



## **Disease specific PT program**

### The Objectives of the disease specific PT scheme:

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

### The disease specific PT Program

Twice a year, QualiGene sends a certain number of DNA samples (The number of samples and the diseases will be determined according to the tested diseases in each participating laboratory). Each laboratory is required to determine the sample's genotype, using their routinely used methods. The participating laboratory needs to submit the results to QualiGene, which in-turn verifies the accuracy of the identified mutations and reports its findings back to the participating laboratory.

### Disease specific PT Laboratory Practice

- The laboratory needs to treat the PT test item as if it was a routine genetic testing sample, in the laboratory
- The methods and procedure need to be carried out by the materials and protocols, routinely used in the laboratory

### Disease specific PT material storage instructions

- The DNA samples should be stored at 4°C



### The Disease specific 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 samples will be numbered and the specific test for each sample will be specified in the result form, provided by QualiGene.
- The results will be submitted by fax or e-mail, using the result form provided by QualiGene.
- The lab will receive QualiGene's evaluation and feed-back within four weeks from the results submission deadline.
- The grades will not be reported back until 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.
- Following each PT round, QualiGene will publish a report containing announcements, disease specific 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 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 Criteria (percent of total grade)

- Correct identification of the mutations      50%
- Nomenclature      18%
- Final reports (for Israeli laboratories)      22%
- Timely submission of results      10%