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

Sede legale
00138 Roma - Via di Castel Giubileo, 11
C.F. e P.Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Laboratori e Studi Medici Roma
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Laboratori e Studi Medici Milano
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

CHIAMATA GRATUITA
NUMEROVERDE
800-501651

GENOMA Molecular Genetics Laboratories Group

AZIENDA CON SISTEMA DI GESTIONE QUALITÀ CERTIFICATO DA DNV GL = ISO 9001 =

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, ± 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 ", iin 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:

Results verified by: Giuliano Cottone ***Verification date:*** 11/07/2016

Results validated by: Andrea Nuccitelli ***Validation date:*** 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

Lab Director
Dr. Francesco Fiorentino

Genoma Group Srl

Rome, 11 July 2016

Sede legale
00138 Roma - Via di Castel Giubileo, 11
C.F. e P.Iva: 05402921000
REA: 883.955
Iscr. Reg. Impr. 369761/1997

Laboratori e Studi Medici Roma
00138 Roma - Via di Castel Giubileo, 11
Tel. +39 06 881 1270 - Fax +39 06 6449 2025
Web: www.laboratoriogenoma.eu
E-mail: info@laboratoriogenoma.eu

Laboratori e Studi Medici Milano
20161 Milano - Affori Centre,Via Enrico Cialdini, 16
Tel. : + 39 02 3929 7626 - Fax : + 39 02 3929 76261
Web: www.genomamilano.it
E-mail info@genomamilano.it

CHIAMATA GRATUITA NUMEROVERDE 800-501651