CLINICAL UPDATE
Case study 4: Pallister Killian mosaic syndrome
Bulletin 5 | October 2016

33 YEAR OLD FEMALE
LATE TO CARE 27 WEEKS GA
Ultrasound findings (NOS—not otherwise specified)

MaterniT® GENOME
ORDERED AT 27 WEEKS
Positive: 34.3Mb gain 12(p11.1-p13.33)
This region has been reported to be involved in Pallister-Killian mosaic syndrome

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 12p, signifying a gain of material on that genomic location.
Case study 4 summary

- Patient late to care
- 27 weeks GA – Ultrasound findings: positive; club foot, diaphragmatic hernia, increased nuchal fold, patient initially refused amniocentesis
- 27 weeks GA – MaterniT GENOME ordered (ultrasound findings not disclosed to the lab); positive for gain of chromosome 12p
- 28 weeks GA – Amniocentesis with microarray reported 80% mosaicism i(12p) consistent with Pallister Killian syndrome. Karyotype showed isochromosome 12p in all metaphase cells
- Will pursue palliative care upon delivery

Chromosome 12 – 34.3Mb gain 12(p11.1-p13.33)

Ideogram from the MaterniT GENOME lab report with close-up view of the impacted chromosomal trace provide a detailed view of the region of interest. The purple trace shows the deviation: a gain on chromosome 12p. (Note the purple trace in relation to the blue trace.)

Key points

- MaterniT GENOME correctly identified complex chromosomal abnormalities consistent with Pallister-Killian mosaic syndrome, confirmed by diagnostic testing (Microarray and Karyotype)
- "Pallister-Killian mosaic syndrome appears to be a rare condition, although its exact prevalence is unknown. This disorder may be underdiagnosed because it can be difficult to detect in people with mild signs and symptoms. As a result, most diagnoses are made in children with more severe features of the disorder. More than 150 people with Pallister-Killian mosaic syndrome have been reported in the medical literature."\(^1\)
- Esoteric findings including Pallister Killian are individually rare, but collectively common, and not associated with advanced maternal age. Using an advanced NIPT screening test like MaterniT GENOME offers more clinically relevant information to the clinician and patient
- As illustrated by this case study, only a genome-wide cfDNA test is capable of identifying abnormalities on other chromosomes. 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

Summary: MaterniT GENOME is the only genome-wide NIPT to date; it detects up to 30% more chromosomal information than other NIPTs\(^2\); detects chromosomal aneuploidies missed by traditional NIPT; thereby providing earlier awareness and more proactive pregnancy management options.