GENOMA Molecular Genetics Laboratories Group

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

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

Report date: 14/08/2017 ***Time:*** 14:30

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:*** H48577

Acceptance Date: 09/08/2017 ***Acceptance Time:*** 11:20 ***Collection Date:*** 08/08/2017

Analysis details

Analysis performed: GeneSafe™ De Novo - 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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GENOMA Molecular Genetics Laboratories Group

Results and Conclusions

Result: **NEGATIVE Test Result**
No pathogenic/likely pathogenic mutation(s) detected.
Fetal sex consistent with **Female.**
Fetal Fraction: 4%

Interpretation: This result shows the test has not detected any known pathogenic/likely pathogenic mutation in the targeted genes screened. Negative screening results mean that there is a very **low risk** that the fetus has one of the disorders screened with this test, although no guarantee may be given that the fetus is actually healthy. 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, 25 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: 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

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Eurofins Genoma Group Srl

Lab Director

Dr. Francesco Fiorentino

Eurofins Genoma Group Srl

Rome, 14 August 2017

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