AMA FEMALE

HISTORY OF MULTIPLE MISCARRIAGES
- First trimester ultrasound (12 weeks)
- Nuchal translocation (NT) - Normal
- First trimester serum screen
- Subsequently positive
- Increased risk of trisomy 13 or trisomy 18

MATERNIT® GENOME ORDERED AT 12 WEEKS
Positive: 10.90 Mb loss at 5p15-ter and 30.85 Mb gain at 16q21-q24.3.

Normal 50 Kb trace (for comparison)
Each number below the trace 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 downward deviation on the orange line for chromosome 5p and upward deviation for chromosome 16q signifying a loss and gain of material, respectively.
Key points

- Normal ultrasound > followed by abnormal serum screen > MaterniT GENOME ordered at 12 weeks
- MaterniT GENOME correctly identified chromosomal abnormalities different than those suggested by serum screen and in the presence of a normal ultrasound. Supported by confirmatory amniocentesis
- As illustrated by this case study, self reported family history can be incomplete, 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 and detects up to 30% more chromosomal information than other NIPTs

Case study 1 followup

- Subsequent ultrasound at 16 weeks - Normal
- Amniocentesis at 16 weeks: Revealed abnormal result: 46,XX,der(5)t(5;16)(p15.2;q21)
- Unreported maternal translocation: t(5;16)(p15.1;q21) only disclosed by patient to Genetic Counselor after MaterniT GENOME test results were issued
- Follow-up testing on POC (products of conception) confirmed amniocentesis results
- MaterniT GENOME test result confirmed by cytogenetics

1. Incidence of fetal chromosomal and subchromosomal abnormalities based on Wellseley et al. Eur J Hum Genet. 2012 May; 20(5):521-6. Results from case studies are not predictive of results in other cases. Results in other cases may vary.