Analysis Report : GeneScreen® - Genetic Carrier Screening 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: blood ***Our Sample's ID:*** E12790

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

Analysis details

Analysis performed: GeneScreen® - Genetic Carrier Screening 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

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C.F. e P. Iva: 05402921000 - REA: 883.955 - Iscr. Reg. Impr. 369761/1997

Analysis Results

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 patient 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:

GeneScreen® is a diagnostic test which allows multiple carrier testing of more than 700 genetic diseases, including the most common in the European population. The DNA, isolated from the peripheral blood, is amplified by PCR. Through massively parallel sequencing (MPS), which uses Next Generation Sequencing (NGS) techniques with ILLUMINA sequencing instruments, 550 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 the presence of potential mutations in the genes under investigation. Only mutations classified as "known pathogenic outcome" in accordance with the relevant scientific literature and the current classification in the Human Gene Mutation Database (HGMD), updated on the date of the sample collection, are 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: **Genetic counseling is adviced**

Further action:

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

Lab Director
Dr. Francesco Fiorentino

Genoma Group Srl

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Rome, 01 July 2016

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