GENOMA Group
Molecular Genetics Laboratories

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

Analysis Report : PrenatalScreen® Prenatal Test

Report date: 01/07/2016 ***Time:*** 18:21

Referring Centre details

Referring Centre:

City:

Patient's details

Surname: ***Name:***

Date of birth: ***Place of birth:***

Ethnicity: N.A. ***Sex:*** F

Physician: ***Sample's ID:***

Indication:

Clinical details:

Sample's details

Sample Type: Amniotic Fluid ***Our Sample's ID:*** E12790

Acceptance Date: 28/05/2016 ***Acceptance Time:*** 09:57 ***Collection Date:*** 27/05/2016

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: 30/05/2016 ***Analysis completed:*** 01/07/2016

Eurofins Genoma Group S.r.l a socio unico

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

Results Summary

Result:

- gene GJB2 (Deafness):
Heterozygote for mutation c.229 T>C (W77R) [rs104894397]
- gene HBB (Thalassaemia beta):
Heterozygote for mutation c.91+1 G>A (IVS1-1 G>A) [rs33971440]

Interpretation:

The fetus resulted **carrier** of the following mutations:

c. 229 T>C (W77R) of the gene GJB2;
Ref: Carrasquillo (1997) Hum Mol Genet 6, 2163

c. 91+1 G>A (IVS1-1 G>A) of the gene HBB.
Ref: Waye (2002) Hemoglobin 26, 87

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. highlighted by the bioinformatics analysis algorithm.

Comments:

Further action:

Carrier screening for the patient's partner is suggested. This screening can be performed at Genoma Group or another laboratory. The patient may wish to discuss these positive results with family members to allow them the option to be screened for this condition. Comprehensive genetic counseling is recommended to discuss the implications of these test results and possible associated reproductive risk. Patients who have questions about their results can contact Genoma and set up a phone consult with a Lab Genetic Counselor by calling +39 06 8811270 or emailing info@laboratoriogenoma.eu. If clinicians would like to discuss this patient's results with Genoma, please call +39 06 8811270 to be connected to the genetic counselor on call, or email info@laboratoriogenoma.eu.

Results verified by: Giuliano Cottone ***Verification date:*** 28/06/2016

Results validated by: Francesco Fiorentino ***Validation date:*** 01/07/2016

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, 01 July 2016

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