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**

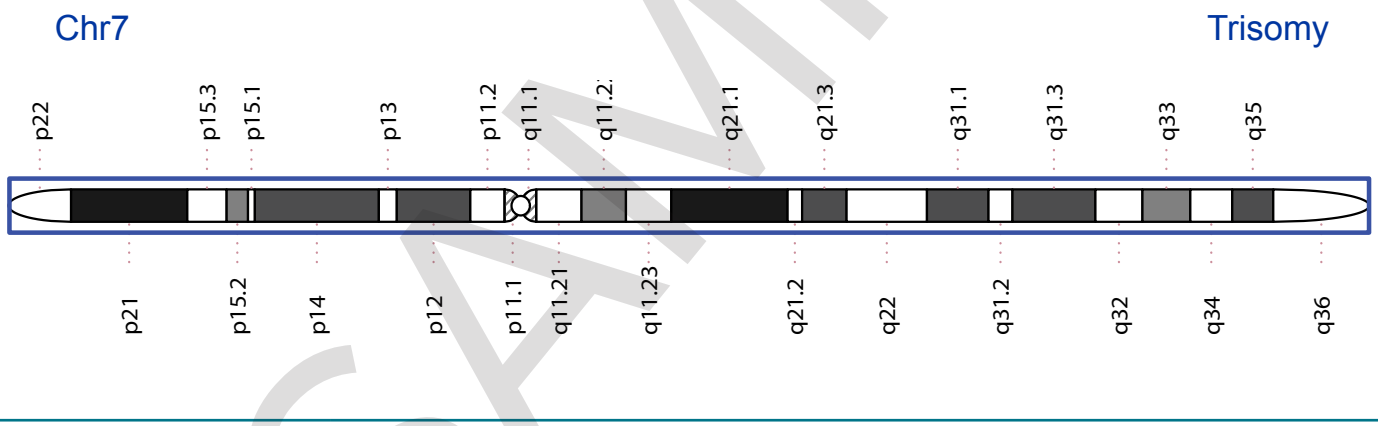
Test Result

Positive
Trisomy 7

Lab Director's Comments

This specimen showed an increased representation of chromosome 7, suggestive of trisomy 7. Genetic counseling and clinical correlation are recommended. Trisomy 7 mosaicsm is commonly reported. Confined placental mosaicism (CPM) is likely. Chromosome 7 is known to be imprinted. Therefore, subsequent trisomy rescue in fetal tissue carries residual risk for UPD. 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: 7%



An approximate 159.00Mb gain of chromosome 7 material was observed, suggestive of trisomy 7.

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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	**Positive**
SEX CHROMOSOME ANEUPLOIDIES	
Fetal sex	Consistent with female
Monosomy X (Turner syndrome)	Negative
XYY (Jacobs syndrome)	Negative
XXY (Klinefelter syndrome)	Negative
XXX (Triple X syndrome)	Negative
GENOME-WIDE COPY NUMBER VARIANTS ≥7 Mb	
Gains/Losses ≥7 Mb	Negative
SELECT MICRODELETIONS	
22q11 deletion (associated with DiGeorge syndrome)	Negative
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