VERAgene FAQs

What is NIPT?

NIPT is a non-invasive prenatal test which analyzes cell-free DNA in the mother's blood to identify genetic conditions such as trisomies and sex chromosome aneuploidies in the fetus.

What are genetic conditions?

The human genome is sorted into twenty-three pairs of chromosomes. Genetic conditions are caused by unwanted changes in the genome that happen during conception. There are three types of genetic conditions:

- aneuploidies are genetic conditions that occur when a chromosome has an extra copy (trisomy) or is missing a copy (monosomy)
- microdeletions are genetic conditions caused by the deletion of part of a chromosome
- single gene diseases are genetic conditions caused by pathogenic mutations (genetic changes) in a gene.

What is VERAgene comprehensive NIPT?

VERAgene is the only non-invasive prenatal test that can simultaneously screen for aneuploidies, microdeletions and single gene diseases. The diseases screened by VERAgene are associated with moderate to severe phenotype with significant impact on the quality of life. These genetic diseases exhibit symptoms such as congenital anomalies, development delays, hearing loss, blindness, metabolic diseases, etc.

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 from both the mother and the fetus. This cell-free DNA is isolated and analyzed for aneuploidies and microdeletions. Additionally, the maternal DNA along with the father's DNA sample is analyzed for any potential genetic mutations, to compute the risk for monogenic disorders.

What genetic conditions can be detected by VERAgene?

VERAgene can screen for trisomies such as Down syndrome, Edwards syndrome, Patau syndrome, sex chromosome aneuploidies, microdeletions such as DiGeorge syndrome, 1p36 deletion syndrome, Smith-Magenis syndrome and Wolf-Hirschhorn syndrome, and 100 monogenic diseases with autosomal recessive and X-linked inheritance. These include Beta Thalassemia, Cystic Fibrosis, Tay-Sachs disease, Canavan disease, Sickle Cell disease, Phenylketonuria, Alport syndrome and many more. 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.

What are Down syndrome, Edwards syndrome and Patau syndrome?

Down syndrome, Edwards syndrome and Patau syndrome are genetic disorders caused by chromosomal trisomies. Down syndrome is caused by an extra copy of chromosome 21 whereas Edwards syndrome and Patau syndrome are caused by an extra copy of chromosome 18 and chromosome 13, respectively. Down syndrome is characterized by intellectual impairment and congenital abnormalities. Down syndrome occurs approximately in 1 in 700 pregnancies but is more frequent in pregnancies in women over 35. Edwards syndrome and Patau syndrome are less common occurring in 1 in 5000 and 1 in 16000 pregnancies, respectively. Edwards and Patau syndromes are characterized by severe congenital abnormalities and rarely survive past the first year of life.

What are monogenic diseases?

Monogenic diseases are caused by a mutation in a single gene (single gene diseases). Such conditions can be autosomal dominant which are caused when a mutation exists on only one chromosome, or autosomal recessive where a mutation needs to be present in both chromosomes. There are also X-linked diseases, where the mutation is always on the X chromosome and affects males and females differently. VERAgene analyses over 2000 variants to detect the risk for 100 autosomal recessive and X-linked monogenic diseases.

What are sex chromosome aneuploidies?

Sex chromosome aneuploidies are genetic disorders caused by the presence or absence of a sex chromosome. The 23rd pair of chromosomes determine the gender of an individual. Women have two X chromosomes and men have one X and one Y chromosome. VERACITY tests for the following sex chromosome aneuploidies:

- Turner syndrome, characterized by the presence of a single X chromosome.
- Triple X syndrome, characterized by the presence of three X chromosomes.
- Klinefelter syndrome, characterized by the presence of two X chromosomes and one Y chromosome.
- Jacob syndrome, characterized by the presence of one X chromosome and two Y chromosomes.
- XXYY syndrome, characterized by the presence of two X chromosomes and two Y chromosomes.

What are microdeletions?

Microdeletions are genetic conditions caused by the loss of a part of a chromosome. Microdeletions are characterized by congenital abnormalities and intellectual impairment. The severity of the symptoms varies depending on the size and location of the microdeletion. The most common microdeletion syndrome is DiGeorge syndrome which occurs approximately once in 1000 pregnancies. The prevalence of DiGeorge syndrome increases to 1 in 100 in pregnancies with major structural anomalies such as congenital heart disease.

What are autosomal diseases?

Autosomal diseases are conditions that affect chromosome pairs 1 to 22, the 'autosomal' chromosomes. These genetic conditions can either be autosomal dominant or autosomal recessive.

What are X-linked diseases?

X-linked diseases are those that affect the X chromosome on the 23rd chromosome pair, which determines gender. Generally, they are not as severe as autosomal diseases, but they may affect quality of life as common symptoms include fertility problems and cognitive difficulties. X-linked diseases are most often passed from affected or carrier mothers to their sons, as fathers pass the Y chromosome on all their male offspring.

Why do X-linked diseases affect males and females differently?

As males have one X and one Y chromosome, when their X chromosome has a mutation the disease always manifests. Females have two X chromosomes, and when one of their X chromosomes has a mutation they are carriers of an X- linked disease. They may exhibit some symptoms of the disease or no symptoms at all, due to 'X-inactivation' – a mechanism that always 'shuts off' one of the two X chromosomes in females so they don't have twice the number of genes as males, which would be toxic. As this process is random in the female cells, the X chromosome with the mutation can be silenced in varying degree in female carriers; thus explaining the symptom variability shown.

Why should expecting parents consider VERAgene?

Microdeletions and single gene diseases – unlike aneuploidies – do not have a maternal age-associated risk, and most of them do not have chemical or ultrasound biomarkers that can help in early detection. The cumulative risk for the fetus to have either an aneuploidy, microdeletion or a monogenic condition screened by VERAgene is 1 in 50 in moderate to high-risk pregnancies. VERAgene can accurately screen for these conditions to help parents take informed decisions about possible treatment and clinical management.