

PGD for single gene diseases- PT program

The Objectives of the PGD PT Scheme:

- To maintain a high level of the PGD process. The PGD PT will enable laboratories to evaluate their Amplification Rate, Allele Drop-Out Rate and to detect contaminations, if present.
- To monitor flaws in PGD by using 10 pg DNA samples (mimicking embryos) and pre-characterized DNA samples for haplotype analysis.
- To provide feedback on the lab's PGD procedure.
- To provide guidance and recommendations for PGD improvements.

The PGD PT Program

Once a year, participating laboratories are challenged with a new PGD case. The labs are provided with DNA samples in TE buffer from parents and from an affected sibling. In addition, 10 pg DNA samples from two or more additional siblings are provided (mimicking the embryos). The lab is required to genotype, create and analyze the haplotypes, and select an embryo to transfer back to the woman. The lab needs to share with QualiGene the PGD results as well as the haplotype analysis.

PGD PT Laboratory Practice

- The laboratory needs to treat the PT test samples as if it was a routine PGD test in the laboratory
- The haplotype analysis should be carried out using the primers used routinely in the laboratory

PGD PT material storage instructions

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

The PGD PT Protocol

- QualiGene will inform all participating laboratories two weeks before sample shipment.
 - A date for submission of results 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 form provided by QualiGene.
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- The haplotype analysis and test results will be attached to the result and submitted to QualiGene.
- The lab will receive QualiGene's evaluation and feed-back within four weeks from the results due date.
- At the end of each year, registered labs will receive a certificate attesting their participation in QualiGene's PT program.
- QualiGene will publish an annual report with PGD PT related statistics, general information as well as conclusions drawn from the latest PT scheme.

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 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)

- Correct identification of the 10 pg samples genotype (mimicking the embryos) 40%
- Genotyping procedure quality of the samples (mimicking embryos) 20%
- Haplotype analysis 30%
- Timely submission of results 10%