

MLPA PT program

The Objectives of the MLPA PT scheme:

- To maintain a high level of MLPA-based tests.
- To monitor flaws in MLPA testing by using pre-characterized DNA samples.
- To provide feedback on the lab's MLPA testing procedure.
- To provide guidance and recommendations for MLPA testing improvements.

The MLPA PT Program

Twice a year, QualiGene sends a blinded, purified human genomic DNA sample diluted in TE buffer, harboring one or more specific mutations (detectable by MLPA), a gene-specific probemix and instructions specifying the tested gene. The laboratory determines the sample's genotype and submits the results to QualiGene for evaluation. QualiGene in turn verifies the accuracy of the analysis and reports its findings back to the participating laboratory.

MLPA PT Laboratory Practice

- The laboratory needs to treat the PT test samples as if these were routine MLPA test samples in the laboratory
- The laboratory will use the probemix and DNA samples, according to MLPA kit protocol
- The result analysis needs to be carried out using coffalyzer software, provided by MRC-Holland

MLPA PT material storage instructions

- The DNA samples should be stored at 4°C.
- The probemix should be stored at -20°C, if not used three days from the day of receipt (could be kept at RT if used within three days)

The MLPA 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**)
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- The results will be submitted by fax or e-mail, on a result form provided by QualiGene.
- The raw data will be attached to the result form and submitted to QualiGene.
- The lab will receive QualiGene's evaluation and feed-back within four weeks from the results submission deadline
- The grades and PT results will not be reported to the participating labs before all participants have submitted their results
- At the end of each year, the registered labs will receive a certificate attesting their participation in QualiGene's PT program.
- QualiGene will publish a report following each PT round, containing MLPA PT related statistics, general information and conclusions

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 reasons for the appeal and supporting arguments
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 laboratories' names are kept strictly confidential.
- QualiGene's evaluation and grading of your results is confidential and will be submitted back only to the laboratory manager.
- Statistics and grades published in QualiGene's report are always anonymous. Grades of Israeli laboratories are reported annually to the Israeli MOH.

Grading (percent of total grade)

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| • Correct identification of deletions and duplications | 40% |
| • Quality of MLPA reaction | 35% |
| • Electrophoresis parameters | 15% |
| • Timely submission of the results | 10% |