MaterniT® GENOME

CLINICAL UPDATE

Case study 4: Pallister-Killian mosaic syndrome

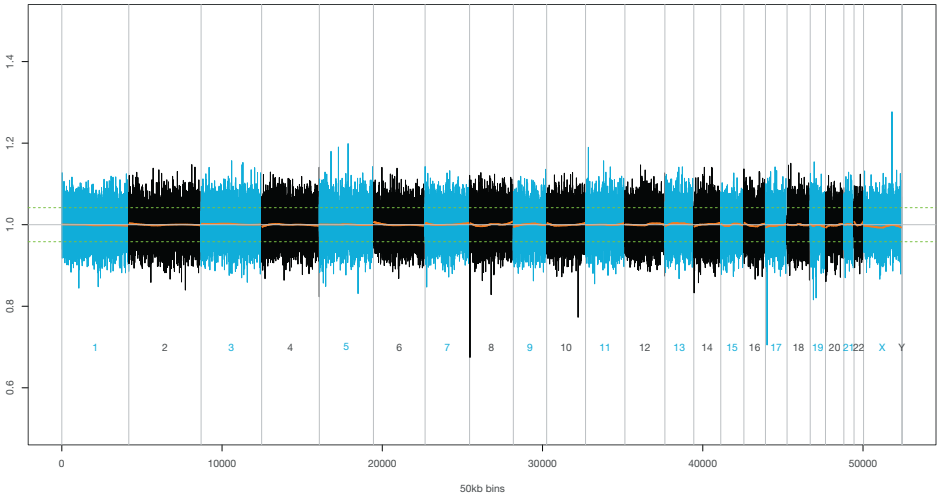
33 YEAR OLD FEMALE

LATE TO CARE 27 WEEKS GA

Ultrasound findings (NOS–not otherwise specified)

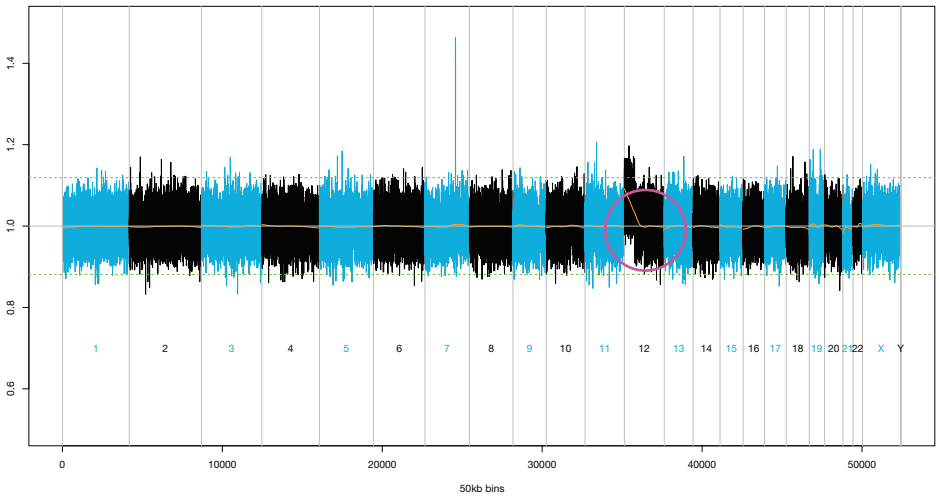
MATERNIT® GENOME ORDERED AT 27 WEEKS

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.

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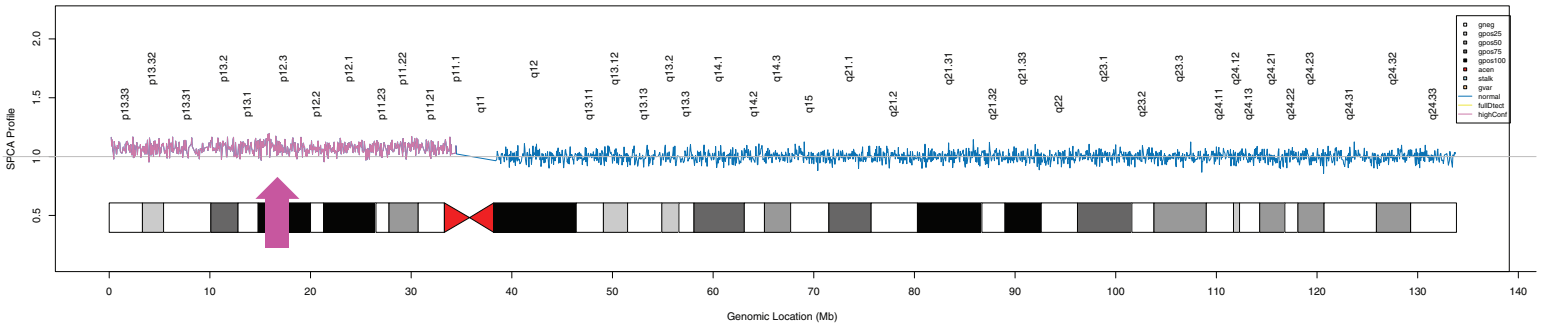
Key points

- MateriT GENOME correctly identified complex chromosomal abnormalities consistent with Pallister-Killian mosaic syndrome, confirmed by diagnostic testing (Microarray and Karyotype)
- According to Genetics Home Reference, "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 potentially associated with advanced maternal age.[2] Using an advanced NIPT screening test like MateriT GENOME offers more clinically relevant information to the clinician and patient
- As illustrated by this case study, 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

MateriT GENOME detects up to 30% more chromosomal information than other NIPTs[3,4]; detects chromosomal aneuploidies missed by traditional NIPT; thereby providing earlier awareness and more proactive pregnancy management options.



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

Ideogram from the MateriT 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.)

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 – MateriT 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-Kilian syndrome. Karyotype showed isochromosome 12p in all metaphase cells
- Will pursue palliative care upon delivery

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

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1. US National Library of Medicine. Genetics Home Reference. Pallister-Killian mosaic syndrome. https://ghr.nlm.nih.gov/condition/pallister-killian-mosaic-syndrome. Accessed January 9, 2018.
2. Blyth M, Maloney V, Beal S, et al. Pallister-Killian syndrome: a study of 22 British patients. *J Med Genet* 2015;52:454-464.
3. 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.
4. Internal data