COVREPORT

VISUALIZATION TOOL FOR NGS REPORTS

APPLICATION

The visualization tool aims to broaden the scope of NGS users and facilitate coverage reading of NGS reports for clinicians in their everyday practice.

PROBLEM ADDRESSED

New Generation Sequencing revolutionized genome sequencing and is getting more and more available. However the comprehension of the generated reports is still a complicated task for non specialists.

An important task is making NGS reports easy to interpret.
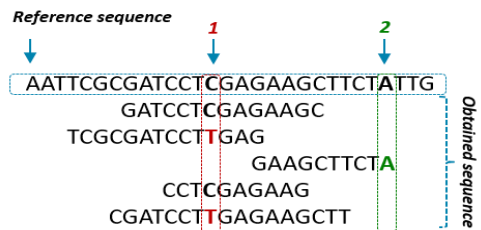
TECHNOLOGY

CovReport is a new gene sequencing coverage visualization tool that greatly facilitates the interpretation of diagnostic test sequencing report reports.

It generates a concise summary of the gene coverage allowing to evaluate the performance of the sequencing test at a glance.

- Position 1 has 4x coverage (description of its sequencing depth) meaning that this position was « seen » 4 times ;
- Position 2 has 1x coverage .

Figure 1. Read coverage of genome sequencing

As a result, the clinician can manage the diagnostic work-up in a more efficient way by requesting additional sequencing tests for the regions with insufficient coverage. A user-friendly interface generates a graphical summary of the coverage that can be directly included in the patient's report.
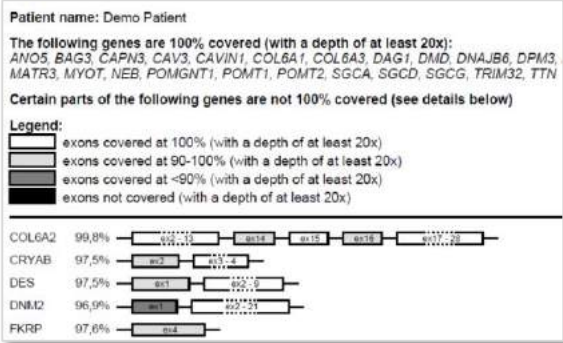

Figure 2. CovReport vizualisation sample

ADVANTAGES VS. STATE OF THE ART

✓ Reliable scientific solution issued from a public research institution specializing in bioinformatics;

✓ User friendly : easy to use tool, readable and understandable reports, suitable for clinicians without special genetics-related training;

✓ In situ approval : the tool has been tested for a year in real hospital conditions and proved its efficiency (including several diagnosis changes).