Master level thesis/degree project at Clinical Genomics Uppsala

Coverage analysis 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.

Most bioinformatics tools that have been developed for processing NGS data are developed for use in the research setting. Most often they can be applied in the clinical setting with no or minimal modifications, but one exception is good tools to analyze the coverage of specific genetic regions that have been analyzed.

Coverage is often referred to as either horizontal or vertical coverage. Most scientific reports on NGS coverage would state that XX% of the targets (horizontal) were covered by at least YYx times (vertical) coverage. In the clinical setting however, we often need to know that ALL bases of a certain gene, or list of genes, has been sequenced with sufficient coverage in order to state that a patient sample has been analyzed correctly.

Aim

The aim of the project is to develop a tool for reporting coverage of NGS data. 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 reporting coverage
2. Specifying required features for the bioinformatics coverage tool
3. Screening for available tools (open source)
4. Developing 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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