GENOMA Molecular Genetics Laboratories Group

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

Analysis Report : GeneSafe™ Complete - non invasive prenatal screening for single-gene disorders

Report date: 14/08/2017 ***Time:*** 15:10

Referring Centre details

Referring Centre:

City:

Patient's details

Surname:		***Name:***	
Date of birth:	14/01/1981	***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:*** H48581

Acceptance Date: 09/08/2017 ***Acceptance Time:*** 12:25 ***Collection Date:*** 08/08/2017

Analysis details

Analysis performed: GeneSafe™ Complete - non invasive prenatal screening for single-gene disorders

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

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

Method of Analysis: DNA sequencing by Next Generation Sequencing (NGS) technology

Diagnostic strategy:

Sample Processing Date: 09/08/2017 ***Analysis completed:*** 14/08/2017

Eurofins Genoma Group S.r.l a socio unico

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CHIAMATA GRATUITA NUMEROVERDE 800-501651

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Result Summary

Result:

POSITIVE Test Result

Pathogenic mutations detected.

Heterozygous mutations **Cod.39 C>T (c.118C>T)(p.Q39X)** and **IVS1-1 G>A (c.93-1G>A)** detected in the **HBB gene.**

Fetal sex consistent with **Female.**

Fetal Fraction: 6%

Interpretation:

This result shows that the test detected the **heterozygous mutations Cod.39 C>T (c.118C>T)(p.Q39X)** and **IVS1-1 G>A** in the **HBB gene**, suggestive of **high risk** for **BetaThalassemia** syndrome. A patient with a positive test result should be referred for genetic counseling and should be followed-up with an invasive diagnostic test for confirmation of test results, before any medical decisions are made. Please refer to the “Performance” and “Limitations of the test” sections of the enclosed technical report for additional information.

Technical notes:

The test screens for pathogenic and likely pathogenic mutations associated with selected single gene conditions, by analyzing circulating cell-free fetal DNA (cfDNA) from a maternal blood sample. Circulating cfDNA is first purified from the plasma component of anti-coagulated maternal whole blood.Through a state-of-the-art technological process, named Next Generation Sequencing (NGS) technique, 29 genes are sequenced at high read depth (>500X). The resulting genetic sequences are analysed via an advanced bioinformatics analysis, to check for the presence of potential mutations in the genes under investigation: CFTR, HBB, CX26(GJB2), CX30(GJB6), ASXL1, BRAF, CBL, CHD7, COL1A1, COL1A2, COL2A1, FGFR2, FGFR3, HDAC8, JAG1, KRAS, MAP2K1, MAP2K2, MECP2, NIPBL, NRAS, NSD1, PTPN11, RAF1, RIT1, SETBP1, SHOC2, SIX3, SOS1. The test is validated for singleton and twin pregnancies with gestational age of at least 10 weeks. Limit of Detection (LOD) of the method: fetal fraction greater than or equal at 2% .

Comments:

Further action:

Verification date: 14/08/2017

Validation date: 14/08/2017

This report represents a true copy to the primary document, that is detained in the archives of Eurofins Genoma Group Srl.

Medical Geneticist

Dr.ssa Marina Baldi

Lab Director

Dr. Francesco Fiorentino

Eurofins Genoma Group Srl

Eurofins Genoma Group Srl

Rome, 14 August 2017

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