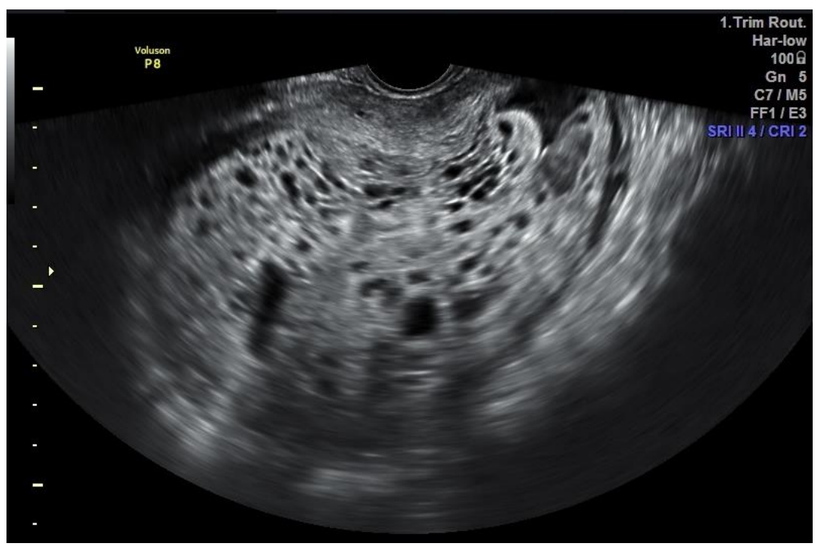
P-37

A Trispermic Tetraploid Pregnancy Mimicking Complete Hydatidiform Mole: Molecular Genetic Resolution of Clinicopathological Discordance.

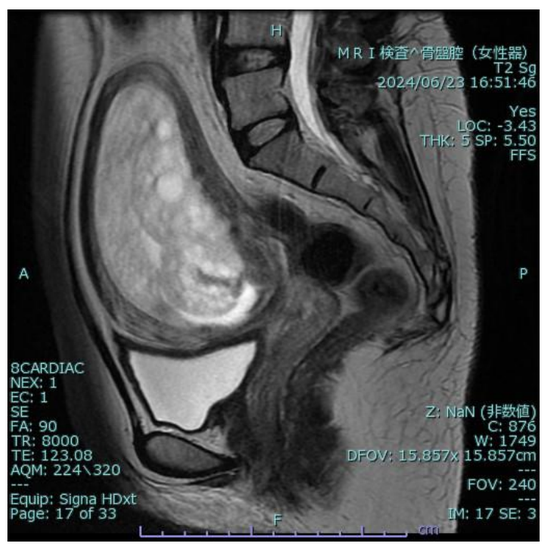
Yasushi Sasaki, Rei Matsuura, Shingo Miyamoto, Ken Nakayama, Miki Morioka
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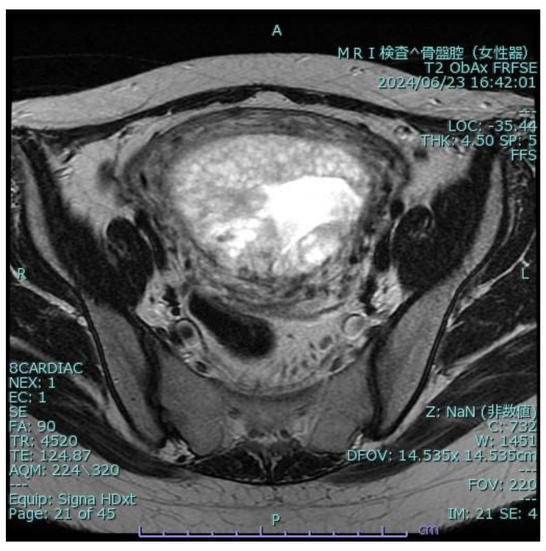
Case:

A 39-year-old gravida 1 para 1 woman presented with abnormal uterine bleeding. Serum human chorionic gonadotropin (hCG) was 16,329.6 mIU/mL. Transvaginal ultrasonography showed a multivesicular intrauterine lesion without identifiable fetal structures. Magnetic resonance imaging (MRI) revealed a 10-cm heterogeneous multicystic intrauterine mass without myometrial invasion, strongly suggesting complete hydatidiform mole (CHM). After uterine evacuation, histopathology showed hydropic villi with features consistent with a partial hydatidiform mole (PHM), and p57 immunostaining was positive. Because these findings were discordant with the clinicoradiological impression, short tandem repeat (STR) genotyping was performed and showed an unusual pattern consistent with three paternal and one maternal genomic complements, suggesting a trispermic tetraploid conception. Subsequent fluorescence in situ hybridization (FISH) analysis using probes for chromosomes 2, 4, 9, and 17 demonstrated four signals in multiple nuclei, confirming tetraploidy. Serum hCG declined to 286.9 mIU/mL at 1 week, 3.5 mIU/mL at 1 month, and <0.5 mIU/mL at 2 months, remained undetectable thereafter, and no gestational trophoblastic neoplasia developed.

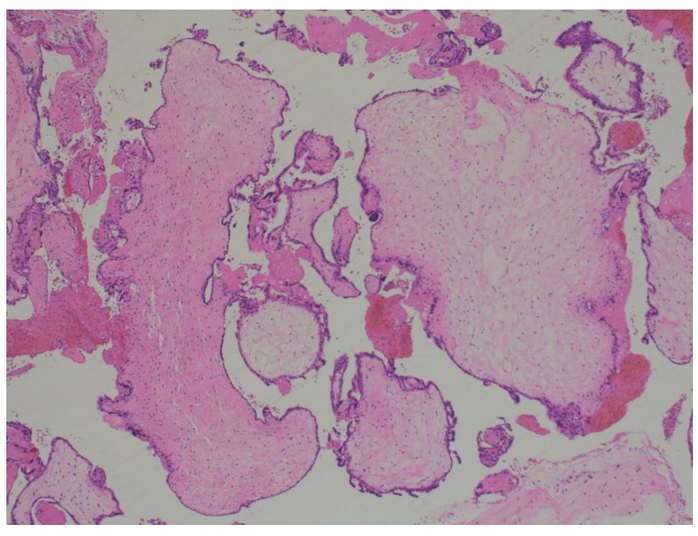


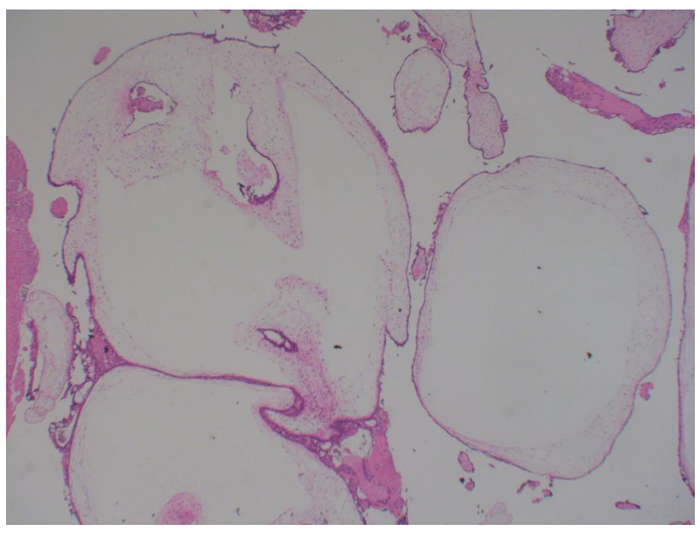
Transvaginal ultrasound finding



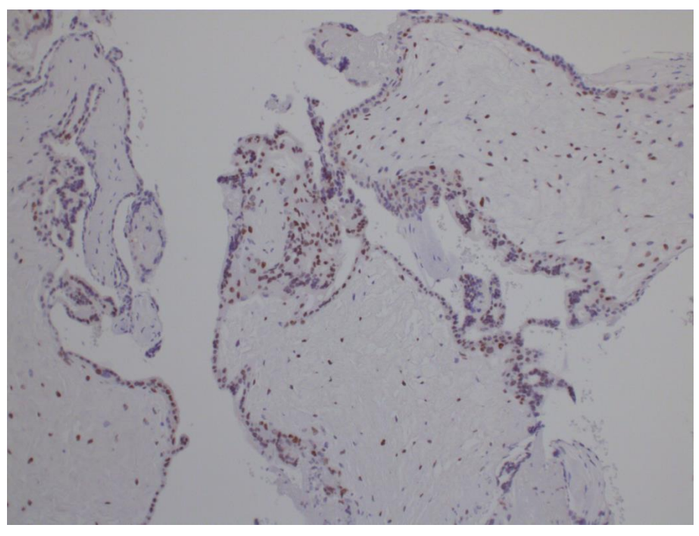


MRI findings before manual vacuum aspiration

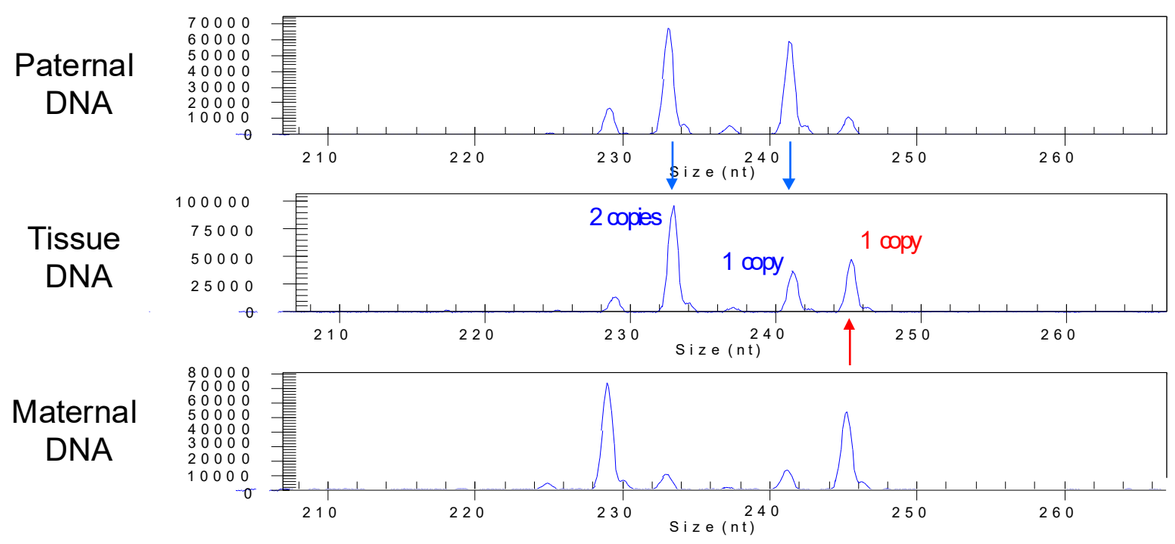




Microscopic findings: HE staining suggested PHM

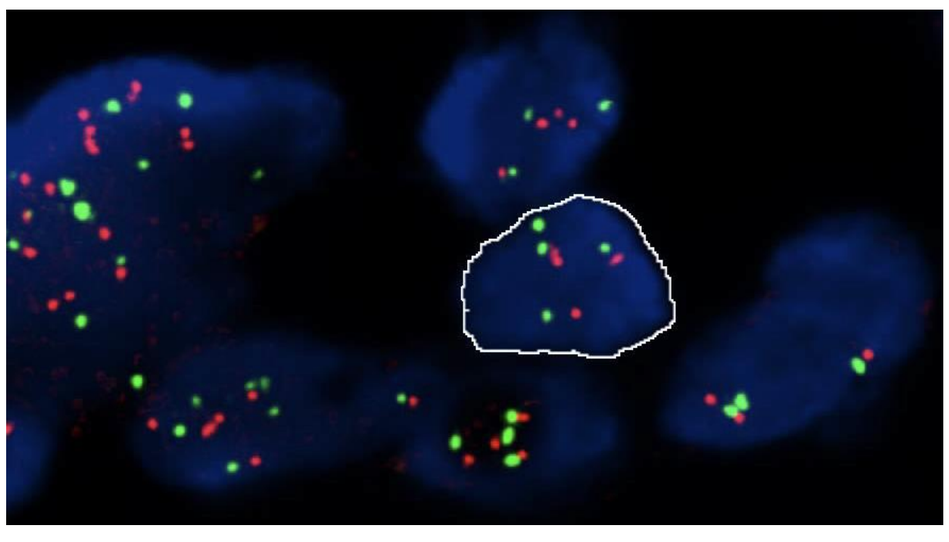


Microscopic findings: p57^{kip2} staining positive

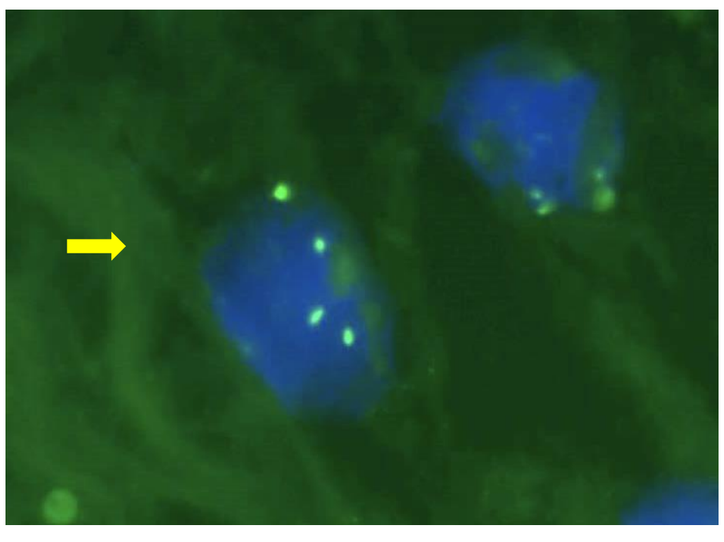


STR analysis of D22S684

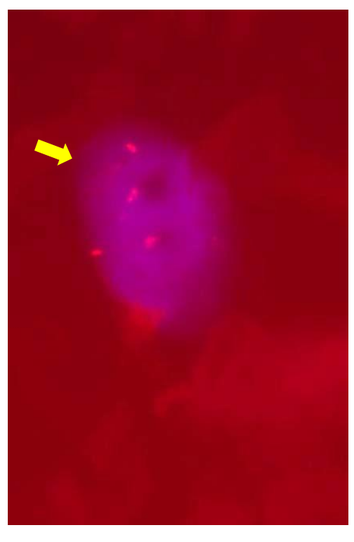
In addition, we performed STR analysis of D18S51, D20S471, FGA, vWA.
All of these analysis suggested **trispermic tetraploid conception**.



HER2 FISH analysis of Chromosome 17



Chromosome 4 FISH analysis



Chromosome 9 FISH analysis

Conclusion:

PHM-like morphology may occur in a tetraploid conception and cannot alone define its genetic constitution. Although clinicoradiological findings strongly suggested CHM and histopathology resembled PHM, combined STR genotyping and FISH established a trispermic tetraploid conception.
This case highlights the complementary roles of morphology and molecular genetic testing in resolving clinicopathological discordance in atypical conceptions.

Conflict of interest statement: The authors wish to report that there are no conflict of interest.