VERAgene®
comprehensive NIPT



Single test for aneuploidies, microdeletions and monogenic diseases

MEDICOVER GENETICS

# **VERAgene** NEW GENERATION NIPT

**VERAgene** is a next-generation, **non-invasive prenatal test** (NIPT) that analyzes cell-free foetal DNA to screen for foetal **aneuploidies** and clinically significant **microdeletions**. Maternal cell-free foetal DNA and paternal genomic DNA are then analyzed to determine the foetal risk of **100 inherited monogenic disorders**.

By integrating chromosomal screening with carrier-informed monogenic risk assessment in a single test, VERAgene delivers comprehensive and clinically meaningful results without adding complexity to the testing process. Suitable from the **9th week of pregnancy**, VERAgene can be offered to all expecting mothers, regardless of maternal age or risk classification.

# WHY RECOMMEND **VERAgene**?

VERAgene complements conventional prenatal screening methods by extending the scope of genetic evaluation. Traditional approaches could identify structural or biochemical markers but cannot assess inherited or subchromosomal abnormalities. Inherited single-gene disorders occur across all pregnancies regardless of maternal age or family history, so incorporating monogenic disease screening into the same test provides clinically relevant information that conventional screening cannot detect.

By incorporating VERAgene into early prenatal care, clinicians can provide:

- Earlier and broader genetic risk assessment across chromosomal, microdeletion, and monogenic conditions
- Fewer unnecessary invasive procedures, due to its high specificity and focused reporting
- Clear results to support confident counseling and evidence-based decision-making

## Clinical Utility

According to professional societies such as ACMG, ACOG and ISPD, NIPT is the most accurate screening test for the detection of common foetal aneuploidies[1,2,3]. The use of NIPT in combination with conventional prenatal screening tests provides thorough evaluation of the pregnancy and improves prenatal care.

NIPT can reduce the number of invasive diagnostic procedures for common foetal autosomal aneuploidies and can also increase the prenatal detection rate for **sex chromosome aneuploidies** (SCAs) and **microdeletions** which:

- are not associated with maternal age
- often do not have ultrasound or biomarker findings

ACMG further highlights the importance of carrier screening and monogenic risk evaluation as part of comprehensive reproductive genetic care. VERAgene aligns with this guidance by integrating both chromosomal aneuploidies and monogenic disease screening into a single, clinically focused test. The disorders screened by VERAgene are often severe with significant impact on the quality of life.


*1. Dungan, Jeffrey S., et al. "Noninvasive Prenatal Screening (NIPS) for Fetal Chromosome Abnormalities in a General-Risk Population: An Evidence-Based Clinical Guideline of the American College of Medical Genetics and Genomics (ACMG)." Genetics in Medicine, vol. 25, no. 2, 2023, p. 100336*

*2. ACOG Committee Practice on Bulletins. "Screening for Fetal Chromosomal Abnormalities." Obstetrics & Gynecology, vol. 136, no. 4, 2020, pp. 859–867*

*3. Hui, Lisa, et al. "Position Statement from the International Society for Prenatal Diagnosis on the Use of Non-Invasive Prenatal Testing for the Detection of Fetal Chromosomal Conditions in Singleton Pregnancies." Prenatal Diagnosis, vol. 43, no. 7, 16 May 2023, pp. 814–828*

# **VERAgene** PANEL

**VERAgene** is available in a **single panel**, delivering high-confidence results across trisomies, clinically significant microdeletions, and inherited monogenic disease risk.

*Reporting of presence/absence of Y chromosome*

### Trisomies

Screens for the most common chromosomal abnormalities associated with significant clinical outcomes.

**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*)
**Jacob's syndrome** (*XYY*)
**XXYY syndrome**

### Microdeletion Syndromes

Screens for the most prevalent microdeletion syndromes of clinical significance.

**DiGeorge syndrome** (*22q11.2*)
**1p36 deletion syndrome** (*1p36*)
**Wolf-Hirschhorn syndrome** (*4p16.3*)
**Smith-Magenis syndrome** (*17p11.2*)
**Cri-du-chat syndrome*** (*5p deletion*)
**Prader–Willi syndrome*** (*15q11–q13 deletion*)
**Angelman syndrome*** (*15q11–q13 deletion*)

**These enhancements are currently implemented at Medicover Genetics' laboratories in Cyprus and will be progressively introduced across additional laboratories within the Medicover Genetics network.*

### 100 Monogenic Diseases

VERAgene targets over 2000 point mutations related to 100 autosomal recessive and X-linked monogenic diseases of hematological, renal, ophthalmological, cardiac, endocrine, neurological, muscular and metabolic nature.

*For a complete list of the monogenic diseases screened by VERAgene, please visit* www.medicover-genetics.com

# WHO IS **VERAgene** APPLICABLE FOR?

| | Autosomal Aneuploidies | Sex Chromosome Aneuploidies | Microdeletions | Monogenic Diseases |
|---|---|---|---|---|
| Singleton | ● | ● | ● | ● |
| Twin/Vanishing Twin | ● | | ● | ● |
| IVF (Self-egg used) Singleton | ● | ● | ● | ● |
| IVF (Self-egg used) Twin/Vanishing Twin | ● | | ● | ● |

# Clinical Results that Matter

## **TARGETED** METHODOLOGY

Unlike whole-genome sequencing, **VERAgene's** targeted methodology reduces background noise and analytical artifacts and focuses only on genomic regions of proven medical significance. This approach supports high sensitivity and specificity for both common aneuploidies and clinically significant microdeletions for reliable results. **VERAgene** uses novel Targeted Capture Enrichment Technology that enables aneuploidy detection as well as foetal fraction measurement with unparalleled **accuracy** and **reliability**. For monogenic conditions, **VERAgene** concurrently analyzes maternal cfDNA and paternal genomic DNA to calculate the foetus's risk based on established inheritance models. This trio-based design minimizes false positives and screens for variants associated with 100 autosomal **recessive and X-linked disorders** with demonstrated clinical significance.

# TEST **PERFORMANCE**

**VERAgene's** performance has been validated through peer-reviewed and internal studies demonstrating **high sensitivity**, **specificity**, and **reproducibility** for common trisomies, sex chromosome aneuploidies, and clinically significant microdeletions. Its analytical process maintains robust performance even in low foetal fraction samples (≥3%).

## Autosomal Aneuploidies

| KARYOTYPE | NO. | FOLLOW-UP | CORRECT CALLS | SPECIFICITY | NPV |
|---|---|---|---|---|---|
| NORMAL | 10280 | 10280 | 10280 | 99.98% (95% CI, 99.93 - 99.998%) | 100% (95% CI, 99.96 - 100%) |
| **KARYOTYPE** | **NO.** | **FOLLOW-UP** | **CORRECT CALLS** | **SENSITIVITY** | **PPV** |
| Trisomy 21 | 126 | 44 | 44 | 100% (95% CI, 92 - 100%) | 100% (95% CI, 92 - 100%) |
| Trisomy 18 | 24 | 10 | 10 | 100% (95% CI, 69 - 100%) | 100% (95% CI, 69 - 100%) |
| Trisomy 13 | 16 | 7 | 5 | 100% (95% CI, 48 - 100%) | 71% (95% CI, 29 - 96%) |

## Sex Chromosome Aneuploidies

| KARYOTYPE | NO. | FOLLOW-UP | CORRECT CALLS | SPECIFICITY | NPV |
|---|---|---|---|---|---|
| NORMAL | 6200 | 6200 | 6200 | 99.95% (95% CI, 99.86 - 99.99%) | 100% (95% CI, 99.94 - 100%) |
| **KARYOTYPE** | **NO.** | **FOLLOW-UP** | **CORRECT CALLS** | **SENSITIVITY** | **PPV** |
| 45, X | 16 | 7 | 4 | 100% (95% CI, 40 - 100%) | 57% (95% CI, 18 - 90%) |
| 47, XXX | 6 | 2 | 2 | - | - |
| 47, XXY | 10 | 4 | 4 | - | - |
| 47, XYY | 3 | 0 | - | - | - |
| 48, XXYY | 1 | 1 | 1 | - | - |

*Kypri et al. "Non-invasive Prenatal Testing of Fetal Chromosomal Aneuploidies: Validation and Clinical Performance of the Veracity Test." Molecular Cytogenetics vol. 12 34. 15 Jul. 2019*

## Microdeletions

| | SENSITIVITY | SPECIFICITY |
|---|---|---|
| DiGeorge syndrome (22q11.2)*† | 100% (95% CI: 73.5-100%) | 100% (95% CI: 99.5-100%) |
| Wolf-Hirschhorn syndrome (4p16.3)*† | 100% (95% CI: 39.8-100%) | 100% (95% CI: 99.5-100%) |
| Smith-Magenis syndrome (17p11.2)*† | 100% (95% CI: 69.2-100%) | 100% (95% CI: 99.5-100%) |
| 1p36 deletion syndrome*† | 100% (95% CI: 29.2-100%) | 100% (95% CI: 99.5-100%) |
| Prader-Willi and Angelman syndromes (15q)* | 91% (95% CI: 72-99%) | 100% (95% CI: 99.3-100%) |
| Cri-du-Chat syndrome (5p)* | 92% (95% CI: 64-99.8%) | 100% (95% CI: 99.3-100%) |

**Internal validation was conducted using spiked samples*

*†Internal validation was conducted with clinical samples*

# BENEFITS OF **VERAgene**

**ACCURATE**
>99% accuracy*

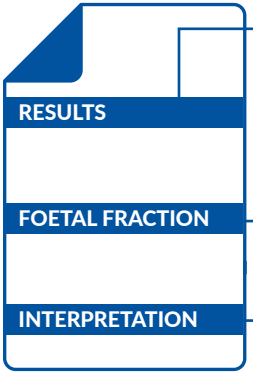
**SAFE**
Non-invasive procedure

**EASY**
Blood sample from pregnant woman and buccal swab from the biological father

**FAST**
Results in 4-7 working days from sample receipt

**The percentage refers to the detection of aberrations covered by VERAgene.*

# WHAT WILL THE **REPORT** TELL ME?




**VERY LOW RISK**
Reduced likelihood of the foetus having one of the tested conditions

**VERY HIGH RISK**
Increased likelihood of the foetus having one of the tested conditions

*As VERAgene NIPT is a screening test, high-risk results should always be confirmed with a diagnostic test.*

According to the ACMG 2016 Position Statement, foetal fraction is accurately measured and reported for every sample using validated bioinformatic analysis (*Gregg et al. 2016)*

Concise, standardised explanation of findings and clear recommendations for appropriate clinical follow-up or confirmatory testing when indicated

*A negative result does not eliminate the possibility of other genetic conditions, nor does it guarantee a healthy baby.*

## What can I do after VERAgene?

- Inform your patient about the result
- Recommend genetic counselling, if applicable
- Confirm very high risk results with a prenatal diagnostic procedure
- Discuss next steps and management
- Genetic counselling can be offered to doctors or patients upon request

*Compliance Disclaimer*
*VERAgene NIPT is a screening test, not a diagnostic test.*
*A high-risk result should always be confirmed by invasive diagnostic testing such as amniocentesis before any clinical decision is made.*
*VERAgene is validated for singleton, twin, and vanishing twin pregnancies (including IVF using self egg), but it is not suitable for patients with a history of malignancy, recent transfusion, or transplant, or for pregnancies achieved through donor eggs or surrogacy.*
*Availability and test configurations may vary by region and local legislation.*
*© Medicover Genetics Ltd. All rights reserved. Reproduction or modification of this content without authorization is prohibited.*

# HOW TO ADMINISTER THE **VERAgene** TEST?

Recommend **VERAgene** to the expectant parents

Collect a blood sample from the pregnant woman and a buccal swab from the biological father



Send the samples to **Medicover Genetics**

The samples will be analysed at **Medicover Genetics** laboratories

Results will be available within 4-7 working days from sample receipt, through a secure digital portal

## MORE **QUESTIONS**?

If you have additional questions or concerns, please contact us at **info.genetics@medicover.com**

MEDICOVER
GENETICS

Medicover Genetics Ltd
**www.medicover-genetics.com**

MKT-PRE-VG-DR-EN-V5