INSTAND-NGS4P
Lot 4

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Current medical needs for reporting of NGS results in molecular pathology

1. Supportive variant annotation tool embedded within the NGS diagnostic pipeline

2. Communication tool between Molecular Tumor Boards to share molecular results and clinical experience

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1. Variant annotation - Current challenges

- DNA and RNA biomarkers with diagnostic and/or predictive value.
- Size of panels in diagnostic service are increasing

  e.g. ThermoFisher - Oncomine, Illumina – TSO500, WGS/WES

- The number of tumor types and stages of disease analysed with NGS is increasing
- Increase in the number of unknown, uncommon and composite variants with uncertain clinical relevance.

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1. Variant annotation – Current challenges

- Interpretation of variants is most often a subjective and manual process
- What variants are reported: Pathogenicity (AMCG) / druggability (AMP / ESCAT)
  Manually interrogate different (public) support tools: Franklin, CKB, ClinVar, VarSEAK, …
- Procedures from sample prep to NGS is already in large part automated.

Objective 1

CE-IVDR cloud-based variant knowledge database integrated into the NGS pipeline to support harmonized data interpretation in a clinical context with transfer of output to local pathology reporting systems.

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2. Communication between Molecular Tumor Boards (MTBs)

- NGS results can impact treatment decisions
- Complex and rare variants should be disscussed in MTBs[1-3]

  (Bi)weekly multidisciplinairy meeting: oncologist, pathologists, clinical scientist in molecular pathology, …
- Local MTBs learn from their experiences

  → professional self-educating system that improves local patient care.

1. Koopman et al., JCO Precision Oncology 2020
2. Koopman et al., The Oncologist, 2020
3. Dutch National NSCLC guideline, 2020

Study-proposal for INSTAND-NGS4P-LOT 4 by Dr. Léon van Kempen and Prof. dr. Ed Schuuring (UMCG, NL)

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2. Communication between Molecular Tumor Boards (MTBs)

- Create a network of MTBs for consultation of rare cases
- Exchange of knowledge between MTBs
- Develop clinical evidence for druggability of uncommon or composite variants to improve patient care

Objective 2

Communication tool between MTBs that allows patient-confidentiality compliant sharing of molecular profiles and treatment outcome.

Sharing rare cases will reduce their rarity

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