



Non - Invasive Prenatal Diagnosis (NIPD) for cystic fibrosis (CF), sickle cell anemia and Beta-thalassemia

DISORDERS TESTED BY NIPD :

- 1. Cystic fibrosis :** Cystic fibrosis (CF) is an autosomal recessive condition caused by mutations in the CFTR gene encoding the cystic fibrosis conductance regulator. It affects epithelia of the respiratory tract, exocrine pancreas, intestine, male genital tract, hepatobiliary system, and exocrine sweat glands, resulting in a complex multisystem disease. Pulmonary disease is the major cause of morbidity and mortality in CF. CF is the most common severe genetic disorder in Western population with a frequency of 1/3000.
- 2. Sickle-cell anaemia :** Sickle-cell anaemia (SCA) is an autosomal recessive condition caused by mutations in the HBB gene encoding B-globin. It results in an abnormality in the oxygen-carrying protein haemoglobin (haemoglobin S) found in red blood cells, leading to attacks of pain ("sickle-cell crisis"), bacterial infections, and stroke. The average life expectancy is 40 to 60 years. SCA is very common in Africa, India and the Middle-East.
- 3. Beta-thalassemia :** Beta-thalassemia is an autosomal recessive condition caused by mutations in the HBB gene encoding B-globin. It results in growth retardation, jaundice, hepatosplenomegaly, leg ulcers, development of masses from extramedullary hematopoiesis, and skeletal changes that result from expansion of the bone marrow. Regular transfusion therapy leads to iron overload-related complications including endocrine complication, cardiomyopathy, liver fibrosis and cirrhosis). B-thalassemia is the most common genetic disorder in mediterranean countries (Spain, Italy, Greece, turkey), North Africa, India and the Middle-East.

METHODS : Both the CFTR and the HBB gene can be analysed in DNA isolated from maternal blood (cell free DNA - cfDNA) which also contains fetal DNA. Such **Non - Invasive Prenatal Diagnosis** 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.

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SAMPLES :

1. 20 ml blood in a STRECK blood tube is required from the mother.
The maternal blood can be taken from week 11 of the pregnancy.
2. 10 ml blood in an EDTA blood tube is required from the father.

The sample have to be sent at room temperature by Express mail to GENDIA in Antwerp, Belgium, and arrive there within 2 days of withdrawal.

TURNAROUND TIME : NIPD takes approximately 3 weeks from arrival of the sample at GENDIA.

INDICATIONS : Every pregnancy.

LIMITATIONS OF NIPD : Samples are analyzed for mutations in the coding exons and intron-exon boundaries of the CFTR and the HBB genes. This does not exclude mutations elsewhere in the CFTR and the HBB genes, or mutations in other genes.

RELIABILITY : The reliability of NIPD is high (99 %).

RESULTS : NIPD results will be sent to the patient and the physician / genetic counselor 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 :** 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 :** Amniocentesis (AC) is advised to confirm the results of the NIPD.

PRICE : 650 Euros.