### Analysis Report: PrenatalSafe® Karyo Plus - Non-Invasive Prenatal Test (NIPT)

<table>
<thead>
<tr>
<th>Report date:</th>
<th>Time:</th>
</tr>
</thead>
</table>

#### Referring Centre details

**Referring Centre:**

**City:**

#### Patient’s details

<table>
<thead>
<tr>
<th>Surname:</th>
<th>Name:</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Date of birth:</th>
<th>Place of birth:</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Ethnicity:** N.A.

**Physician:**

**Sex:** F

**Sample’s ID:**

#### Sample’s details

**Sample Type:** blood

**Our Sample’s ID:**

**Acceptance Date:**

**Acceptance Time:**

**Collection Date:**

#### Analysis details

<table>
<thead>
<tr>
<th>Analysis performed:</th>
<th>PrenatalSafe® Karyo Plus - Non-Invasive Prenatal Test (NIPT)</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Code OMIM:</th>
<th>Mode of Inheritance:</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Gene investigated:</th>
<th>OMIM:</th>
<th>Reference Sequence:</th>
</tr>
</thead>
</table>

<table>
<thead>
<tr>
<th>Method of Analysis:</th>
<th>Massive Parallel Sequencing (MPS) - Next Generation Sequencing (NGS)</th>
</tr>
</thead>
</table>

**Diagnostic strategy:**

**Sample Processing Date:**

**Analysis completed:**
**Analysis Results**

**Result:**  
**POSITIVE Test Result.**  
Results consistent with loss of chromosome 5 (5p15.33-p13.2) material.  
Genome-wide analysis did not detect gains or losses of chromosomal material suggestive of whole chromosome aneuploidies, subchromosomal duplications or deletions ≥7 Mb in other chromosomes.  
**Fetal sex** consistent with **Female.**  
**Fetal Fraction:** 7.3%

**Interpretation:**  
A **loss of chromosome 5 material** was observed. It is estimated to be **34 Mb** in size and is suggestive of a **deletion in the region 5p15.33-p13.2.** This region may contain one or more clinically significant genes. Genetic counseling and clinical correlation are recommended. Confirmatory testing is required if fetal confirmation and clinical interpretation of the suspected event are desired. Please refer to the “Performance” and “Limitations of the Test” sections of the enclosed technical report for additional information.

**Technical notes:**  
PrenatalSafe® Karyo Plus test is designed to detect common chromosome aneuploidies as well as gains or losses in every chromosome of the fetal genome, providing karyotype-level insight. It also detect subchromosomal duplications or deletions ≥7 Mb, and 9 common microdeletion syndromes: Prader-Willi syndrome, 5p-/Cri du Chat syndrome, 22q11.2 deletion syndrome, 1p36 deletion syndrome, or 4p-/Wolf-Hirschhorn syndrome, 8q24/Langer-Giedion syndrome, 11q23/ Jacobsen syndrome and 17p11.2/Smith-Magenis syndrome. The PrenatalSafe® Plus test is performed by a directed analysis of cell-free fetal DNA (cfDNA) in maternal blood by Massively Parallel Sequencing (MPS) of the whole fetal genome, using Next Generation Sequencing (NGS) technology. Bioinformatic analysis has been performed as reported elsewhere (Bayindir et al., Eur J Hum Genet 2015; 23:1286-1293). The test is validated for singleton pregnancies with gestational age of at least 10 weeks. The test is neither intended nor validated for diagnosis or for use in pregnancies with more than two fetuses, mosaicism, partial chromosomal aneuploidy, translocations, or maternal aneuploidy. These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal or subchromosomal abnormalities, birth defects and other conditions. This test is not intended to identify pregnancies at risk for open neural tube defects. A negative test result does not eliminate the possibility of chromosomal abnormalities. In addition, conditions caused by other molecular mechanisms cannot be detected with this assay. When an aneuploidy detected result is reported in a twin pregnancy, the status of each individual fetus cannot be determined. Although the presence or absence of Y chromosome material can be reported in a twin pregnancy, the occurrence of sex chromosome aneuploidies such as Monosomy X, XXX, XXY, and XYY cannot be evaluated in twin pregnancies. There is a small possibility that the test results might not reflect the chromosome status of the fetus, but may reflect chromosomal changes of the placenta (confined placental mosaicism) or of the mother (chromosomal mosaicism). Limit of Detection (LOD) of the method: fetal fraction greater than or equal at 2% (Fiorentino et al., 2016, Prenatal Diagnosis).

**Comments:**

**Further action:**

**Results verified by:**

**Verification date:**

**Results validated by:**

**Validation date:**

This report represents a true copy to the primary document, that is detained in the archives of Genoma Group Srl.

**Medical Geneticist**  
Dr.ssa Marina Baldi

**Lab Director**  
Dr. Francesco Fiorentino

**Rome, 21 June 2016**  
Genoma Group Srl