MaterniT® GENOME

CLINICAL UPDATE

Case study 5: three-way translocation

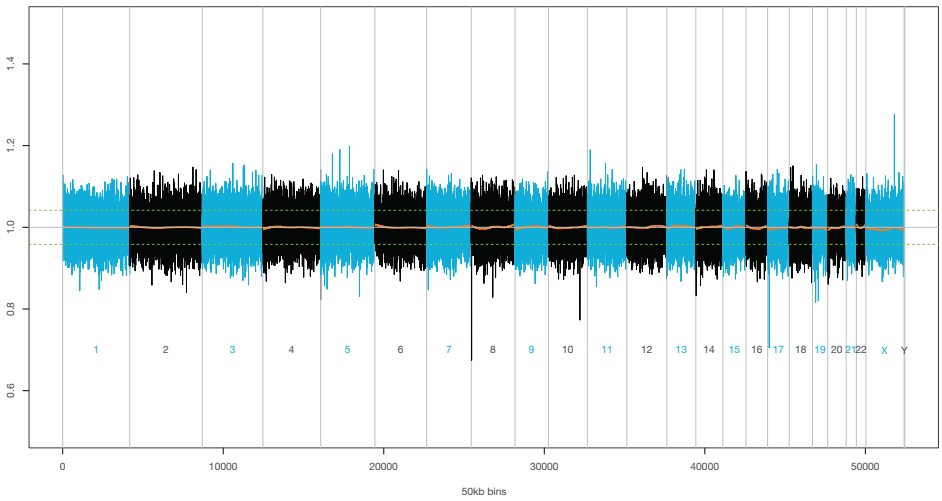
26 YEAR OLD FEMALE

17 WEEKS GA

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

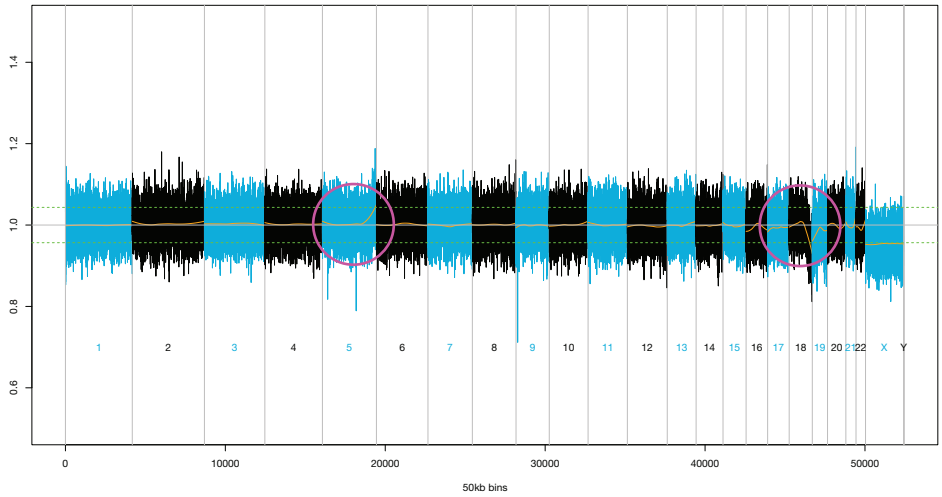
MATERNIT® GENOME ORDERED AT 17 WEEKS

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.

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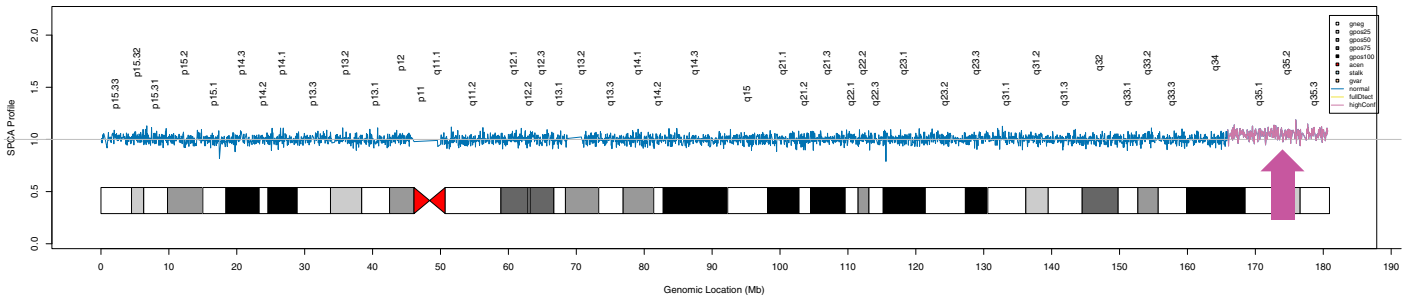
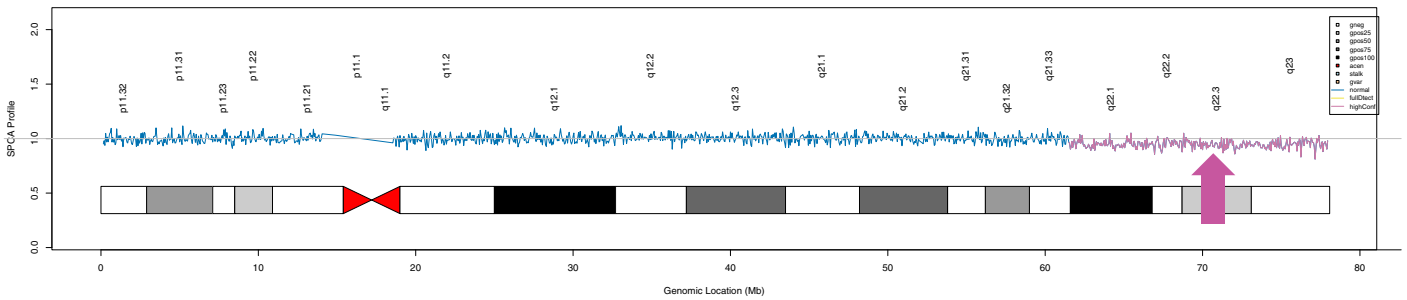
Key points

- 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.[1] MaterniT GENOME is a genome-wide NIPT screening option 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

MaterniT GENOME detects up to 30% more chromosomal information than other NIPTs[2,3]; detects chromosomal aneuploidies missed by traditional NIPT; thereby providing earlier awareness and more proactive pregnancy management options.



Chromosome 5 – 14.8 Mb gain of 5(q34-q35.3)



Chromosome 18 – 16.5 Mb loss of 18(q21.33-q23)

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.)

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

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

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1. Gardner R, Sutherland G, Shaffer L. *Chromosome abnormalities and genetic counseling*. Oxford, UK: Oxford University Press; 2012.
2. Ehrich M, Tynan J, Mazloom A, et al. Genome-wide cfDNA screening: clinical laboratory experience with the first 10,000 cases. *Genet Med*. 2017;19(12):1332-1337.
3. Internal data.