26 YEAR OLD FEMALE

17 WEEKS GA
- Ultrasound findings: normal
- Family history of translocation; mother, child, and sister

MaterniT® GENOME
ORDERED AT 17 WEEKS
Positive: 14.8 Mb gain on 5q and a 16.5 Mb loss on 18q

Normal 50 Kb trace (for comparison)
Each number represents a chromosome, from 1 to 22, X, Y. Note that the orange line stays relatively flat in a normal trace.

Positive MaterniT GENOME trace
Note the significant upward deviation on the orange line for chromosome 5q and downward deviation for chromosome 18q, signifying a gain of material on chromosome 5 and loss of material on chromosome 18.
Case study 5 summary

- 17 weeks GA – Ultrasound findings: normal, family history of translocation
- Mother carries two independent translocations, including a three-way translocation: t(5;10;18)(q33q11.2q21.3) and t(9;13)(p22q22). Mother also has one child who carries the three-way translocation (balanced) and a sister who has an unbalanced form of the t(9;13) translocation
- 17 weeks GA – MaterniT GENOME ordered; positive for gain on chromosome 5q and a loss on chromosome 18q
- 18 weeks GA – Amniocentesis with microarray confirmed 5q duplication and 18q deletion
- 20 weeks GA – Subsequent ultrasound findings: increased nuchal fold, overlapping fingers, edema of the scalp. Fetal echo revealed ASD, VSD and possible anomalous pulmonary venous return
- 25 weeks GA – Ultrasound findings: ventriculomegaly

Ideograms from the MaterniT GENOME lab report with close-up views of the impacted chromosomal trace provide a detailed view of the regions of interest. The purple trace shows the deviations: a gain of chromosome 5q and a loss of chromosome 18q. (Note the purple trace in relation to the blue trace.)

Key points

- MaterniT GENOME correctly identified complex chromosomal abnormalities consistent with an unbalanced translocation, confirmed by ultrasound and diagnostic testing with microarray
- Translocation carriers have a significant risk to experience early pregnancy miscarriage as well as an increased risk of fetal abnormalities due to inheritance of an unbalanced version of the translocation. MaterniT GENOME is the only genome-wide NIPT screening option to date for couples who carry a balanced translocation
- As illustrated by this case study, using traditional NIPT and screening for only common aneuploidies (T13/18/21) with cfDNA may miss clinically relevant abnormalities on other chromosomes, potentially delivering false reassurance

Summary: MaterniT GENOME is the only genome-wide NIPT to date; it detects up to 30% more chromosomal information than other NIPTs; detects chromosomal aneuploidies missed by traditional NIPT; thereby providing earlier awareness and more proactive pregnancy management options.


Results from case studies are not predictive of results in other cases. Results in other cases may vary.

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