INFO BROCHURE NIPT

DETERMINATION OF FETAL SEX
IN CASE OF A SEX-LINKED DISORDER

Non - Invasive Prenatal Diagnosis of fetal sex in the case of a sex-linked disorder

Sex-linked disorders : Sex-linked disorders are genetic disorders transmitted through a single mutation in a gene located on the X chromosome. Most of the X-linked disorders are only present in men because men have only 1 X chromosome, whereas women have 2. Frequently female members are carriers of the mutation, but have no symptoms of the disease. The risk that a carrier female has an affected son is 1 on 4. The risk that an affected man transmits an X-linked disease to his sons is not increased as he transmits his Y and not his X chromosome to his sons.
As most of the X-linked disorders are only present in men, it is important to determine fetal sex in case of sex-linked disorders.

Methods : The fetal sex can be determined in DNA isolated from maternal blood (cell free DNA - cfDNA) which also contains fetal DNA. The sex of the fetus is determined by the presence of Y-specific sequence in the case of a male fetus and the absence of Y-specific material in the case of a female fetus. This test also includes screening for Down syndrome (trisomy 21), trisomy 18 and trisomy 13.
Such **N**on - **I**nvasive **P**renatal **T**est is referred to as NIPT. In contrast to invasive procedures such as amniocentesis (AC) or chorionic biopsy (CVS) that have an overall risk of miscarriage of 1 %, NIPT is non-invasive and has no risk for the fetus.

Samples : At least 20 ml blood in specific blood tubes provided by GENDIA is required from the mother. The maternal blood 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.

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

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Indications : NIPT for the determination of fetal sex can only be performed when the fetus is at risk for a documented X-linked genetic disorder.

Contraindications : NIPT for the determination of fetal sex can NOT be performed when :

1. The fetus is not at risk for a documented X-linked genetic disorder.

2. Samples from twin / multiple pregnancies or missed abortion / vanishing twin cannot be accepted for NIPT.

Limitations of NIPT : Samples are analyzed for the determination of fetal sex in case of sex-linked disorders only. The test also screens for the most common fetal aneuploidies Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau syndrome).

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

Results : NIPT 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 NIPT result indicating a female fetus : 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 a NIPT result indicating a male fetus: when the fetus is male, there is a risk that he will be affected with the sex-linked disorder. The physician / genetic counseler will discuss the implications of this finding with the parents. Amniocentesis (AC) or chorionic biopsy (CVS) to analyse whether the fetus is affected is possible when the mutation causing the X-linked disorders is known .

3. In case of an abnormal NIPT result indicating a fetal trisomy : in case of an abnormal result with the finding of an abnormal number (aneuploidy) of any of the 3 chromosomes tested (chromosomes 21, 18, and 13), the physician/genetic counseler will discuss the implications of such chromosomal anomaly with the patient, who can then decide to confirm the NIPT results with chromosome studies after amniocentesis or chorionic biopsy (CVS).

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