



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

Report date: 14/08/2017

Time: 15:30

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: H48582

Acceptance Date: 09/08/2017

Acceptance Time: 12:20

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



Results and Conclusions

Result: **POSITIVE Test Result**
Pathogenic mutation detected.
Heterozygous mutation **G380R (c.1138G>C)** detected in the **FGFR3** gene.
Fetal sex consistent with **Female**.
Fetal Fraction: 4%

Interpretation: This result shows that the test detected the **heterozygous mutation G380R (c.1138G>C) in the FGFR3 gene**, suggestive of **high risk** for **Achonodroplasia** 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, 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:

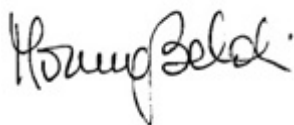
Verification date: 14/08/2017

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

Dr.ssa Marina Baldi



Eurofins Genoma Group Srl

Rome, 14 August 2017

Lab Director

Dr. Francesco Fiorentino



Eurofins Genoma Group Srl