatics data fro	A	5	N/A

168. Blood/plasma						
	1	2	3	4	5	N/A
DNA						
RNA						
cfDNA	[
cfRNA						
69. Fine needle asp	oirates					
	1	2	3	4	5	N/A
DNA						
RNA						
170. Circulating Tum	or Cells					
	1	2	3	4	5	N/A
DNA						
RNA						
171. Extracellular ve	sicles					
	1	2	3	4	5	N/A
DNA						
RNA						
72. Saliva						
	1	2	3	4	5	N/A
DNA	[
173. Other						
	1	2	3	4	5	N/A
DNA						
RNA						
174. If other, please	specify and let u	s know if this d	ata is confiden	ti al.		

175.	what is the indication that you are looking for a solution for?
\bigcirc	Adult cancer
0	Pediatric cancer
0	Rare diseases
0	Other (please specify)
176	Which type of NGS data do you process?
170.	
0	
0	Whole Exome Sequencing (WES)
0	
0	Chill acc
0	Methylation
\cup	
\bigcirc	Other (please specify)
177.	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis?
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
177. (Plea	Where are the raw NGS (unprocessed) data produced for the bioinformatics analysis? ase choose all relevant points) Inhouse Another department External non-commercial organization Commercial provider Other (please specify)
(Plea	ase choose all relevant points)
-------	---
	At the place of NGS data generation
	Inhouse
	Another department
	External non-commercial organization
	Commercial provider
	Other (please specify)
179.	Who assesses the results of the NGS bioinformatics analysis for actionable items?
(Plea	ase choose all relevant points)
	The user
	We provide a service for the user
	Other (please specify)
180.	How do users provide the raw NGS data for the bioinformatics analysis?
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify)
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify)
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify) How do you currently access the results of the NGS bioinformatics analysis?
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify) How do you currently access the results of the NGS bioinformatics analysis? ase choose all relevant points)
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify) How do you currently access the results of the NGS bioinformatics analysis? ase choose all relevant points) By downloading from a server
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify) How do you currently access the results of the NGS bioinformatics analysis? ase choose all relevant points) By downloading from a server By running our solution locally
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify) How do you currently access the results of the NGS bioinformatics analysis? ase choose all relevant points) By downloading from a server By running our solution locally Other (please specify)
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify) How do you currently access the results of the NGS bioinformatics analysis? Asse choose all relevant points) By downloading from a server By running our solution locally Other (please specify)
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify) How do you currently access the results of the NGS bioinformatics analysis? ase choose all relevant points) By downloading from a server By running our solution locally Other (please specify)
180.	How do users provide the raw NGS data for the bioinformatics analysis? By uploading to a server By running our solution locally Other (please specify) How do you currently access the results of the NGS bioinformatics analysis? Asse choose all relevant points) By downloading from a server By running our solution locally Other (please specify)

182. Which type of software is used for the bioinformatics analysis?	
(Please choose all relevant points)	
open-source software (Non-commercial)	
open-source software (Commercial)	
Commercial software (source code not available)	
Other (nlesse specify)	
183. Do you use any workflow manager / workflow description standard? (multiple answer possible)	
No, I only use command line	
No, I only support my stand-alone solution	
Yes, Galaxy	
Yes, Common Workflow Language (CWL)	
Yes, Taverna	
Yes, Orange	
Yes, KNIME	
Yes, Workflow Definition Language (WDL)	
Yes, Next Flow	
Yes, Snake Make	
Yes, other (please specify)	
184. Which file formats are used to exchange data?	
○ FASTQ	
BAM / CRAM	
○ VCF	
⊖ csv	
OSC	
Other (please specify)	

(Plea	se choose all relevant points)
	GNU General Public License
	MIT License
	Apache License
	BSD License
	Commercial products
	Not sure
	Other (please specify)
196 1	What two of appotations do you currently got?
100.	
0	Variant consulation frequency
0	Variant Effect prediction
	Disease gene panel
0	Disease-gene association
0	Clinical actionable items
Õ	Drug-gene interactions
0	Dosage sensitivity
0	Other (please specify)
187.	s the annotation information from
(Plea	se choose all relevant points)
	Findable Accessible Interoperable Reusable (FAIR) data providers
	Free available data
	Commercial data
	Other (please specify)

188.	What kind of QC do you perform for your bioinformatics pipeline?
0	Validation using criteria for accredited labs, shared with users
0	CE certification by notified bodies
0	Other (please specify)
180	Is the NGS bioinformatics analysis certified / accredited to
100.	ISO27001:2013 (information security standard)
\sim	IEC 62304 (Software Lifeevelo)
	ISO 14155-2011 (Clinical investigation of medical devices for human subjects)
	ISO 14103.2011 (Chinical Investigation of medical devices for Human subjects)
\bigcirc	ISO13485:2016 (Medical devices)
\subseteq	
0	Other (please specify)
Future of	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	r NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.
Future of NGS bioin widely ap	f NGS bioinformatics analysis for diagnostics nformatics analyses have been performed in a research setting for a few years, but their use in the diagnostic setting is not plied yet.

190. Which challenges are the highest immediate priority? (scoring 0-5)							
C C	0	1	2	3	4	5	
Automation / Reduced manual work	0	0	0	\bigcirc	\bigcirc	Q	
Reproducibility	\bigcirc	\odot	\odot	\bigcirc	\odot	0	
Packaging (e.g. containers)	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	0	
Portability	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	0	
Long-term availability	\bigcirc	\odot	\bigcirc	\bigcirc	\bigcirc	\odot	
Long-term maintainability	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	0	
Standardized data exchange	\bigcirc	\bigcirc	\bigcirc	0	\bigcirc	0	
Standardized data storage formats	\odot	0	0	\odot	\odot	0	
Encryption / data security	0	0	\bigcirc	\bigcirc	0	Q	
Other (please specify)							

191. Which challenges are the highest long-term priority? (scoring 0-5)

	0	1	2	3	4	5
Automation / Reduced manual work	\bigcirc	\bigcirc	\bigcirc	0	0	0
Reproducibility	\bigcirc	\bigcirc	\odot	0	\odot	\bigcirc
Packaging (e.g. containers)	\bigcirc	\odot	\bigcirc	\odot	\bigcirc	0
Portability	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc
Long-term availability	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0	0
Long-term maintainability	\bigcirc	\odot	\bigcirc	0	\bigcirc	0
Standardized data exchange	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	0
Standardized data storage formats	\bigcirc	\odot	\bigcirc	\odot	\bigcirc	0
Encryption / data security	\bigcirc	\odot	\odot	\odot	0	0
Other (please specify)						

Software and pipelines

Automation offers the execution of different software products at different steps, which allows a standardized and reproducible analysis at different places. This can include different software solutions but ultimately allows harmonizing the detection of genomic events.

192. What do you believe are the key challenges to overcome in relation to introducing standardized pipelines and software to detect actionable items for diagnostics purposes? Score each (0-5)

	0	1	2	3	4	5
managing software versions	0	0	0	\bigcirc	0	\bigcirc
compatibility between tools or databases	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	0
reproducibility	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	0
complying with standard data formats	\bigcirc	\bigcirc	\bigcirc	\odot	0	\odot
adjusting workflow for different customers	\bigcirc	\odot	\bigcirc	\odot	0	0
integration in open- source pipeline systems	\odot	\bigcirc	\bigcirc	\odot	0	0
testing of pipelines with standardized samples	0	\bigcirc	0	\odot	\odot	0
scalability to whole genome sequencing	\odot	\odot	\bigcirc	\odot	0	\odot
Other (please specify)						

Which are in your opinion the relevant standards on this topic and how ready are they to be used in diagnostics? (please rate the relevant standards for readiness 0-5)

193. Software file formats and secure data exchange protocols

	0	1	2	3	4	5
BAM/SAM/FASTQ format	\bigcirc	\odot	0	0	\bigcirc	0
VCF format	\bigcirc	\bigcirc	\odot	\bigcirc	\odot	\bigcirc
gVCF format	\bigcirc	0	\bigcirc	\odot	0	\odot
GA4GH HTSGet	\odot	\odot	\odot	\bigcirc	\odot	\odot
Encryption (Crypt4GH)	0	0	\bigcirc	\odot	\bigcirc	0
CRAM format	\odot	\bigcirc	\bigcirc	\bigcirc	\odot	
0					Other (please	e specify)

ISO standards

ISO 20397-2:2021 "Biotechnology - Massively parallel sequencing - Part 2: Quality evaluation of sequencing data"

194. Please score the relevance of software license models regarding their use in diagnostics? (score each 0-5)

	0	1	2	3	4	5
Open Source (https://docs.github.com/en/github/creating- cloning-and-archiving- repositories/licensing-a- repository#choosing-the-right-license)	\bigcirc	0	0	\bigcirc	0	\bigcirc
Closed source	\odot	\bigcirc	\odot	\odot	\odot	\odot
BSD 3	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc	\odot
Apache	\bigcirc	\bigcirc	\odot	\odot	\bigcirc	\odot
СС	\bigcirc	\bigcirc	\odot	\odot	\odot	\odot
GNU GPL (v1,v2, v3)	\bigcirc	\bigcirc	\odot	\odot	\bigcirc	\odot
OSL	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc	\bigcirc
MIT	\bigcirc	\bigcirc	\odot	\bigcirc	\odot	\odot
Other (please specify)						

Annotation resources

The annotation of variants or genomic events is fundamental to identify actionable items.

195. What do you believe are the key challenges to overcome the standardized use of annotation resources for diagnostics purposes?

Up	odating external resources
Sta	andardized list of approved annotation resources
Qu	ality of resources (CE marking, ISO standard)
Lif	etime of resource
Siz	ze of resource
Lic	censing of resources
Ot	her (please specify)

Data storage and sharing

The storing and sharing of data is an integral part of a bioinformatics pipeline. The types of data include raw NGS data, result files from the pipeline as well as metadata.

Finally, it needs to be decided which data should be stored, where should they be stored, for how long and who should have access to?

196. What do you believe are the key challenges to overcome in relation to storing & sharing of relevant data for diagnostics purposes? score each (0-5)

	0	1	2	3	4	5
Location of storage (GDPR)	\odot	0	0	\bigcirc	0	0
GA4GH standards	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	0
encryption	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	\bigcirc
longtime storage	\bigcirc	\bigcirc	\odot	\odot	\odot	\odot
Patient access	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	\odot
Data size	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot	
\bigcirc					Other (p	lease
specify)						

Security

The acquired NGS data in diagnostics are very sensitive and can identify patients.

197. What do you believe are the key challenges to overcome in relation to the secure handling of NGS data in diagnostics? score each (0-5)

	0	1	2	3	4	5
standardization	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
awareness of personnel	\bigcirc	\odot	\bigcirc	0	\odot	0
training of personnel	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
quality of solutions (CE marked)	0	\bigcirc	0	0	\bigcirc	0
security updates	\bigcirc	\odot	\odot	\odot	0	0
GDPR requirements properly addressed to	0	0	0	Ona	U tional implementati	ion
Other (please specify)						

198. If you have any further comments in the context of bioinformatics analysis, please let us know.

199. Is this information confidential?

O Yes

SOLUTION PROVIDERS Questionnaire

Introduction Lot 4

* 200. Would you like to answer questions related to Lot 4 (Integrated reporting)?

⊖ Yes

O No

SOLUTION PROVIDERS Questionnaire

Lot 4

The aim of Lot 4 is to provide innovative solutions for translating NGS results into medical decision-making reports. This should be achieved by integrating NGS results with pharmacogenomics panels and existing e-medication tools containing information on dosing and drug interactions. Since this information has to be made available to healthcare professionals and patients at the bedside for rapid interpretation, it will be important to determine the optimal method to clearly present NGS results and their medical relevance. The relevant clinical information should be reported in a concise and clear way, reporting only data with validated evidence for clinical decisions and in a form minimizing the risk of data misinterpretation. The questionnaire should help to define the specifications of the integrated reporting tool.

201. Do you use tools for integrated reporting?

- () Yes
- O No

202. Are you interested in implementing tools for integrated reporting?

O Yes

203. 32. Are you a solution provider for integrated reporting tools?

- Yes
- O No

204. Do you have a solution for integrated reporting of NGS results that integrates the following data and information (multiple answers may apply)

	Yes	No
Cancer-related variants	\bigcirc	\odot
Actionable items	0	\bigcirc
Pharmacogenomic variants	0	\odot
Level of evidence for cancer-related variants (level of evidence could be companion- diagnostics, drug-label, guidelines, databases, scientific literature)	Q	\odot
Level of evidence for pharmacogenetic variants (level of evidence could be for example companion-diagnostics, drug-label, guidelines, databases, scientific literature)	\bigcirc	\bigcirc
Information on clinical trials	0	0
Other (please specify)]

205. How important is the integration of the following data and information in reporting of NGS results in order to support medical decision making (please select score; 0=not relevant; 5=highly relevant).

	0	1	2	3	4	5	N/A
Information on informed consent	\bigcirc	\bigcirc	\bigcirc	\bigcirc	0	\bigcirc	\bigcirc
Information on the sample analyzed	0	\odot	\bigcirc	\bigcirc	0	\bigcirc	\odot
Information on the analytical method	\cdot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Information on the quality of the analysis	0	\odot	0	\odot	0	\bigcirc	\bigcirc
Results on cancer – related variants	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Information on actionable items	\odot	\bigcirc	\bigcirc	\bigcirc	\odot	\odot	\bigcirc
Information on pharmacogenomics variants	0	\odot		\bigcirc	\odot	\bigcirc	Ō
Information on the level of evidence for cancer- related variants	\odot	\bigcirc	\bigcirc	\odot	0	0	\bigcirc
Information on the level of evidence for pharmacogenomics variants	0	\bigcirc	\bigcirc	0	0	\bigcirc	\odot
Information on drug-drug interaction, dosing, side effects and contra indications	0	0	0	0	0	0	0
(e.g., e-medication)							
Information on relevant clinical data (e.g., heart, liver, kidney function)	0	\odot	0	0	\odot	0	\odot
Information on running clinical trials	0	\odot	0	\odot	0	0	0
Information on possible compassionate use	0	\bigcirc	0	\bigcirc	0	\bigcirc	\bigcirc
Other (please specify)							

206. Please indicate how important is the following information on clinical evidence for decision making. Evidence based on the following facts (please select score; 0=not relevant; 5=highly relevant).

	0	1	2	3	4	5	N/A
Companion-diagnostics	\bigcirc	\odot	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc
Drug-label	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc
Guidelines of medical societies	\odot	\odot	\bigcirc	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Curated databases	\odot	\odot	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc
Scientific literature	\bigcirc						
Information on open clinical trials – national	\odot	0	0	0	0	0	\odot
Information on open clinical trials – Europe	0	\bigcirc	0	\bigcirc	0	0	\bigcirc
Information on open clinical trials – international	0	0	\bigcirc	0	0	0	\bigcirc

What is your preferred reference for assessing the clinical evidence? Please describe

207. How important are the following features of integrated reporting for decision support

(please select score; 0=not relevant; 5=highly relevant)

	0	1	2	3	4	5	N/A
Desktop solution	\odot	\bigcirc	\odot	0	\bigcirc	\bigcirc	\bigcirc
Mobile device (e.g., tablet 12')	\bigcirc	\bigcirc	\odot	\odot	\bigcirc	\bigcirc	\bigcirc
Touch screen	\odot	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	\bigcirc
WLAN	\odot	\bigcirc	\bigcirc	\odot	\bigcirc	\odot	\bigcirc
Off-line mode	0	\odot	\odot	\bigcirc	\odot	\odot	\odot
Other (please specify)							

208. How important is graphical presentation (data visualization) of results for decision support? (please select score; 0=not relevant; 5=highly relevant)

0	1	2	3	4	5	N/A
\odot						

If ves please describe	the solution						
0. How important i	s it that the res	sults of integ	rated reporting	g are also integra	ated in electro	onic health	records
nk) and the hospi		Systems (mi	S)? (please se	elect score, 0-no	t Televant, 5-		vant)
	0	1	2	3	4	5	N/A
Relevance of integratior n EHR	0	\odot	\odot	\bigcirc	0	0	\odot
televance of integratior າ HIS	0	\odot	\odot	\bigcirc	0	0	0
lease describe:							
lease describe: 212. Have you tes Yes No	sted different p	products for in	ntegrated repo	orting and decision	on support?		
lease describe: 212. Have you tes Yes No If yes, describe which	sted different p products	products for in	ntegrated repo	orting and decisio	on support?		
lease describe: 212. Have you tes Yes No If yes, describe which	sted different p	products for in	ntegrated repo	orting and decisio	on support?		
lease describe: 212. Have you tes Yes No If yes, describe which 3. How satisfied an lease select score; 0	sted different p products re you with cur 0=not relevant 1	rrent products t; 5=highly re	ntegrated repo s on the mark elevant) 3	orting and decision	on support? reporting and	d decision s	upport? N/A
lease describe: 212. Have you tes Yes No If yes, describe which 3. How satisfied at lease select score; 0	sted different p products re you with cur 0=not relevant 1	products for in rrent products t; 5=highly re 2	ntegrated reports s on the mark elevant) 3	orting and decision et for integrated 4	on support? reporting and 5	d decision s	upport? N/A
lease describe: 212. Have you tes Yes No If yes, describe which 3. How satisfied at lease select score; 0 4. How important if patients? (please 0	sted different p products re you with cur 0=not relevant 1 s it that the inte select score; C	egrated repo products for in t; 5=highly re 2 egrated repo)=not relevar	ntegrated reports s on the mark elevant) 3 rting and deci nt; 5=highly re 3	et for integrated 4 ision support sys levant) 4	on support? reporting and 5 tem generate	d decision s es a special	upport? N/A I report
lease describe: 212. Have you tes Yes No If yes, describe which a. How satisfied and lease select score; 0 4. How important in patients? (please 0	sted different p products re you with cur 0=not relevant 1 s it that the into select score; 0	egrated repo prot relevan 2	ntegrated reports s on the mark elevant) 3 arting and deci at; 5=highly re 3	et for integrated 4 ision support sys levant) 4	on support? reporting and 5 tem generate 5	d decision s es a special	upport? N/A I report N/A

215. How important is i	t that the da	ta analysis fo	or integrated	reporting is p	erformed loo	ally or via a			
webservice? (please select score; 0=not relevant; 5=highly relevant)									
	0	1	2	3	4	5	N/A		
Local data analysis	\odot	\bigcirc	\bigcirc	\bigcirc	\odot	\bigcirc	0		
Data analysis via webservice	0	\odot	\odot	\odot	\odot	\bigcirc	\bigcirc		

216. If you have any further comments in the context of integrated reporting and e-medication, please let us know.

217. Is this information confidential?

O Yes O No

SOLUTION PROVIDERS Questionnaire

Last question

218. Would you like to share with us any additional information related to any section of the questionnaire? If yes please use the free field to do so.

219. Is the provided information confidential?

