19 YEAR OLD FEMALE

Gestational age: 12 weeks, 4 days

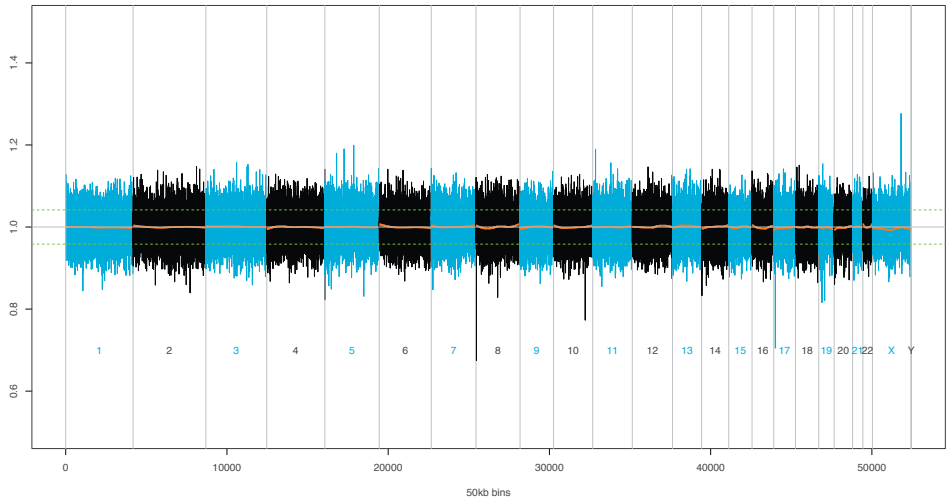
FAMILY HISTORY

No known risk factors
MaterniT 21 PLUS + SCA + ESS ordered, result negative

SCA - Sex Chromosome Aneuploidies
ESS - Enhanced Sequencing Series
(Clinically relevant microdeletions and trisomies)

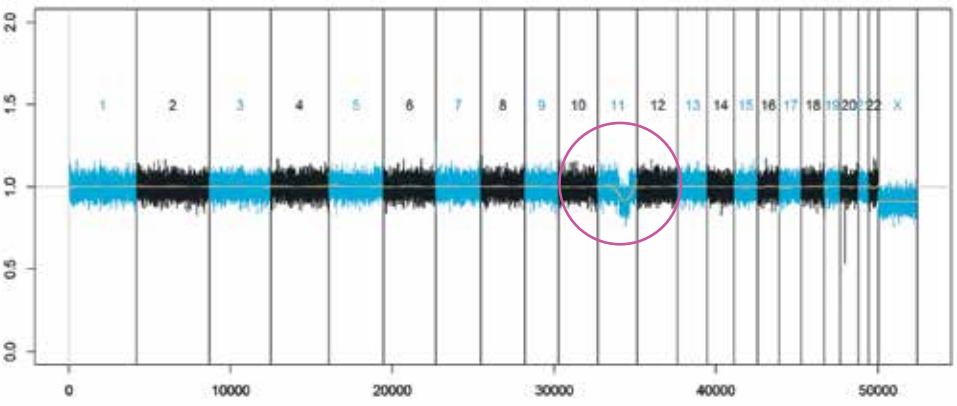
GENOME-FLEX ORDERED AT 27 WEEKS GA

Due to ultrasound findings of clubfoot and heart defect
Positive: 32.25 Mb 11q interstitial deletion



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 GENOME-Flex trace

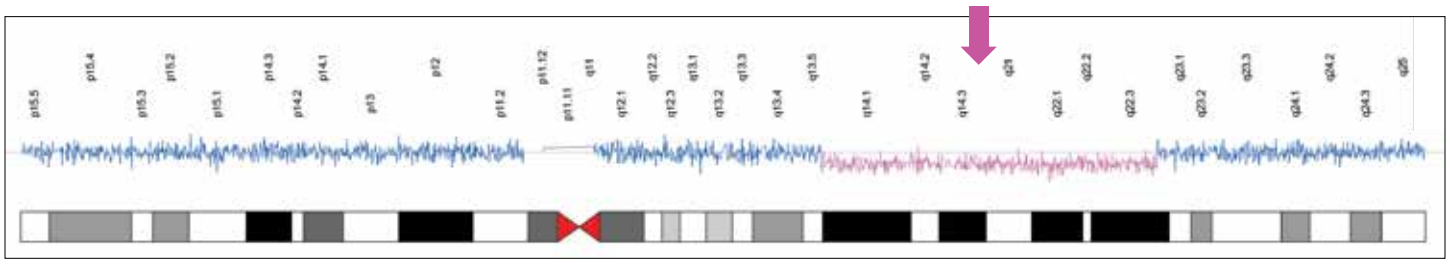
Note the significant downward deviation on the orange line for chromosome 11q signifying a loss of chromosomal material.

Key points

- A traditional NIPT will only report on specific regions of the genome (typically T13/18/21, SCAs, X/Y), therefore missing chromosomal abnormalities located in regions not screened
- **MaterniT 21 PLUS now with GENOME-Flex** allows previously run MaterniT 21 PLUS samples to be rapidly re-sequenced using the deeper sequencing power of MaterniT GENOME to screen all 23 pairs of chromosomes
- As illustrated by this case study, using traditional NIPT screening for average risk patients may miss clinically relevant abnormalities on other chromosomes. However, a flexible NIPT like MaterniT 21 PLUS offers a new NIPT pathway should the patient become high-risk later in pregnancy due to suspected chromosomal abnormalities

MaterniT 21 PLUS is the only NIPT to offer GENOME-Flex, a new NIPT high risk pathway. MaterniT GENOME detects up to 30% more chromosomal information than other NIPTs[2,3]; detects chromosomal aneuploidies and copy number variants missed by traditional NIPT; thereby providing earlier awareness and more proactive pregnancy management options.

Chromosome 11 - 32.25 Mb 11q interstitial deletion



Ideogram from the MaterniT GENOME lab report with a closeup view of the impacted chromosomal trace provides a detailed view of the region of interest. The amplitude of the purple trace shows the deviation: in this case a loss on chromosome 11q.

Case study 6 summary

- 19 year old average risk patient, no known risk factors
- MaterniT 21 PLUS (with SCA + ESS) ordered at 12 weeks GA - Negative result
- Subsequent ultrasound identified suspected clubfoot and heart defect
- **GENOME-Flex** ordered at 27 weeks GA (no additional blood draw required) - **Positive result - 32.25 Mb 11q interstitial deletion**
- A confirmatory diagnostic procedure may be recommended according to clinical practice and society guidelines, but this option may not be desired by the patient. This patient declined amniocentesis with microarray
- Baby delivered with multiple anomalies
- **GENOME-Flex was able to re-sequence this patient's original MaterniT 21 PLUS sample and identify clinically relevant abnormalities not detected by traditional NIPT (typically without a redraw and with a rapid 72 hour turnaround time)**

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. Illumina. https://www.illumina.com/clinical/illumina_clinical_laboratory/verifi-prenatal-tests.html. Accessed August 15, 2018.
2. 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.
3. Internal data.