Analysis Report : GeneScreen® Easy - Genetic Carrier Screening Test

Report date: ***Time:***

Referring Centre details

Referring Centre:

City:

Patient's details

Surname: ***Name:*** .

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

Ethnicity: ***Sex:***

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

Indication:

Clinical details:

Sample's details

Sample Type: ***Our Sample's ID:***

Acceptance Date: ***Acceptance Time:*** ***Collection Date:***

Analysis details

Analysis performed: GeneScreen® Easy - Genetic Carrier Screening Test

Code OMIM: ***Mode of Inheritance:***

Gene investigated: ***OMIM:*** ***Reference Sequence:***

Method of Analysis:

Diagnostic strategy:

Sample Processing Date: ***Analysis completed:***

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REA: 883.955
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CHIAMATA GRATUITA NUMEROVERDE 800-501651

Analysis completed

Result:

Genetic variants identified:
- NM_001127701.1(SERPINA1): c.863 A>T (p.Glu288Val) heterozygote. [rs17580]
Genotype IVS8 polyT CFTR: 7T/7T
No mutations were detected in the other investigated genes.
(ClinVar updated Jun 06, 2019)

Interpretation:

The patient resulted **CARRIER** of the following mutation:
- c.863 A>T (p.Glu288Val) heterozygote in the SERPINA1 gene.
rs17580 ClinVar ncbi; Last evaluated: Nov 20, 2018; Last Updated: May 19, 2019. Clinical significance: Pathogenic; Ref.: Curiel (1989) J Biol Chem 264, 10477
No other pathogenic mutations were detected in the genes that were screened. A complete list of all conditions tested can be found in the enclosed technical report.
Risk to Child: Any child of this patient has a 50% chance of inheriting the patient's mutation(s) associated with this disease(s) and being a carrier. If the patient's partner also carries a mutation for this disease(s), there is a 25% chance that each child of the patient will inherit both parents' mutations and may develop the disease(s).
Risk to Patient: This patient is a carrier of a genetic mutation for this disease(s) but is not likely to be affected.

Technical notes:

GeneScreen® Easy is a diagnostic test which allows multiple carrier testing of more than 500 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, 330 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. 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. Gene dosage analysis by Multiplex Ligation-dependent Probe Amplification (MLPA) of the SMN1 gene was performed for SMA carrier screening. Fluorescent PCR was used for Fragile-X carrier screen, to detect the (CGG)n repeat expansions in the promoter region of the FMR-1 gene.

Comments:

Further action:

Comprehensive genetic counseling is recommended to discuss the implications of these test results. 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: ***Verification date:***

Results validated by: ***Validation date:***

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