

## **Biomnis**

## **Non-Invasive Prenatal Testing (NIPT)**



## How does it work?



During pregnancy, some fragments of the foetal DNA circulate in the maternal blood and are detectable starting from the 7<sup>th</sup> week of gestation. The circulating foetal DNA quantity increases with the increasing gestational period, and from the 10<sup>th</sup> week of gestation it is sufficient to guarantee a high specific and sensitive result of the NIPT screening test.

## **Eurofins NIPT Advantages**



### More than a simple NIPT screening test

# A range of options to choose which prenatal screening test is most suitable to you

	Commonly found trisomies				Other trisomies	All other chromosomes	Microdeletion syndromes		Single gene disorder	Foetal sex
		T18	T13	X/Y	T9 ස T16		6 panel	+3 panel		
PrenaTest	0									Ø
Ninalia 3	0	0	<b>Ø</b>							0
Ninalia <b>5</b>	0	0	0	0						0
) <sup>(5)</sup> PrenatalSafe* <b>3</b>	0	0	<b>Ø</b>							0
) <sup>(5)</sup> PrenatalSafe* <b>5</b>	Ø	Ø	<b>Ø</b>	Ø						0
MPrenatalSafe* <b>Pius</b>	0	Ø	<b>Ø</b>	0	0		0			0
PrenatalSafe*	Ø	Ø	<b>Ø</b>	Ø	0	0				0
PrenatalSafe*Plus	Ø	Ø	<b>Ø</b>	Ø	0	0	0	0		0
PrenatalSafe <sup>®</sup>	0	Ø	<b>I</b>	0	0	0			<b>Ø</b>	0
PrenatalSafe"Pius	Ø	Ø	Ø	Ø	0	0	Ø	0	Ø	0
, GeneSafe									0	

Through analysis of circulating cell-free foetal DNA (cffDNA) in maternal blood, the NIPT test screens for common trisomies, genome-wide chromosomal abnormalities, microdeletion syndromes, and single gene level genetic disorders in the foetus.

Always consult your healthcare provider before deciding which test is right for you.

#### **Chromosomal Aneuploidy**

The test screens for chromosomal abnormalities in the foetus, for example, the presence of whole extra chromosome (trisomy) such as Down Syndrome, Edwards Syndrome, Patau Syndrome, Trisomy X, Klinefelter Syndrome, and Jacobs Syndrome, or absence of whole chromosomes (monosomy), such as Turner Syndrome.

#### Structural Chromosomal Alterations

Analyses abnormalities in every chromosome of the foetal genome, providing karyotype-level insight. It provides information about gains or losses of chromosomal material across the genome.

#### **Microdeletion Syndromes**

Chromosomal abnormalities which are very small in size. The test screens for up to 9 clinically significant microdeletion syndromes. A panel of 6 or a panel of 9 microdeletion syndromes screening is available.

1	DiGeorge syndrome 22q11.2 deletion	1 in 4,000	Learning problems, congenital heart defects, palatal abnormalities			
2	1p36 deletion syndrome	1 in 4,000 to 1 in 10,000	Characteristic craniofacial features, intellectual disability, seizures, brain and heart defects			
3	Angelman syndrome (15q11.2 deletion)	1 in 12,000	ntellectual disability, speech impairment, seizures			
4	Prader-Willi syndrome (15q11.2 deletion)	1 in 10,000 to 1 in 25,000	Hypotonia, morbid obesity, delayed motor and language skills, intellectual disability, hypogonadism			
5	Cri du Chat syndrome (5p15.3 deletion)	1 in 20,000 to 1 in 50,000	Intellectual disability, speech delay, cat-like cry			
6	Wolf-Hirschhorn syndrome (4p16.3 deletion)	1 in 50, 000	Growth deficiency, hypotonia, craniofacial features, intellectual disability, heart and brain abnormalities			
7	Langer-Giedion syndrome (8q24 deletion)	1 in 200,000	Benign bone tumours (exostoses), short stature, and distinctive facial features			
8	Jacobsen syndrome (11q23 deletion)	1 in 100,000	Developmental delay, distinctive facial, bleeding disorders and some behaviour disorders			
9	Smith-Magenis syndrome (17p11.2 deletion)	1 in 15,000 to 1 in 25,000	Variable intellectual deficit, sleep disturbance, craniofacial and skeletal anomalies, psychiatric disorders and speech and motor delay			

#### Single Gene Disorder

The GeneSafe and PrenatalSafe Complete test also screen mutations in 29 genes that could be missed by traditional prenatal screening. These gene mutations cause life altering genetic disorders such as cystic fibrosis, sickle cell anaemia, B-thalasemia, skeletal dysplasias, cardiac defects, multiple congenital anomalies, autism, epilepsy, and/or intellectual disability among 49 others. This test is highly suitable for screening of genetic disorders associated with advanced paternal age. This is a paradigm shift in prenatal screening.

#### **Evolution of non-invasive parental screening**

# Our NIPT screening test is intended for patients who meet any of the following criteria.



## Why choose Eurofins?

While the other test provider screens only T13, T18, T21 and sex chromosome abnormalities, we offer a comprehensive approach covering the whole genome to also detect the genetic abnormalities prevalent in the remaining 17% cases\*.

Moreover, we use market leading NGS technology from Illumina. Our test failure rate is lowest (0.3%) compared to other NIPT providers.



\*Wellesley D, et al: Eur J Hum Genet. 2012; 20:521-526



### Next steps...

To find out more, Call: 0808 1691 022 Email: NIPT@eurofins.co.uk

#### Alternatively contact your local healthcare provider:

#### About Eurofins clinical diagnostics

Eurofins clinical diagnostics is fully focused on excellence, innovation and bringing technologically advanced genetic tests to every patient, wherever he or she lives. With a network of highly specialised diagnostic laboratories in the U.K., Italy, France, and Germany we provide the best possible diagnostic services. Our accredited laboratories are equipped with state-of-the-art instruments and technologies to process voluminous samples every day. We have helped >200,000 future parents by providing their foetal genetic conditions so they can feel assured. We pride ourselves in the timely delivery of results through efficient logistical arrangements and electronic result reporting. Every test is performed to the highest standard by our team of dedicated staff with extensive experience in prenatal diagnostics.



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