

# CONVENTIONAL PRENATAL TESTS



Cell-free DNA extracted from mother's blood and paternal DNA sample

Can detect several fetal genetic disorders



#### **HIGH ACCURACY**

> 99% detection rate for aneuploidies



#### SAFE

No risk of fetal miscarriage



#### EARLY

Can be done from the 10<sup>th</sup> week of pregnancy Screening combines biochemical results, ultrasound findings and other parameters

Most microdeletions and monogenic diseases do not have biochemical and ultrasound markers



LOW ACCURACY 80-95% detection rate for aneuploidies



of miscarriage through amniocentesis or CVS (0.5%)



Screening for aneuploidies after the 12<sup>th</sup> week of pregnancy

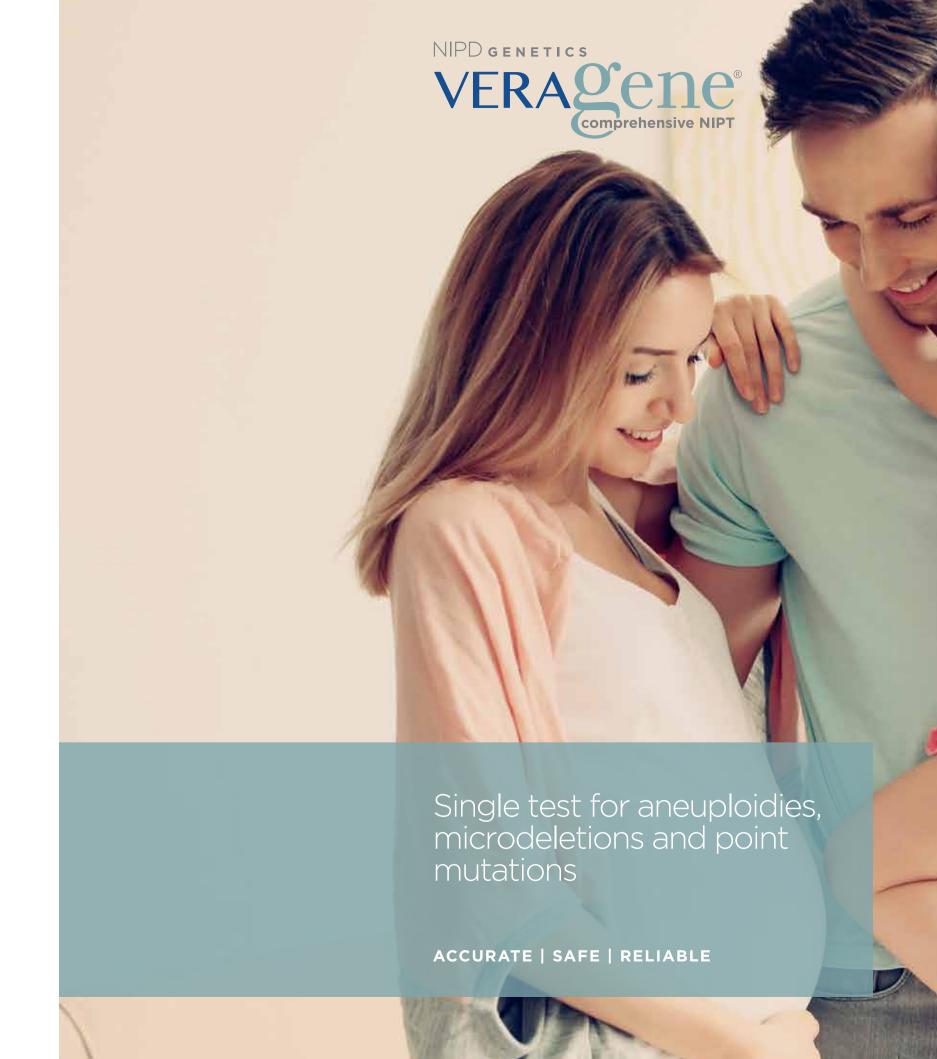








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## **VERAgene NIPT**

Can be done from the 10th week of pregnancy

**Single screening test** for aneuploidies, microdeletions and point mutations

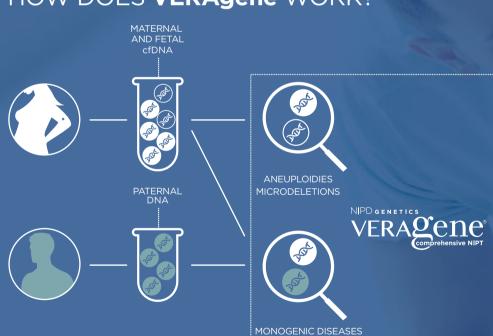
Validated for **singleton** and **twin** pregnancies

Applicable also for **IVF** pregnancies

## WHAT IS **VERAgene** NIPT?

VERAgene is the first comprehensive non-invasive prenatal test (NIPT) that can simultaneously screen for aneuploidies, microdeletions and point mutations. The diseases screened by VERAgene are often severe with significant impact on the quality of life. Besides aneuploidies and microdeletions, VERAgene targets 2000 mutations to screen for 100 monogenic diseases. By combining detection of aneuploidies and microdeletions with the screening of monogenic diseases, VERAgene provides a comprehensive picture of the pregnancy using a single test.

## HOW DOES **VERAgene** WORK?



VERAgene needs a maternal blood sample during pregnancy, and a buccal swab sample from the biological father. The maternal blood contains cell-free DNA (cfDNA) from both the mother and the fetus. The cfDNA is isolated and analyzed independently for aneuploidies and microdeletions, and concurrently with the paternal DNA sample for parental carrier status determination for monogenic diseases using our proprietary technology. Sophisticated algorithms are then used to compute the risk of the fetus having an aneuploidy, microdeletion or monogenic disease. The results are sent to the clinician who communicates them to the parents and provides the necessary counselling.



#### **AUTOSOMAL ANEUPLOIDIES**

Down syndrome (Trisomy 21)
Edwards syndrome (Trisomy 18)
Patau syndrome (Trisomy 13)

#### SEX CHROMOSOME ANEUPLOIDIES

Turner syndrome (Monosomy X)

Triple X syndrome (Trisomy X)

Klinefelter syndrome (XXY)

Jacobs syndrome (XYY)

XXYY syndrome

#### **MICRODELETIONS**

DiGeorge syndrome (22q11.2)
1p36 deletion syndrome (1p36)
Smith-Magenis syndrome (17p11.2)
Wolf-Hirschhorn syndrome (4p16.3)

#### MONOGENIC DISEASES

**Myotubular Myopathy** 

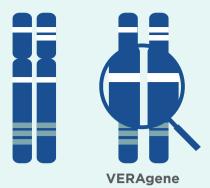
Panel of 100 autosomal and X-linked monogenic diseases, including:

**Cystic Fibrosis Alstrom syndrome** Sickle-Cell disease **Abetalipoproteinemia Beta Thalassemia Bardet-Biedl syndrome Tav-Sachs disease** Alport syndrome, X-linked Gaucher disease **Pendred syndrome** Phenylketonuria **Familial Dysautonomia Autosomal Recessive** Joubert syndrome, Type 2 **Polycystic Kidney disease** Isovaleric Acidemia Canavan disease Glutaric Acidemia. Type 2A Fanconi Anemia, Type C Maple Syrup Urine disease, Usher syndrome, Type 1F Type 1B

The hereditary monogenic diseases screened by VERAgene are associated with moderate to severe phenotypes, including hematological, renal, ophthalmological, cardiac, endocrine, respiratory, neurological, muscular and metabolic diseases. Inherited metabolic diseases also include many inborn errors of metabolism.

**Factor XI Deficiency** 

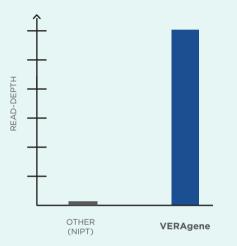
For a complete list of the monogenic diseases screened by VERAgene please visit *www.nipd.com* 



# UNIQUE FEATURES OF VERAgene

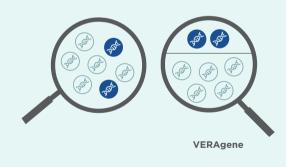
#### TARGETED GENOMIC ANALYSIS

VERAgene uses proprietary technology, specifically designed to avoid genomic regions with complex architecture that affect test performance. This overcomes problems associated with other NIPTs and increases the **precision** and **accuracy** of VERAgene.



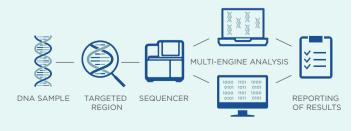
#### HIGH READ-DEPTH

Read-depth is the number of times a nucleotide in the genome is read during analysis. VERAgene captures DNA fragments from targeted regions on chromosomes of interest. VERAgene is able to analyze these selected regions at an extremely high read-depth which improves the statistical accuracy of the analysis and increases the **sensitivity** and **specificity** of VERAgene.



### FETAL FRACTION MEASUREMENT

A proprietary bioinformatics software accurately calculates fetal fraction. Accurate fetal fraction measurement increases the **robustness** and **reliability** of VERAgene.



### MULTI-ENGINE ANALYSIS PIPELINES

Proprietary bionformatics pipelines analyze the sequencing data produced from each test. This multi-engine analysis increases the **sensitivity** and **specificity** of aneuploidy, microdeletion and fetal gender detection.