MaterniT® GENOME

CLINICAL UPDATE
Case study 2: holoprosencephaly

FEMALE

Advanced maternal age (AMA)

PRIMIGRAVIDA

UNREMARKABLE FAMILY HISTORY

- First trimester ultrasound (13 weeks)
  - Physician suspected alobar holoprosencephaly not reported to lab at time of first draw

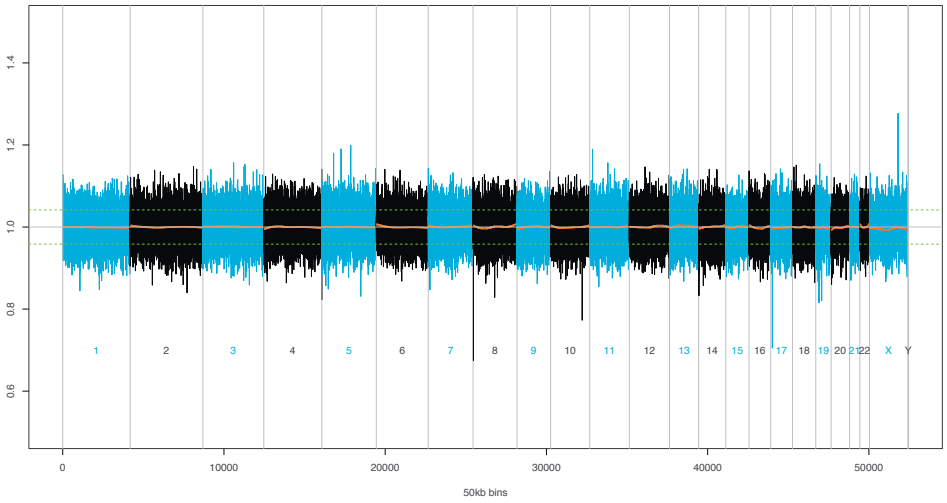
MATERNIT® GENOME ORDERED AT 13 WEEKS

(AMA only indicated on TRF) – QNS

Redraw ordered at 15 weeks - positive:

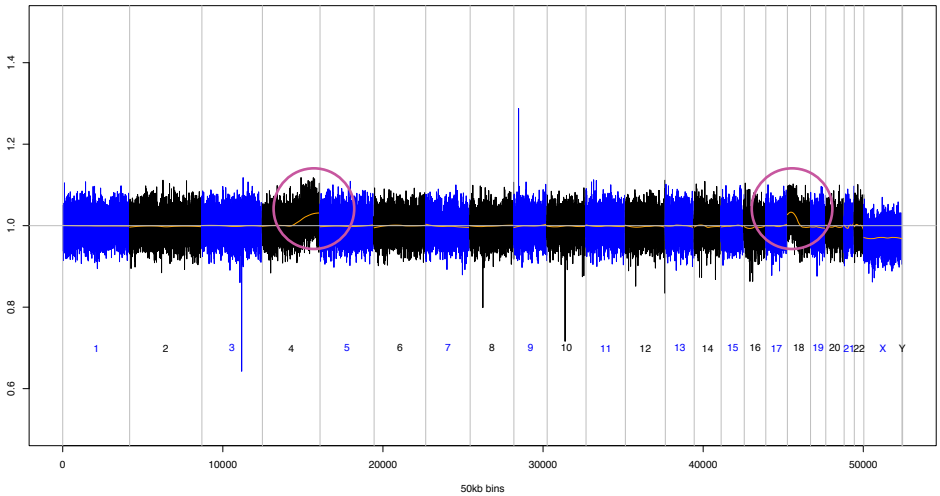
60.40 Mb gain 4q28.2-q35.2

35.15 Mb gain 18p11.32-q12.2



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 4q and for chromosome 18p signifying a gain of material on both chromosomes.

Integrated GENETICS
LabCorp Specialty Testing Group

rep-1167-v1-0618 | L18791-0618-1

MaterniT GENOME

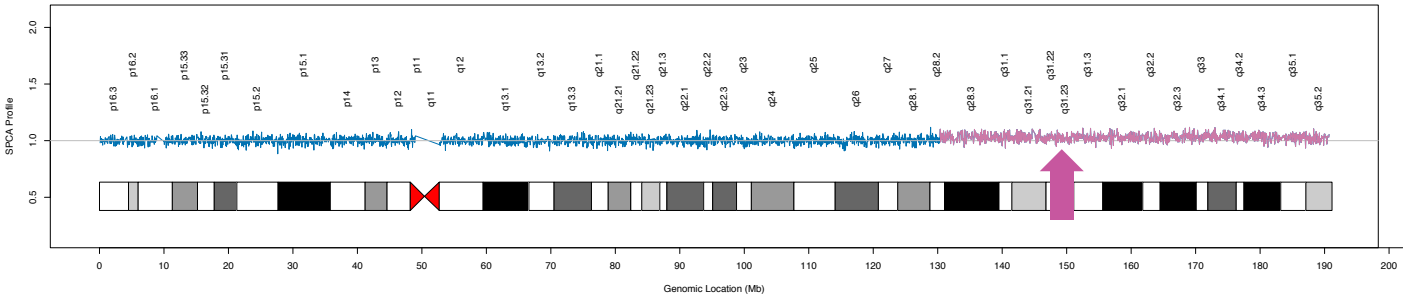
CLINICAL UPDATE
Case study 2: holoprosencephaly

Key points

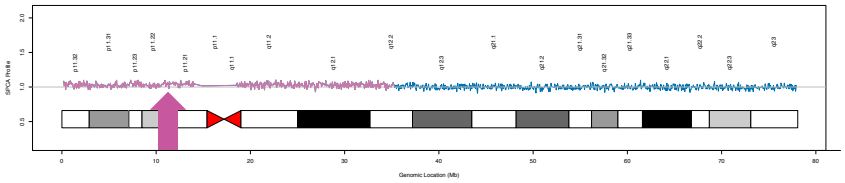
- MaterniT GENOME correctly identified complex chromosomal abnormalities following an ultrasound finding suggestive of different chromosomal and genetic conditions, confirmed by diagnostic testing
- MaterniT GENOME detects chromosomal abnormalities not detected by traditional NIPT (T13/18/21)
- As illustrated by this case study, ultrasound alone, or in combination with family history, can provide incomplete results, 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[1,2]; detects chromosomal aneuplodies missed by traditional NIPT; thereby providing earlier awareness and more proactive pregnancy management options.

Chromosome 4 – 60.40 Mb gain 4q28.2-q35.2



Chromosome 18 – 35.15 Mb gain 18p11.32-q12.2



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 4q and gain on chromosome 18p (note the purple trace in relation to the blue trace).

Case study 2 summary

- Initial ultrasound (13 weeks) – suggested alobar holoprosencephaly, but not reported to lab
- MaterniT GENOME ordered (13 weeks, AMA only) – QNS, redraw (15 weeks) – positive for gain on chromosomes 4 and 18
- Amniocentesis performed at 17 weeks
  - FISH: consistent with trisomy 18
  - Karyotype: 47,XY,+18,add(18)(q21.3)
  - Partial trisomy of 18 and “an unknown chromosome segment”
- Miscarriage at ~20 weeks
- Parental karyotype confirmed both parents to be translocation carriers
- Maternal balanced translocation (4;18) subsequently confirmed by maternal blood testing
- MaterniT GENOME test results confirmed by amniocentesis, maternal testing and POC testing
- **MaterniT GENOME test results offered more clarity than ultrasound alone**

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

Integrated GENETICS
LabCorp Specialty Testing Group

1. 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.
2. Internal data.