Screenings Test for Inherited Disease (STID)

DESCRIPTION : **STID** (**S**creenings **T**est for **I**nherited **D**isease) is a test to screen for the most common gene anomalies (mutations) in common genetic diseases with recessive inheritance.

WHY STID : If a healthy couple carries a mutation in the same gene they have a 25 % risk that their offspring will be affected by a recessive disease. The overall frequency of such recessive diseases is 1 %, which is higher than the frequency of Down syndroom.
STID screens healthy couples for carriership of 409 frequent mutations in 104 common recessive disease genes. When both partners are carrier of a mutation in the same disease gene, prenatal diagnosis with chorion biopsy (CVS) or amniocentese (AC) can be offered.

SAMPLE : 5 ml blood from the couple (both husband and wife) has to be taken into an EDTA blood tube at the GENDIA lab in Antwerp, or sent at room temperature to GENDIA's lab in Antwerp, Belgium, and arrive there within 2 days of withdrawal.
Also DNA (5 ug) is suitable for STID.

METHODS : Samples are analyzed for the 409 most common mutations in 104 frequent genetic disease genes with recessive inheritance. The list of these diseases can be seen by clicking on Disease list, and a detailed description of the 104 genetic disease genes can be found by clicking on Disease description.

STID was developed and is performed by the Counsyl laboratory in the USA under the Clinical Laboratory Improvement Amendments (CLIA).

TURNAROUND TIME : STID takes 2 months to complete from arrival of the sample in the lab to the result report. In case of pregnancy STID can be expedited to 1 month.

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INDICATIONS : STID can be performed in all pregnancies without specific indication, as recessive disorders occur in the offspring of a healthy couple that is unaware they carry a mutation in the same gene.

LIMITATIONS : Samples are only analyzed for the 409 most common mutations in 104 frequent genetic disease genes with recessive inheritance. Other mutations in these genes or other genes are not analysed. Therefore, a normal STID test does not exclude carriership for any of the diseases tested.

RESULTS : STID results will be sent to the patient and/or physician who ordered the test. Genetic counseling with explanation of the test results and recommended follow-up steps if necessary, can be provided by genetic counselers from GENDIA.

FOLLOW UP :

1. In case of normal STID results in both partners : no specific follow up is necessary unless ultrasound examination of the fetus reveals anomalies and further fetal studies might be indicated.

2. In case one of the couple is carrier:

2A. If one of the partners is a carrier of a mutation in a rare disorder, and the calculated the risk for affected offspring remains low, no specific further genetic studies are necessary.

2B. If one of the partners is a carrier of a mutation in a frequent genetic disease (eg cystic fibrosis), further studies with complete sequencing of the gene might be indicated in the other partner.

3. In case both partners are carrier of a mutation in the same gene : in that case the risk that a child from the couple is affected with the disease is 25 %. Therefore, prenatal testing with molecular studies of the culprit gene after chorion biopsy (CVS) or amniocentesis (AC).

PRICE : 390 Euro.

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