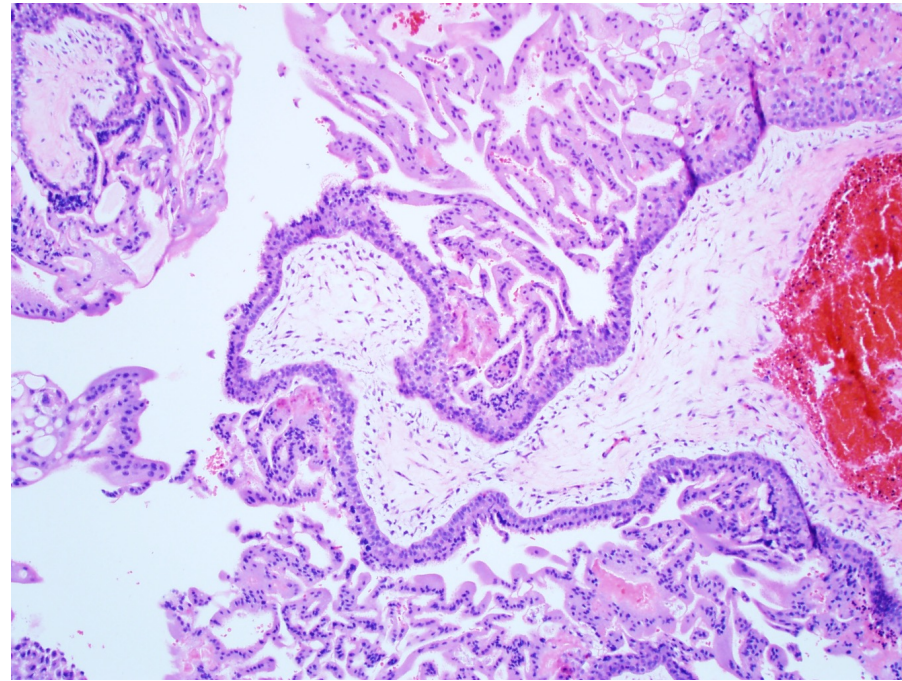
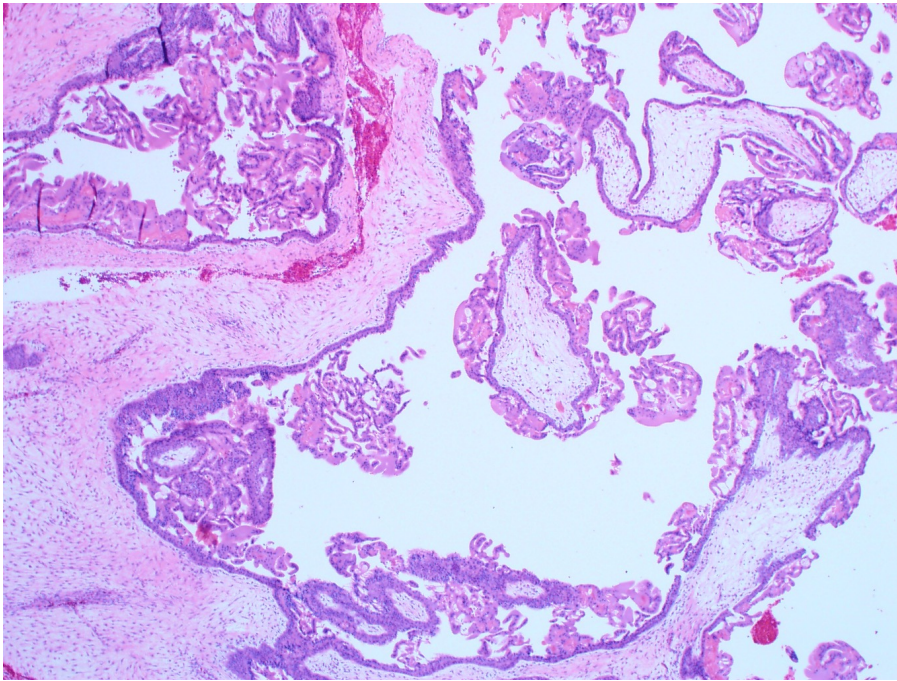




38-year-old woman presented with missed abortion



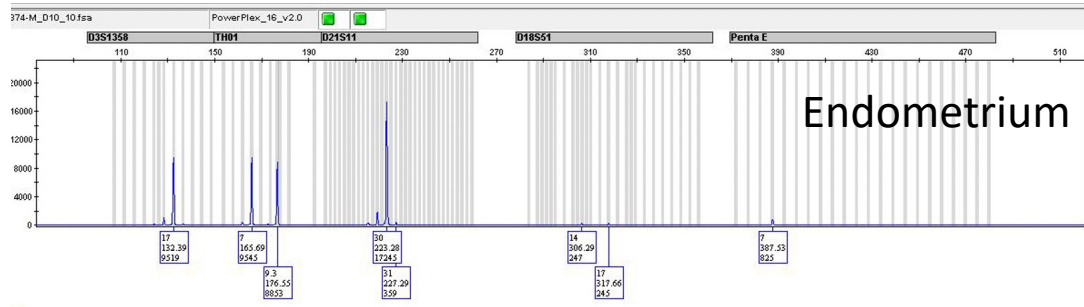
Diagnostic Options

A: Non-molar missed abortion

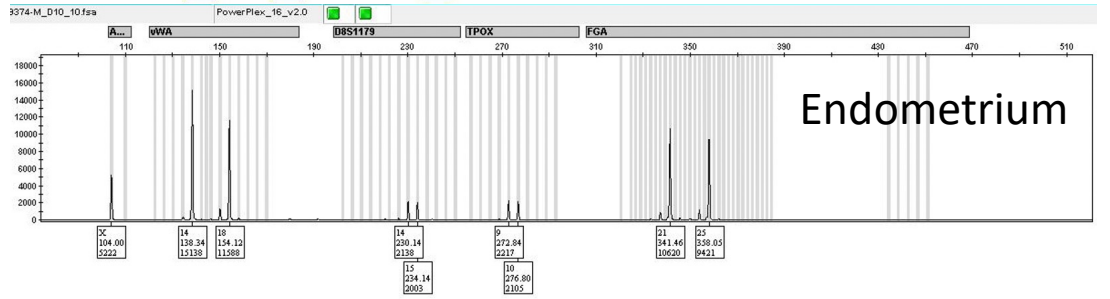
B: Monospermic partial mole

C: Down syndrome

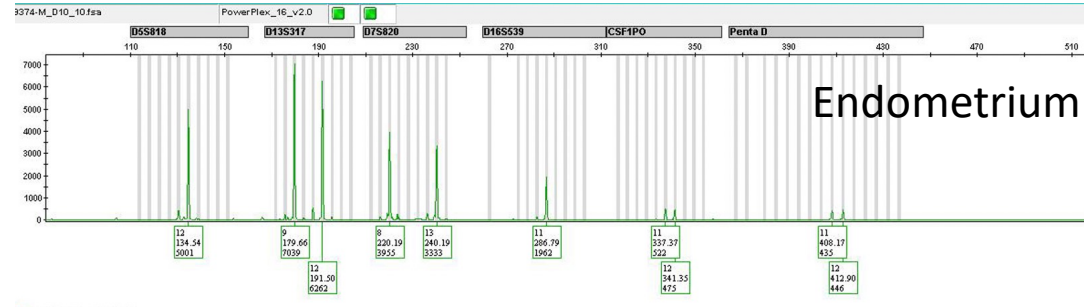
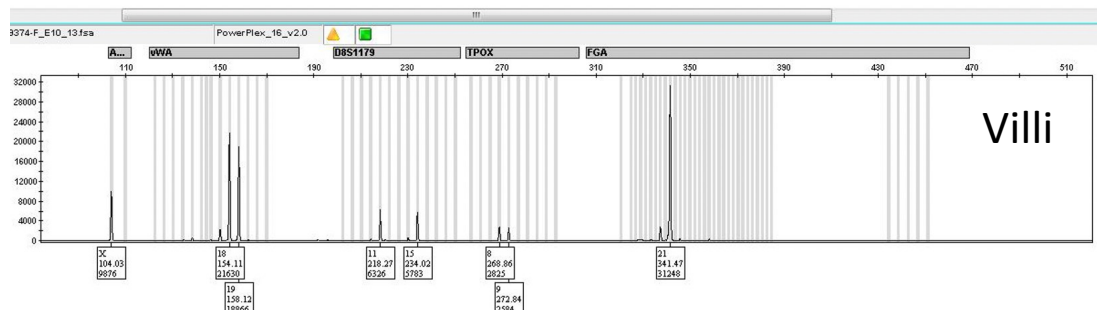
D. Dispermic complete mole



S17-G374-M_D10_10 fsa



S17-G374-M_D10_10 fsa



S17-G374-M_D10_10 fsa



Full genotyping panel: The allelic pattern of the chorionic villi shows a balanced biparental profile at most STR loci except the presence of three allelic copies (*) at D21S11 locus (2 copies of allele 30 and 1 copy of allele 32.2) and Penta D locus (3 distinct alleles – 9, 11 and 12).

Case Summary

Chorionic villi show abnormal configurations, hydropic change and marked villous trophoblastic hyperplasia. STR genotyping demonstrates an isolated allelic copy gain at two chromosome 21 loci (D21S11 and Penta D), consistent with Trisomy 21 gestation (Down syndrome).

Final Diagnosis: Trisomy 21 Gestation (Down Syndrome)

Discussion

Trisomy gestations are among the most frequent products of conception in missed or spontaneous abortions and can histologically simulate a partial hydatidiform mole. Trisomy 16 and Trisomy 21 are the most common. Multiple trisomies can also occur.

All histological features traditionally described in a partial mole are commonly seen in a trisomy gestation. These include hydropic change, abnormal villous contours, trophoblastic pseudoinclusions, two populations of chorionic villi, and abnormal trophoblastic hyperplasia. It is important to note that none of the above histological parameters (single or combined) are specific and sensitive enough to diagnose a partial mole (International Journal of Gynecological Pathology. 2013, 32:307-315). Ancillary studies are essential to separate various trisomy gestations from a true hydatidiform mole.