

Master level thesis/degree project at Clinical Genomics Uppsala

Amplicon Mapping in clinical next-generation sequencing

Background

Next-generation sequencing (NGS) is increasingly used in the clinical setting, a transition from traditional sequencing technologies. The more advanced technology requires tools to process the large amount of data produced, but promises better diagnostic yields, more personalized treatment strategies etc.

In general, technologies for enrichment of library pools for targeted sequencing can be subdivided into two main groups, target capture by hybridization and amplicon sequencing. The facility is using amplicon based strategies for several assays, in particular for the detection of somatic variants in several types of cancer. These assays have proven good to filter FFPE artefacts and detect relatively low frequency variants. When working with amplicon it is relevant to map what amplicon any single read originated from, a procedure known as amplicon mapping.

The interest of using unique molecular identifiers, UMI, is increasing globally. The UMIs can remove PCR artefacts, improve the overall sensitivity of the assays thus providing new possibilities to improve our clinical assays.

Aim

The aim of the project is to develop a tool for amplicon mapping that also processes UMIs. You will be working together with a team of bioinformaticians at the facility and the work will be subdivided into different parts:

1. Studying our current method for amplicon mapping
2. Specifying required features for the tool
3. Screening for available UMI and amplicon mapping tools (open source)
4. Develop the tool
5. If time permits, implementation into our pipeline tool

More specifications are available upon request from interested applicants.

About us

Within the Clinical Genomics Uppsala facility (www.scilifelab.se/facilities/clinical-genomics-uppsala) we implement NGS analyses in the clinical setting. For that purpose, we develop tools and pipelines for clinical NGS analyses, in particular for inherited disorders, hematology and solid tumors.

Questions and/or application:

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