



LEIDEN UNIVERSITY MEDICAL CENTER

NGS data flow

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Overview

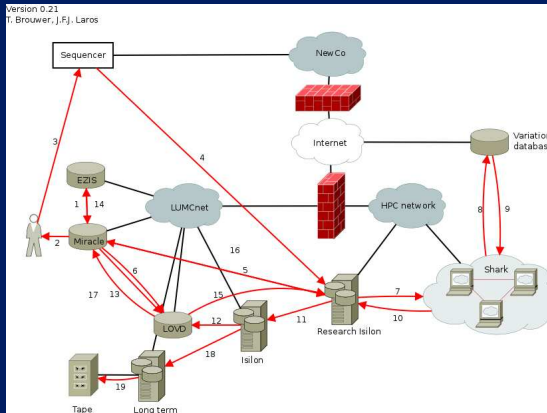


Figure 1: Data flow for NGS of clinical samples.

Overview

Before sequencing:

- Miracle receives pedigree information and an order for sequencing.
- Miracle makes a batch for the sequencing company using its own IDs.
 - DNA is extracted and put in a test tube, barcode is linked to ID.
 - Digital sample sheet is made for the sequencing company.
 - ID.
 - Test (exome / target / array / NIPT).

Overview

Primary data analysis:

- Miracle checks that sequencing does not take too long.
- Raw data arrives.
 - Miracle gets a signal that the raw data is available.
- Miracle sends pedigree information and the test to the pipeline.
- Primary data analysis on the cluster.
 - Miracle gets a signal when the primary data analysis is finished or has failed.

Overview

Secondary data analysis:

- Miracle sends IDs to the analysis program.
- Annotated varianten are analysed with the analysis program.
 - Miracle gets a signal when the data is loaded.
- Secondary data analyse in starts.
 - Miracle receives list of variants for validation.
 - Miracle receives primers.
- Validation tests are done and reported back to Miracle.
- Miracle sends results back to analysis program.
- Miracle reports back to patient registration system.

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