Prenatalsafe®

eurofins | Clinical Genetics

Form No. MARKETING1 Issue No. 1.00 Active Date 25/04/2025

NON-INVASIVE PRENATAL TESTING

NIPT

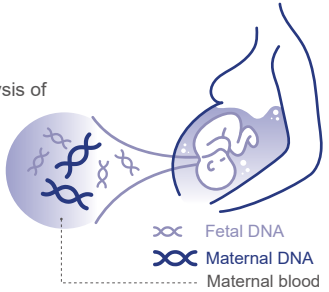
Non-invasive prenatal testing (NIPT), since its introduction into clinical practice over 10 years ago, has contributed greatly to the area of prenatal diagnostics[1]. NIPT has established itself as a safe alternative to invasive investigations (i.e., amniocentesis and villocentesis), while ensuring high sensitivity by comparison to serological testing, e.g. Bi-Test.

Recommended for **all pregnant women**

HOW DOES **NIPT WORK**?

NIPT is a non-invasive test that allows the analysis of fetal genetic material with a simple blood sample from the mother.

The test can detect fetal DNA circulating in maternal blood and analyse this DNA to identify the presence of chromosomal abnormalities and genetic diseases in the fetus.

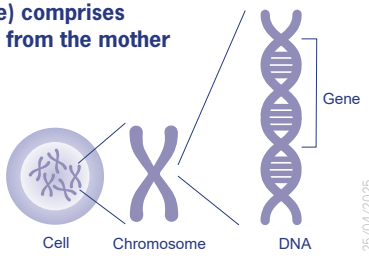


The amount of fetal DNA present in maternal blood increases throughout the pregnancy and is generally adequate for screening from week 10 of gestation. If the amount of fetal DNA in the sample is not adequate (determined during testing), it may be recommended that a new sample is obtained from the mother.

The chromosome set (called a karyotype) comprises 23 pairs of chromosomes, half inherited from the mother and half from the father:

- 22 pairs of non-sex chromosomes
- 1 pair of sex chromosomes

Chromosomes are formed from DNA. Some DNA segments are defined as GENES and provide the cell with the information required to perform its function.



Abnormalities in the process that leads to the formation of the embryo can cause different types of chromosomal alterations:

- Abnormalities in the **number** of chromosomes: ANEUPLOIDIES
- Abnormalities in the **structure** of CHROMOSOMES

Variations in the DNA sequence of the fetus, known as genetic mutations, may be inherited from the parents or occur for the first time in the fetus. These variations, depending on where they occur in the DNA sequence, can lead to different Genetic Diseases.

Genetic Diseases

Several studies have shown that the frequency of genetic alterations increases with maternal age. Advanced paternal age has also been shown to be a risk factor.

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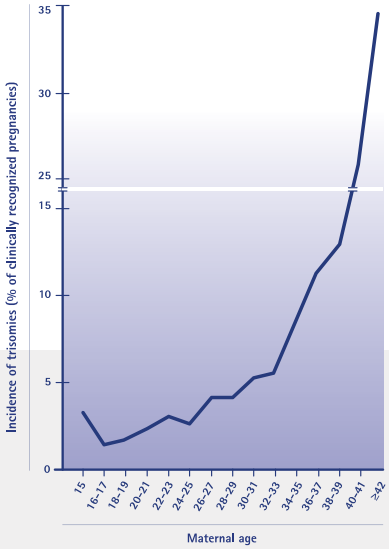
WHAT CAN BE INVESTIGATED
WITH NIPT?

1) Abnormalities in the number of chromosomes (Aneuploidies)

TRISOMY: three copies of a chromosome

MONOSOMY: single copy of a chromosome



Common Aneuploidies[2]

- Trisomy of chromosome 21 (Down Syndrome): 1 in 700 births
- Trisomy of chromosome 18 (Edwards Syndrome): 1 in 3000 births
- Trisomy of chromosome 13 (Patau Syndrome): 1 in 6000 births

Incidence increases with increasing maternal age[3].

2) Abnormalities in the structure of CHROMOSOMES

DELETION: loss of a chromosome segment

DUPLICATION: doubling of a chromosome segment

If these rearrangements are very small, they are called microdeletions and microduplications.

Microdeletion 22q11.3 is the most frequent microdeletion and is linked to DiGeorge syndrome, which has an incidence of 1 / 2000–4000 people, regardless of maternal age[4].

3) Genetic DISEASES

DE NOVO: caused by DNA mutations that occur for the first time in the fetus

HEREDITARY: caused by mutations inherited from parents

It is important to test specifically for the possibility of being a HEALTHY CARRIER*.

*A healthy carrier is an individual who can pass on a genetic mutation to their offspring, but is not affected by the mutation themselves

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Prenatalsafe®

In Eurofins Clinical Genetics Laboratory, Dublin, we currently offer Prenatalsafe® 3 & 5. The laboratory has been established since 2024 in partnership with Eurofins Genoma who have over 20 years experience in genetic testing.

	Prenatalsafe® 3 (PNS3)	Prenatalsafe® 5 (PNS5)
Fetal sex	●	●
Trisomy 21 Down Syndrome	●	●
Trisomy 18 Edwards Syndrome	●	●
Trisomy 13 Patau Syndrome	●	●
Sex Chromosome Aneuploidies		●

WHO IS IT FOR?

Any expectant mother, single or twin pregnancies, conceived either through natural conception or medically assisted reproductive technologies, whether autologous or heterologous.

Reporting times:

3-7 days

- PNS3 is available for singleton and twin pregnancies.
- PNS5 only available for singleton pregancies.
- Fetal sex determination in twin pregnancies only detects presence or absence of Y chromosome.

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Extended Testing Range

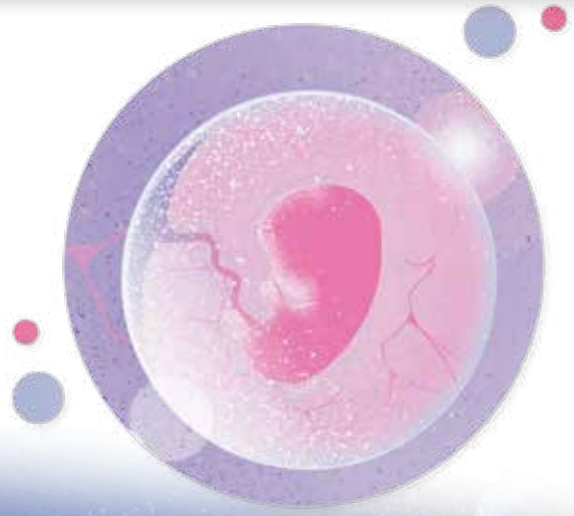
Through our partners Eurofins Genoma Italy we are able to offer the comprehensive range of Prenatalsafe® testing options. In the below table you can see that these additional options cover everything from the addition of DiGeorge screening test up to full Carrier Screening. For more information on these options please email sales@ctie.eurofinseu.com.

	Tested in Eurofins Genoma Italy						
Prenatalsafe®	5DiGeorge	Plus	Karyo	Karyo Plus	Complete	Complete Plus	Full Risk
Fetal sex	●	●	●	●	●	●	●
Trisomy 21 Down Syndrome	●	●	●	●	●	●	●
Trisomy 18 Edwards Syndrome	●	●	●	●	●	●	●
Trisomy 13 Patau Syndrome	●	●	●	●	●	●	●
Sex Chromosome Aneuploidies	●	●	●	●	●	●	●
Rare Autosomal Aneuploidies		9 and 16	●	●	●	●	●
Deletions and Duplications			●	●	●	●	●
Microdeletions	22q11.2	●		●		●	●
Inherited genetic diseases					●	●	●
De novo Genetic diseases					●	●	●
Carrier screening test*							●

Reporting times:

10-15 days
gene analysis

15-20 days
carrier testing on parents



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Microdeletions:

	Microdeletion Syndromes	Chromosome regions
Prenatalsafe® 5DiGeorge	DiGeorge Syndrome	deletion 22q11.2
Prenatalsafe® Plus	includes Prenatalsafe® 5DiGeorge +	
	Cri-du-chat Syndrome	deletion 5p15.3
	Prader-Willi Syndrome	deletion 15q11.2
	Angelman Syndrome	deletion 15q11.2
	1p36 Deletion Syndrome	deletion 1p36
	Wolf-Hirschhorn Syndrome	deletion 4p16.3
Prenatalsafe® Karyo Plus	includes Prenatalsafe® Plus +	
	Jacobsen Syndrome	deletion 11q23
	Langer-Giedion Syndrome	deletion 8q24.11-q24.13
	Smith-Magenis Syndrome	deletion 17p11.2

Inherited genetic diseases:

- CFTR Cystic Fibrosis
- CX26 (GJB2) Deafness Autosomal Recessive Type 1A
- CX30 (GJB6) Deafness Autosomal Recessive Type 1B
- HBB Beta Thalassemia
- HBB Sickle Cell Anemia

De novo genetic diseases:

Syndromic Disorders

Alagille Syndrome	JAG1
CHARGE Syndrome	CHD7
Cornelia de Lange Syndrome, type 5	HDAC8
Cornelia de Lange Syndrome, type 1	NIPBL
Rett Syndrome	MECP2
Sotos Syndrome, type 1	NSD1
Bohring-Opitz Syndrome	ASXL1
Schinzel-Giedion Syndrome	SETBP1
Holoprosencephaly	SIX3

Noonan Spectrum Disorders

Cardiofaciocutaneous Syndrome, type 1	BRAF
Noonan Syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSLL)	CBL
Noonan Syndrome, type 3	KRAS
Cardiofaciocutaneous Syndrome 3	MAP2K1
Cardiofaciocutaneous Syndrome 4	MAP2K2
Noonan Syndrome, type 6	NRAS
Noonan Syndrome, type 1 LEOPARD Syndrome 1	PTPN11
Noonan syndrome, type 5 LEOPARD Syndrome 2	RAF1
Noonan syndrome, type 8	RIT1
Noonan syndrome-like disorder with loose anagen hair	SHOC2
Noonan syndrome, type 4	SOS1

Skeletal Disorders

Achondrogenesis, type II	COL2A1
Achondroplasia	FGFR3
CATSHL Syndrome	
Crouzon syndrome with acanthosis nigricans	
Hypochondroplasia	
Muenke syndrome	
Thanatophoric dysplasia, type I	
Thanatophoric dysplasia, type II	COL1A1
Ehlers-Danlos syndrome, classic	
Ehlers-Danlossyndrome, type VIIA	
Osteogenesi imperfecta, type I	
Osteogenesi imperfecta, type II	
Osteogenesi imperfecta, type III	
Osteogenesi imperfecta, type IV	
Ehlers-Danlos Syndrome cardiac valvular form	COL1A2
Ehlers-Danlos, type VIIB Syndrome	
Osteogenesi imperfecta, type II	
Osteogenesi imperfecta, type III	
Osteogenesi imperfecta, type IV	

Craniosynostosis

Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	FGFR2
Apert Syndrome	
Crouzon Syndrome	
Jackson-Weiss Syndrome	
Pfeiffer Syndrome, type 1	
Pfeiffer Syndrome, type 2	
Pfeiffer Syndrome, type 3	

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Prenatalsafe® QUALITY PROVEN

LATEST GENERATION CE-IVD TECHNOLOGY

+

PROPRIETARY CE-IVD NIPT FLOW™ ALGORITHM

Sensitivity and specificity > 99% demonstrated on 71,740 pregnancies

	Sensitivity (95% CI)	Specificity (95% CI)
Common Aneuploidies		
Trisomy 21	99.54% (98.36% - 99.94%)	100% (96.11% - 100.00%)
Trisomy 18	100% (96.11% - 100.00%)	100% (99.99% - 100.00%)
Trisomy 13	100% (90.51% - 100.00%)	99.99% (99.98% - 100.00%)
Sex Chromosome Aneuploidies		
X0	98.11% (89.93% - 99.95%)	99.98% (99.97% - 99.99%)
XXX	100% (87.23% - 100.00%)	100% (99.99% - 100.00%)
XXY	100% (86.77% - 100.00%)	99.99% (99.99% - 100.00%)
XYY	100% (86.77% - 100.00%)	99.99% (99.99% - 100.00%)
Rare Autosomal Aneuploidies, Deletions, Duplications and Microdeletions		
Rare Autosomal Aneuploidies	100% (89.42% - 100.00%)	99.92% (99.89% - 99.95%)
Deletions and Duplications	100% (83.16% - 100.00%)	99.97% (99.96% - 99.99%)
Microdeletions	83.33% (35.88% - 99.58%)	99.99% (99.99% - 100.00%)

Validation on a large sample cohort (Eurofins Genoma)

- Analysis of over 70,000 samples for common aneuplodies (trisomies)
- Over 65,000 samples for sex chromosome aneuploidies
- Over 40,000 samples for other abnormalities

Reliability on all abnormalities

almost comparable to invasive investigation

Validation of rare Autosomal Aneuploidies (RAA), segmental abnormalities (deletions and duplications) and microdeletions was performed at Eurofins Genoma.

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Precision Genetic Testing
Improving Clinical Practice

Prenatalsafe®, combined with an accurate ultrasound investigation, allows early identification of fetal abnormalities.

Customer care available at any time on the path, from counselling to reporting

Logistics authorised for transporting biological material UN3373

Sample traceability

eurofins | Clinical Genetics

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www.clinicalgenetics.ie/non-invasive-prenatal-testing

Bibliography

1. Non invasive prenatal testing (NIPT) for common aneuploidies and beyond. Eur J Obstet Gynecol Reprod Biol 2021 Mar;258:424-429
2. Screening for Fetal Chromosomal Abnormalities. ACOG Practice Bulletin, Number 226. Obstetrics & Gynecology: October 2020 - Volume 136 - Issue 4 - p e48-e69
3. To err (meiotically) is human: the genesis of human aneuploidy. Nature Reviews Genetics volume 2, pages280–291 (2001)
4. Cell-free DNA screening for prenatal detection of 22q11.2 deletion syndrome. Maternal and Fetal Medicine, held virtually, January 25–30, 2021

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Eurofins Clinical Genetics,
Sample Reception

Eurofins Biomnis, Unit 3, Sandyford Business Centre, Sandyford Business Park, Dublin 18, D18 E528, Ireland.

Eurofins Genoma

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