

NGS Proficiency Testing Program

Objectives of the NGS PT scheme:

- To maintain a high level of your NGS-service.
- To monitor flaws in your NGS-based testing using pre-characterized DNA samples.
- To provide feedback on the laboratory's NGS procedure.
- To provide guidance and recommendations for improvements in your NGS protocol.

The NGS PT Program

Twice a year, QualiGene sends a blinded, purified human genomic DNA sample diluted in TE buffer, harboring pre-characterized mutations (detectable by NGS), and instructions specifying the tested gene and reference NM number. The laboratory determines the sample's genotype and submits the results to QualiGene for evaluation. QualiGene in turn verify verifies the accuracy of the analysis, and will reports its findings back to the participating laboratory.

NGS PT Laboratory Practice

- The laboratory will treat the PT test item as if it was a routine NGS test in the laboratory
- The NGS test will be carried out by the methods routinely used in the lab
- The result analysis will be carried out using the software routinely used in the lab, to analyse NGS results

NGS PT material storage instructions

- The DNA samples should be stored at 4°C.

The NGS PT Protocol

- QualiGene will inform all participating laboratories two weeks before sample shipment.
 - A submission date will be set for **90 days from delivery day**. (The delivery day will be announced and **late submission, without a prior approval for extension by QualiGene, will result in grade reduction**)
 - The results will be submitted by fax or e-mail, on a result form provided by QualiGene.
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- The FASTQ files will be uploaded to our FTP server (username and password details will be provided)
- The grades will not be reported back to the participating laboratories, before all participant have submitted their results
- The lab will receive QualiGene's evaluation and feed-back within four weeks from the results due date.
- At the end of each year, participating laboratories will receive a certificate attesting to their participation in QualiGene's PT program.
- QualiGene will publish an annual newsletter containing announcements, NGS PT related statistics, general information and conclusions, which were drawn from the past PT schemes.

Appeals

- Each participating laboratory may appeal the grade, reported back by QualiGene, up to one week from result receipt
- Appeals should be submitted by e-mail, specifying the appeal arguments and reasons
- QualiGene will re-evaluate the results submitted by the participating laboratory, and will report its decision to the laboratory up to 10 days from appeal receipt.

Confidentiality

- All participating lab's names are kept confidential.
- QualiGene's evaluation and grading is confidential and will be submitted back only to the laboratory manager.
- Statistics and grades published in the newsletter are anonymous.
- Grades of Israeli participating laboratories are reported to the MOH, annually

Grading (percent of total grade)

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| • Correct identification of the mutation | 50% |
| • Coverage per target region | 35% |
| • False positive calls | 5% |
| • Timely submission of results | 10% |