MaterniT™ GENOME Lab Report

Sequenom Laboratories
3595 John Hopkins Court, San Diego, CA 92121 Tel: 877.821.7266
CLIA #: 05D2015356 CAP #: 7527138

FINAL REPORT

Ordering Provider:	**Doe, John, MD**	Patient:	**Sample, Jane**
Provider Location:	**Grand Rapids**	DOB:	**09/13/1970**
Provider Phone:	**555-555-5555**	Patient ID:	**12345-01234**
Date Ordered:	**06/28/2015**	Specimen:	**1035600024**
Date Collected:	**06/29/2015**	Referral Clinician:	**Smith, Jane, GC**
Date Received:	**06/30/2015**	Lab Director:	**Nilesh Dharajiya, MD**
Order ID:	**ORD12345-01234**	Date Reported:	**07/07/2015 6:00 PM PT**

Test Result

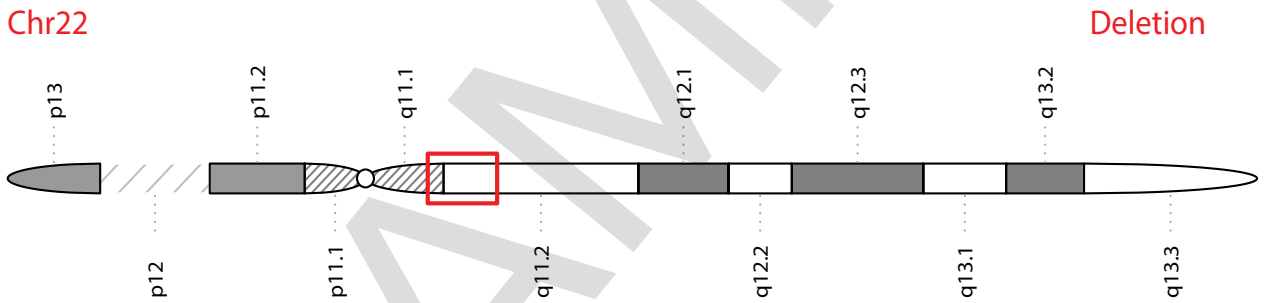
Positive

Loss of chromosome 22(q11.21-q11.21) material

Laboratory Director's Comments

A loss of chromosome 22 material was observed. It is estimated to be 2.6 Mb in size and is suggestive of a deletion in the region 22q11.2, which is associated with DiGeorge syndrome. Genetic counseling and clinical correlation are recommended. Confirmatory testing is required if fetal confirmation and clinical interpretation of the suspected event are desired. Please refer to the "Performance" and "Limitations of the Test" sections of this laboratory report for additional information.

Fetal Fraction: 13%



An approximate 2.35Mb loss of chromosome 22 material was observed, suggestive of a deletion in the region q11.21 - q11.21, associated with DiGeorge syndrome.

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Result Table

Content	Result
AUTOSOMAL ANEUPLOIDIES	
Trisomy 21 (Down syndrome)	Negative
Trisomy 18 (Edwards syndrome)	Negative
Trisomy 13 (Patau syndrome)	Negative
Other autosomal aneuploidies	Negative
SEX CHROMOSOME ANEUPLOIDIES	
Fetal sex	Consistent with male
Monosomy X (Turner syndrome)	Negative
XYY (Jacobs syndrome)	Negative
XXY (Klinefelter syndrome)	Negative
XXX (Triple X syndrome)	Negative
XXY (Klinefelter syndrome)	Negative
XXX (Triple X syndrome)	Negative
SELECT MICRODELETION REGIONS	
22q11 deletion (associated with DiGeorge syndrome)	**Positive**
15q11 deletion (associated with Prader-Willi / Angelman syndrome)	Negative
11q23 deletion (associated with Jacobsen syndrome)	Negative
8q24 deletion (associated with Langer-Giedion syndrome)	Negative
5p15 deletion (associated with Cri-du-chat syndrome)	Negative
4p16 deletion (associated with Wolf-Hirschhorn syndrome)	Negative
1p36 deletion syndrome	Negative