



**Analysis Report : PrenatalScreen® Prenatal Test**

**Report date:** 11/07/2016

**Time:** 13:37

**Referring Centre details**

**Referring Centre:**

**City:**

**Patient's details**

**Surname:**

**Name:**

**Date of birth:**

**Place of birth:**

**Ethnicity:** N.A.

**Sex:** M

**Physician:**

**Sample's ID:**

**Indication:**

**Clinical details:**

**Sample's details**

**Sample Type:** Amniotic Fluid

**Our Sample's ID:** E17941

**Acceptance Date:** 16/06/2016 **Acceptance Time:** 17:58 **Collection Date:** 16/06/2016

**Analysis details**

**Analysis performed:** PrenatalScreen® Prenatal Test

**Code OMIM:**

**Mode of Inheritance:**

**Gene investigated:**

**OMIM:**

**Reference Sequence:**

**Method of Analysis:** Next Generation Sequencing (NGS)

**Diagnostic strategy:**

**Sample Processing Date:** 17/06/2016

**Analysis completed:** 11/07/2016



## Results Summary

**Result:** **NEGATIVE Test Result.**  
No mutations have been detected in the genes under investigation.

### Interpretation:

**Technical notes:** PrenatalScreen® is a diagnostic test that analyses fetal DNA, obtained from CVS or amniotic fluid following an invasive prenatal diagnosis, to screen 744 genes for mutations causing over 1000 monogenic disorders in the fetus, including the most common in the European population. The fetal DNA, isolated from CVS or amniotic fluid, is amplified by PCR. Through massively parallel sequencing (MPS), which uses Next Generation Sequencing (NGS) techniques with ILLUMINA sequencing instruments, 744 genes are completely sequenced (exons and adjacent intronic regions,  $\pm 5$  nucleotides) (see technical report) at high read depth. The resulting genetic sequences are analyzed via an advanced bioinformatics analysis, to check for the presence of Known pathogenic or likely pathogenic mutations in the genes under investigation. Only mutations classified as "known or likely pathogenic", in accordance with the relevant scientific literature and the current classification in the ClinVar – NCBI, dbSNP – NCBI, and other NCBI resources, Human Gene Mutation Database (HGMD), updated on the date of the sample collection, will be reported.. Moreover, in compliance with the indications of the American College of Medical Genetics (ACMG), only mutations with a Minor Allele Frequency (MAF) <5% (1000 Genomes Project) are considered as pathogenic or possibly pathogenic; this measurement refers to the frequency in which the less common allele is present in the general population. Variations with a read depth (i.e. number of reads) lower than 30X are not highlighted by the bioinformatics analysis algorithm.

### Comments:

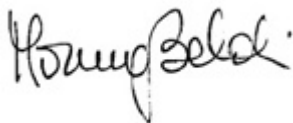
#### Further action:

|                              |                   |                           |            |
|------------------------------|-------------------|---------------------------|------------|
| <b>Results verified by:</b>  | Giuliano Cottone  | <b>Verification date:</b> | 11/07/2016 |
| <b>Results validated by:</b> | Andrea Nuccitelli | <b>Validation date:</b>   | 11/07/2016 |

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



Genoma Group Srl

Rome, 11 July 2016

#### Lab Director

Dr. Francesco Fiorentino



Genoma Group Srl