FEMALE
Advanced maternal age

PRIMIGRAVIDA
UNREMARKABLE FAMILY HISTORY
- First trimester ultrasound (13 weeks)
- Physician suspected alobar holoprosencephaly
- Ultrasound finding not reported to lab at time of first draw

MATERNIT® GENOME ORDERED AT 13 WEEKS
(AMA listed as only indication on test form) – 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.

Results from case studies are not predictive of results in other cases. Results in other cases may vary.
Key points
• MaterniT GENOME correctly identified complex chromosomal abnormalities following an ultrasound finding that could be suggestive of a number of different chromosomal abnormalities or genetic disorders, 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

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

Case study 2 summary
• Initial ultrasound (13 weeks) – suggested alobar holoprosencephaly, but not reported to lab
• MaterniT GENOME ordered (13 weeks, AMA indication only)–QNS, redraw (15 weeks)–positive for gain on chromosomes 4 & 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 detected 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