NIPD GENETICS
VERAgene™ comprehensive NIPT

VS

CONVENTIONAL PRENATAL TESTS

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

Screening combines biochemical results, ultrasound findings and other parameters

Can detect several fetal genetic disorders

Biochemical and ultrasound markers don't exist for microdeletions and monogenic diseases

HIGHLY ACCURATE
> 99% detection rate for aneuploidies

LOW ACCURACY
80-95% detection rate for aneuploidies

SAFE
No risk of fetal miscarriage

RISK
of miscarriage through amniocentesis or CVS (0.5%)

FAST
Can be done from 10 weeks of pregnancy

Screening for aneuploidies after the 12th week of pregnancy

NIPD GENETICS

CAP ACCREDITED

ISO 13485:2016

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NIPD GENETICS
VERAgene™ comprehensive NIPT

Single test for aneuploidies, microdeletions and point mutations

ACCURATE | SAFE | RELIABLE

VERAgene NIPT

Can be done from **10 weeks of pregnancy**

Single screening test for aneuploidies, microdeletions and point mutations

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

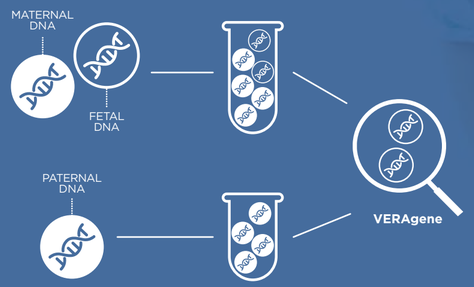
Applicable also for **IVF** pregnancies

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. VERAgene targets 500 mutations to screen for 50 monogenic disorders.
By combining detection of aneuploidies and microdeletions with the screening of monogenic disorders, VERAgene provides a comprehensive picture of the pregnancy using a single test.

HOW DOES **VERAgene** WORK?

VERAgene needs a maternal blood sample, and a buccal swab sample from the biological father. The maternal blood contains cell-free DNA (cfDNA) from both the mother and the fetus. This cfDNA is isolated and analyzed along with the father's DNA sample for any potential genetic mutations using next generation sequencing. Sophisticated bioinformatics algorithms are then used to compute the risk of the fetus having a monogenic disease.
The results are sent to the clinician who communicates them to the parents and provides the necessary counseling.

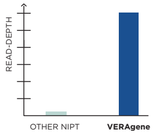


UNIQUE FEATURES OF **VERAgene**

VERAgene captures, counts and analyses cfDNA fragments from selected genomic regions using targeted enrichment and next generation sequencing (NGS) with proprietary genetic and analytical tools.
The main features of VERAgene are:

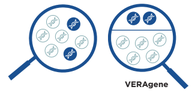
TARGETED GENOMIC ANALYSIS

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.



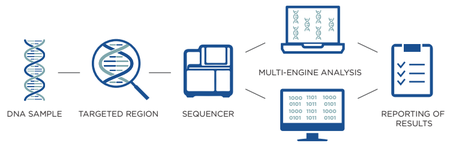
HIGH READ-DEPTH

These fragments are then counted several hundreds of times using NGS to achieve very high statistical accuracy and precision.



ACCURATE FETAL FRACTION

VERAgene uses the high read-depth of maternal and fetal DNA counts from the genome to accurately measure the fetal contribution to the cfDNA. Accurate fetal fraction measurement protects from false results.



MULTI-ENGINE ANALYSIS PIPELINES

Proprietary bioinformatics pipelines analyze the sequencing data produced from each test. This multi-engine analysis increases the sensitivity and specificity of detecting aneuploidies, microdeletions and monogenic diseases, along with fetal gender.

WHAT DOES **VERAgene** SCREEN FOR?

Aneuploidies

Condition	Impact	Cause
Down syndrome (Trisomy 21)	severe	Three copies of chromosome 21
Edwards syndrome (Trisomy 18)	very severe	Three copies of chromosome 18
Patau syndrome (Trisomy 13)	very severe	Three copies of chromosome 13
Turner syndrome (Monosomy X)	moderate	One copy of chromosome X
Triple X syndrome (Trisomy X)	mild	Three copies of chromosome X
Klinefelter syndrome (XXY)	mild	Extra copy of chromosome X
Jacobs syndrome (XYY)	mild	Extra copy of chromosome Y
XXYY syndrome	severe	Extra copies of chromosomes X and Y

Microdeletions

Condition	Impact	Cause
DiGeorge syndrome (22q11.2)	severe	Deletion of part of chromosome 22
1p36 deletion syndrome	severe	Deletion of part of chromosome 1
Smith-Magenis syndrome (17p11.2)	severe	Deletion of part of chromosome 17
Wolf-Hirschhorn syndrome (4p16.3)	severe	Deletion of part of chromosome 4

Monogenic diseases

Condition	Impact	Gene (Mutations)
3 Methylcrotonyl CoA Carboxylase Deficiency 1	severe	MCCC1 (2)
3 Methylcrotonyl CoA Carboxylase Deficiency 2	severe	MCCC2 (8)
Abetalipoproteinemia	severe (moderate)	MTTP (1)
Arthrogryposis Mental Retardation Seizures	severe	SLC35A3 (1)
Autosomal recessive polycystic kidney disease	severe	PKHD1 (30)
Bardet Biedl syndrome 12	severe (blindness)	BBS12 (4)
Beta thalassemia	very severe	HBB (88)
Canavan disease	severe	ASPA (4)
Choreacanthocytosis	moderate	VPS13A (1)
Crigler Najjar syndrome, Type I	very severe	UGT1A1 (10)
Cystic fibrosis	very severe	CFTR (122)
Factor V Leiden thrombophilia	moderate	F5 (1)
Factor XI deficiency	severe	F11 (4)
Familial dysautonomia	moderate	IKBKAP (3)
Familial Mediterranean fever	moderate	MEFV (8)
Fanconi anemia (FANCG-related)	severe	FANCG (3)
Glycine encephalopathy (GLDC-related)	very severe	GLDC (2)
Glycogen storage disease, Type 3	severe	AGL (14)
Glycogen storage disease, Type 7	severe	PFKM (3)
GRACILE Syndrome	very severe	BCS1L (12)
Inclusion body myopathy, Type 2	moderate	GNE (2)
Isovaleric acidemia	severe	IVD (1)
Joubert syndrome, Type 2	severe	TMEM216 (2)
Junctional epidermolysis bullosa, Herlitz type	severe	LAMC2 (1)
Leber congenital amaurosis (LCA5-related)	severe	LCA5 (3)
Leydig cell hypoplasia [Luteinizing Hormone Resistance]	moderate	LHCGR (10)
Limb girdle muscular dystrophy, Type 2E	severe	SGCB (6)
Lipoamide Dehydrogenase Deficiency [Maple syrup urine disease, Type 3]	severe	DLD (7)
Lipoprotein lipase deficiency	moderate	LPL (1)
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	severe	HADHA (2)
Maple syrup urine disease, Type 1B	severe	BCKDHB (5)
Methylmalonic acidemia (MMAA-related)	very severe	MMAA (14)
Multiple sulfatase deficiency	very severe	SUMF1 (1)
Navajo neurohepatopathy [MPV17-related hepatocerebral mitochondrial DNA depletion syndrome]	severe	MPV17 (1)
Neuronal ceroid lipofuscinosis (MFSD8-related)	very severe	MFSD8 (2)
Nijmegen breakage syndrome	severe	NBN (1)
Ornithine translocase deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome]	severe	SLC25A15 (3)
Peroxisome biogenesis disorders Zellweger syndrome spectrum (PEX1-related)	severe	PEX1 (3)
Peroxisome biogenesis disorders Zellweger syndrome spectrum (PEX2-related)	severe	PEX2 (1)
Phenylketonuria	very severe	PAH (67)
Pontocerebellar hypoplasia, Type 2E	very severe	VPS53 (2)
Pycnodysostosis	severe	CTSK (2)
Pyruvate dehydrogenase deficiency (PDHB-related)	severe	PDHB (2)
Retinal Dystrophy (RLBP1-related) [Bothnia retinal dystrophy]	severe (blindness)	RLBP1 (1)
Retinitis pigmentosa (DHDDS-related)	severe (blindness)	DHDDS (1)
Sanfilippo syndrome, Type D [Mucopolysaccharidosis IIID]	severe	GNS (5)
Sickle-cell disease	very severe	HBB (15)
Sjögren-Larsson syndrome	severe	ALDH3A2 (2)
Tay-Sachs disease	very severe	HEXA (14)
Usher syndrome, Type 1F	moderate (hearing loss)	PCDH15 (2)