Ovarian Insufficiency: The Hidden Uterus
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Background
Primary ovarian insufficiency and hypoplastic uterus are reported in females but often misdiagnosed under infertility.

Objectives
Propose an algorithm for primary amenorrhea with absent uterus

Case Presentation
17 years old female virgin with primary amenorrhea:
• BH:158.5cm(40th percentile), BW:45kg (10th percentile). BMI 17.9 underweight
• Female genitalia, Tanner stage B1PH3, no Turner features
• Vague ovaries and absent uterus under pelvic sonography and magnetic resonance imaging
• Lab tests: High FSH/LH, low Estrogen, normal Testosterone (female reference), TSH & Prolactin normal
• Chromosome 46 XX, karyotype
• Bone age 13 years

Pelvic Ultrasound

Impression and Further Survey
• Suspect Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome variant
• Suspect Primary Ovarian Insufficiency with Mullerian agenesis
• Inform family of possible infertility
• Gonadotropin releasing hormone test: Hypergonadotropic hypogonadism
• Bone marrow density with Z-score -2.7

Bone age 13 years

Karyotype
Chromosome 46 XX, no Turner features

Bone marrow density

Primary Amenorrhea with Absent Uterus
Evaluate female phenotype and Tanner staging
Total Testosterone and Karyotype with Pituitary-Gonadal-Axis evaluation

Other:
Eg. 45,X Turner Syndrome

Absent Uterus:
MRKH syndrome
• Mullerian agenesis
• Testosterone in normal female range, normal FSH, normal breast development

1. Complete androgen insensitivity syndrome
2. 5-alpha reductase deficiency

Final Diagnosis
1. Primary ovarian insufficiency with premature uterus
2. Osteoporosis

Conclusion
• Mullerian agenesis by image modalities can be extremely challenging
• The status of the uterus should be re-evaluated after exposure to exogenous estrogen ≥ 3 months
• Misdiagnosed infertility can cause psychological burden and limit future fertility options
• Monitor for low bone marrow density

Selected References
6. Michala L, Aslan N. The clandestine uterus: or how the uterus escapes detection prior to puberty. BJOG. 2010; 117: 212-5.