NIPT INFO BROCHURE

The Non - Invasive Prenatal Test (NIPT) is a DNA test on maternal blood to safely and reliably screen pregnancies for the most common fetal aneuploidies Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau syndrome).

Chromosome anomalies diagnosed with NIPT :

Trisomy 21 (Down syndrome)	This is caused by an extra copy of chromosome 21 and is also called Down syndrome. This is the most common genetic cause of intellectual disability. Individuals with Down syndrome have some degree of intellectual disability (average IQ of 50). Some children with Down syndrome have congenital defects of the heart or other organs that may require surgery or medical treatment. Some have other medical conditions including hearing or vision loss, and at a later age dementia.
Trisomy 18 (Edwards syndrome)	This is caused by an extra copy of chromosome 18 and is also called Edwards syndrome. Most babies with trisomy 18 have multiple severe birth defects of the brain, heart and other organs. Poor growth during pregnancy is common and many babies are miscarried or stillborn. Of those babies born alive, most die before one year of age. Babies who survive have profound intellectual disabilities and growth and developmental problems.
Trisomy 13 (Patau syndrome)	This is caused by an extra copy of chromosome 13 and is also called Patau syndrome. Most babies with trisomy 13 have multiple severe birth defects of the brain and other organs. Many babies are miscarried or stillborn. Of those babies born alive, most die before one year of age.

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Samples : At least 10 ml blood in a specific blood tube (STRECK) available from 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 3 days of withdrawal.

Methods : DNA isolated from maternal blood, which contains fetal DNA, is sequenced using an high-throughput next generation sequencer (NGS). Sequencing data are analyzed to determine the fetal copy number for chromosomes 21, 18, 13, thereby identifying any aneuploidy of any of these 3 chromosomes. The fetal sex can also be determined. This NIPT test (VERISEQ test) was developed by ILLUMINA in the United States and is carried out by the AML laboratory in Belgium.

Turnaround time : NIPT takes approximately 1 week from arrival of the sample in the GENDIA lab.

Indications : Although NIPT can be performed without specific indication, it might be most appropriate in pregnancies with an increased risk for fetal aneuploidy based upon a high maternal age (> 40 yrs) or abnormal result of the triple test (determination of AFP, oestriol, beta-hCG in maternal blood) or first trimester Down syndrome screening (NT measurement in combination with determination of free beta-hCG and PAPP-A in maternal blood). In these cases NIPT is an alternative to chromosome studies after amniocentesis or chorion biopsy (CVS).

Contraindications : Samples from triplet pregnancies cannot be accepted for testing. Twin testing is possible. NIPT is not the best option in case of ultrasound anomalies of the fetus.

Limitations of NIPT : in line with international recommendations the samples are analyzed for aneuploidy of chromosomes 21, 18, 13 only, whereas the determination of fetal sex is optional. Aneuploidy of other chromosomes, other chromosome anomalies (including mocaicism for chromosomes 21, 18, 13), and triploidy, molecular anomalies or congenital anomalies including neural tube defects cannot be excluded.

Reliability of NIPT results : The reliability of NIPT results is very high. The test has very high sensitivity and specificity for trisomy 21, 18 and 13, and very high reliability for the determination of fetal sex.

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NIPT failures: In a limited number of pregnancies (< 1 %) NIPT cannot be performed for technical reasons (not enough fetal DNA, damaged sample or other technical reasons). In these pregnancies NIPT can be repeated at no extra cost on a repeat maternal blood sample.

NIPT results : NIPT results will be sent to the physician/patient who ordered the test.

Follow up steps :

1. In case of a normal NIPT result : no specific follow up is necessary unless ultrasound examination of the fetus reveals anomalies and further fetal studies might be indicated.

2. In case of test failure : in a limited number of pregnancies (< 1 %) NIPT cannot be performed for technical reasons. In these pregnancies NIPT can be repeated at no extra cost on a repeat maternal blood sample.

3. In case of an abnormal NIPT result : 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 (AC) or chorion biopsy (CVS).

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