| | |
|---|---|
| **Name** | **Sample ID** |
| **Date of birth** | **Sample collected** |
| **Sample type** | **Sample received** |
| **Provider** | **Report date** |

| CLINICAL INFORMATION | | | |
|---|---|---|---|
| **Gestational age:** 10 + 1 | **Pregnancy type:** Singleton | **IVF pregnancy:** No | **Oocytes:** |
| **Clinical indication:** | | | |

myPrenatal — Fetal fraction: 12%

| CHROMOSOMES TESTED | RESULTS CONSISTENT WITH | INTERPRETATION |
|---|---|---|
| **Chromosomes 21, 18, 13** | Absence of aneuploidy | **Low risk of aneuploidy**<br>Regular pregnancy follow-up |
| **Fetal sex** | Absence of Y Chromosome | Female fetus |
| **Sex chromosomes** | Absence of aneuploidy | **Low risk of aneuploidy**<br>Regular pregnancy follow-up |
| **Other chromosomes** | Absence of aneuploidy | **Low risk of aneuploidy**<br>Regular pregnancy follow-up |
| **Deletions and duplications (CNVs) > 7Mb** | Presence of del(14)(q22.1q22.3) | **High risk of partial aneuploidy**<br>Genetic counseling is recommended to evaluate confirmation of the result (see clinical note below) |

## CLINICAL NOTE

The result is consistent with the presence of a deletion in 14q22.1-q22.3 region which includes at least 20 genes and is associated with 14q Deletion Syndrome (Frias Syndrome). Phenotypic features include, among others, structural and facial abnormalities and growth delay. Genetic counseling is highly recommended to evaluate the options available for diagnostic confirmation of this result.

## ANALYSIS METHOD

Cell-free DNA extraction from plasma, paired-end Next Generation Sequencing (NGS) and bioinformatic analysis to determine the risk of aneuploidy as requested (VeriSeq NIPT v2, Illumina Inc. CE-IVD).

## LIMITATIONS AND PERFORMANCE

myPrenatal is a test designed to screen for common fetal trisomies (chromosomes 21, 18 and 13), as also for rare aneuploidies and partial imbalances (CNVs) of at least 7 megabases (Mb) in all the autosomes. In singleton pregnancies it is possible to extend the analysis to sex chromosomes aneuploidies and fetal sex determination; in twin pregnancies only the presence or absence of the Y chromosome can be evaluated. The test is validated for singleton or twin pregnancies of at least 10 weeks of gestation.

Test results might not reflect the true fetal chromosome constitution as false positives or false negatives could occur. Among the main known causes of discrepancies, the most frequent are due to biological factors included but not limited to the presence of a vanishing twin, fetal mosaicism of low proportion, confined placental mosaicism or unrecognized maternal chromosomal abnormalities. In pregnancies with a non-evolving co twin (or vanishing twin), the result could reflect its chromosome constitution.

myPrenatal has not been designed to determine the risk of other genetic disorders such as chromosomal mosaicisms, triploidy, partial aneuploidies of less than 7Mb and single gene defects so that a low risk result does not guarantee a healthy pregnancy or fetus and does not eliminate the presence of other genetic or structural alterations such as neural tube defects.

Some rare chromosomal aneuploidies can only occur in mosaic form. The clinical consequences depend on the chromosome involved and may not be elucidated prenatally.

Performance data:

| | Trisomy 21 | Trisomy 18 | Trisomy 13 | Other aneuploidies | CNVs (>7Mb) |
|---|---|---|---|---|---|
| Sensitivity | >99% | >99% | >99% | 96.40% | 74.10% |
| Specificity | 99,90% | 99,90% | 99,90% | 99.80% | 99.80% |

| Fetal sex results concordance | | |
|---|---|---|
| 100% | 90.5% | 91.7% |
| XX, XY, XXX, XXY | XO | XYY |

VeriSeq NIPT v2 CE-IVD Product insert (Singleton gestations excluding mosaicics).

## DISCLAIMER

Non-invasive prenatal screening based on the analysis of the cell-free DNA cannot be considered diagnostic tests. The results should be interpreted with all the clinical information available by the prescribing physician to provide appropriate advice on pregnancy management. No irreversible clinical decisions should be made based solely on these results. A result consistent with the presence of a chromosomal abnormality should always be confirmed by a prenatal diagnostic test performed on fetal samples obtained by an invasive procedure (i.e. amniocentesis or chorionic villus sampling).