Non - Invasive Prenatal Diagnosis (NIPD) for cystic fibrosis (CF)

Cystic fibrosis : Cystic fibrosis (CF) affects epithelia of the respiratory tract, exocrine pancreas, intestine, male genital tract, hepatobiliary system, and exocrine sweat glands, resulting in complex multisystem disease. Pulmonary disease is the major cause of morbidity and mortality in CF.

Inheritance : Cystic fibrosis (CF) is an autosomal recessive condition caused by mutations in the CFTR gene encoding the cystic fibrosis transmembrane conductance regulator. Both parents of an affected child are carriers of a single CFTR mutation, whereas their other CFTR gene is normal. The risk of another affected child inheriting both mutations from the parents is 25 %.

Methods : The CFTR gene can be analysed in DNA isolated from maternal blood (cell free DNA - cfDNA) which also contains fetal DNA. Such **N**on - **I**nvasive **P**renatal **D**iagnosis is referred to as NIPD. In contrast to invasive procedures such as amniocentesis (AC) or chorionic biopsy (CVS) that have an overall risk of miscarriage of 1 %, NIPD is non-invasive and has no risk for the fetus.

NIPD for CF is performed by relative haplotype dosage analysis (RHDO) using cell free fetal DNA (cffDNA) in pregnancies at risk of cystic fibrosis for confirmed CF carrier couples.
Relative haplotype dosage analysis (RHDO) is used to determine if the fetus has inherited the high risk allele from both parents.

We would require DNA/blood from both parents and a copy of their carrier mutation reports, plus a DNA sample from the proband (and mutation report) to carry out the test.

Such NIPD is only applicable to couples :

1) Who are known carriers of CFTR mutations

   AND

2) DNA is available from the affected proband and both parents

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Samples : At least 5 ug DNA or 5 ml EDTA blood is required from both parents.
The maternal blood in specific Streck blood tubes provided by GENDIA can be taken from week 10 of the pregnancy. The sample has to be sent by Express mail to GENDIA's lab in Antwerp, Belgium, and arrive there within 2 days of withdrawal.
All testing must be arranged in advance by emailing to NIPT@GENDIA.net. It is essential that the laboratory is advised of the pregnancy gestation and that this has been confirmed by ultrasound scan.

Turnaround time : NIPD takes approximately 2 weeks to complete from arrival of the sample in the GENDIA lab.

Contraindications : Samples from twin / multiple pregnancies or missed abortion / vanishing twin cannot be accepted for NIPD.

Reliability of NIPD results : The reliability of NIPD is very high (99 %).

Results : NIPD results will be sent to the physician / genetic counseler who ordered the test and who will explain the test results and recommended follow-up steps if necessary.

Follow up steps :

1. In case of a normal NIPD result : when only 1 or none of the 2 familial CFTR alleles associated with CF in the affected proband are detected by RHDO in the fetus, the fetus will not be affected with CF and no specific follow up is necessary, unless ultrasound examination of the fetus reveals anomalies and further genetic studies might be indicated.

2. In case of an abnormal NIPD result : when the 2 familial CFTR alleles associated with CF in the affected proband are detected by RHDO in the fetus, the fetus will be affected with CF.
In that case the parents will have to decide whether they want to continue the pregnancy after genetic counseling.

Price : 1800 Euros.

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