MaterniT® GENOME

CLINICAL UPDATE
Case study 3: average risk

31 YEAR OLD FEMALE

24 WEEKS GA

- Prior negative Harmony NIPT test
- Fetal anomalies identified via ultrasound
- Positive CMA (NOS)

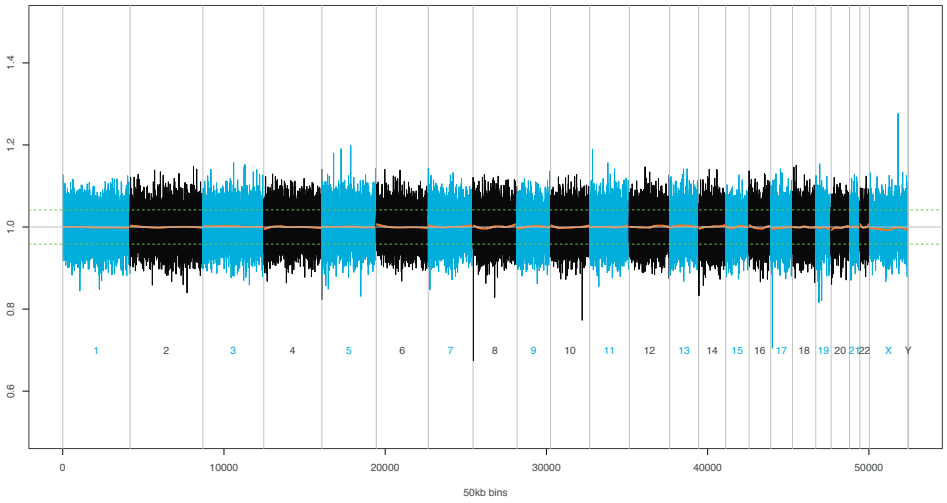
(chromosomal microarray not otherwise specified)

MATERNIT® GENOME ORDERED AT 24 WEEKS*

Positive: 14.6 Mb duplication 1q42.2q44

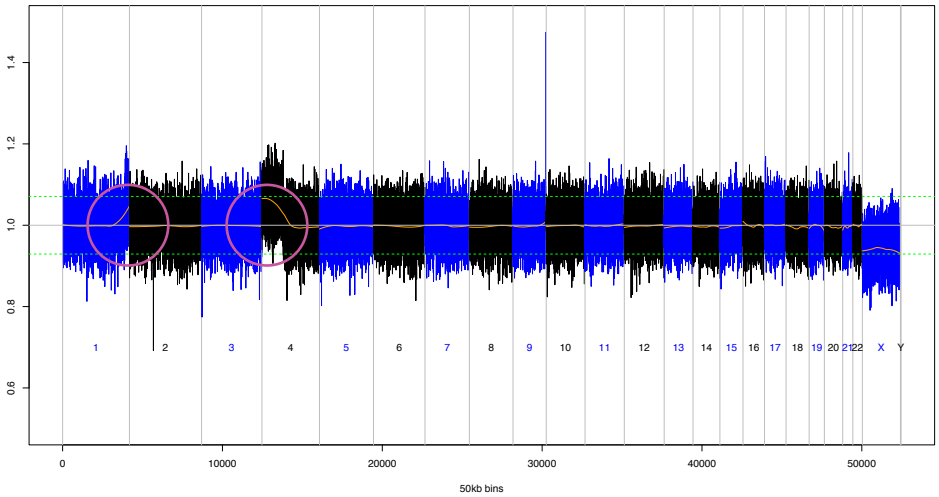
73.45 Mb duplication 4p16.3q13.3

(Lab Director comment: "These results raise the possibility of a supernumerary der(4)(1q;4p).")



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 1q and for chromosome 4p signifying a gain of material on both chromosomes.

*Post CMA at the request of the clinician for research purposes.

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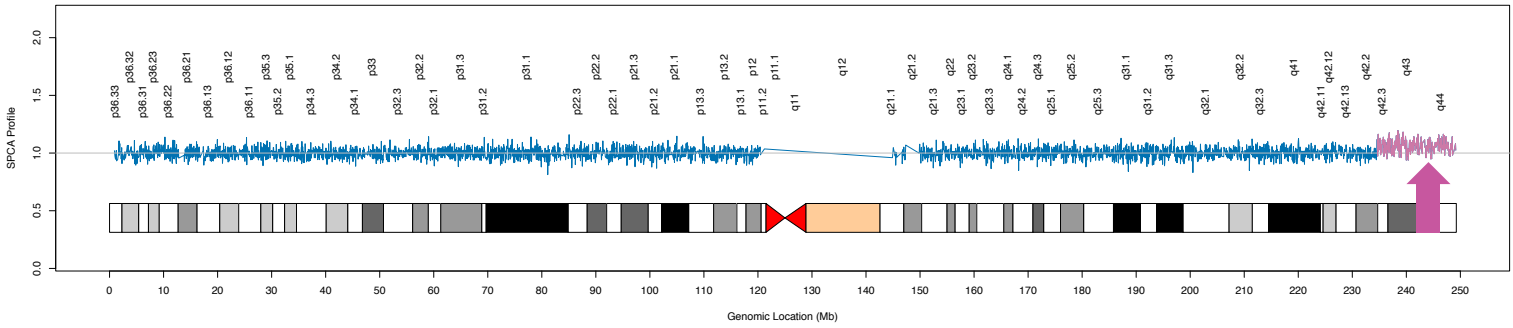
Key points

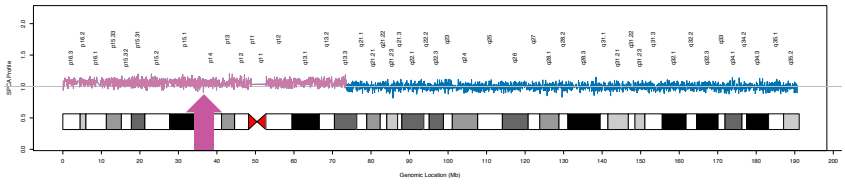
- MaterniT GENOME correctly identified complex chromosomal abnormalities following an ultrasound finding suggestive of different chromosomal and genetic conditions, confirmed by diagnostic testing
- MaterniT GENOME detects chromosomal abnormalities not detected by other NIPTs
- As illustrated by this case study, using traditional NIPT and screening for only common aneuploidies (T13/18/21) may miss clinically relevant abnormalities on other chromosomes, potentially delivering false reassurance

MaterniT GENOME detects up to 30% more chromosomal information than other NIPTs[1,2]; detects chromosomal aneuploidies missed by traditional NIPT; thereby providing earlier awareness and more proactive pregnancy management options.

Chromosome 1 – 14.6 Mb duplication 1q42.2q44



Chromosome 4 – 73.45 Mb duplication 4p16.3q13.3



Ideograms from the MaterniT GENOME lab report with close up views of each impacted chromosomal trace provide a detailed view of the regions of interest. The purple shows the deviations: a gain on chromosome 1q and gain on chromosome 4p (Note the purple trace in relation to the blue trace).

Case study 3 summary

- Initial ultrasound suggested fetal anomalies
- MaterniT GENOME ordered (24 weeks, ultrasound only) – positive for gain on chromosomes 1 and 4
- Maternal balanced translocation (1;4) subsequently detected by karyotype
- MaterniT GENOME test results confirmed by diagnostic testing
- **MaterniT GENOME test results identified chromosomal abnormalities not detected by other NIPTs**

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

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1. Mathias E, Tynan J, Mazloom A, et al. Genome-wide cfDNA screening: clinical laboratory experience with the first 10,000 cases. *Genet Med*. June 2017.
2. Internal data.