



Ovarian Insufficiency: The Hidden Uterus

Biwen Cheng, MD and Chao-Hsu Lin, MD
Department of Pediatrics, Division of Endocrinology
Mackay Memorial Hospital, Hsinchu, Taiwan



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(40thpercentile), 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

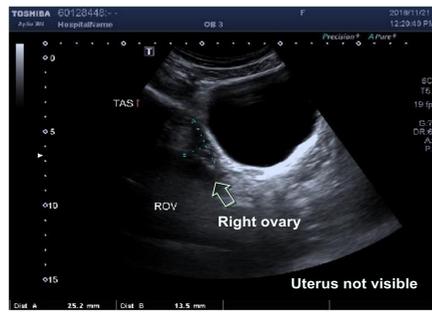
Magnetic Resonance Image



1. Absence of uterus and ovaries
2. Visible bladder

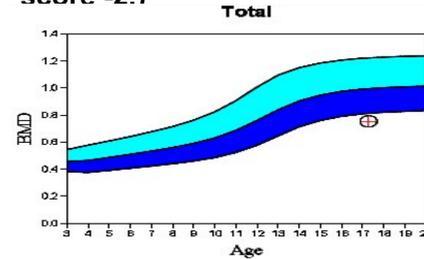
Impression: **Absence of uterus.**

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**

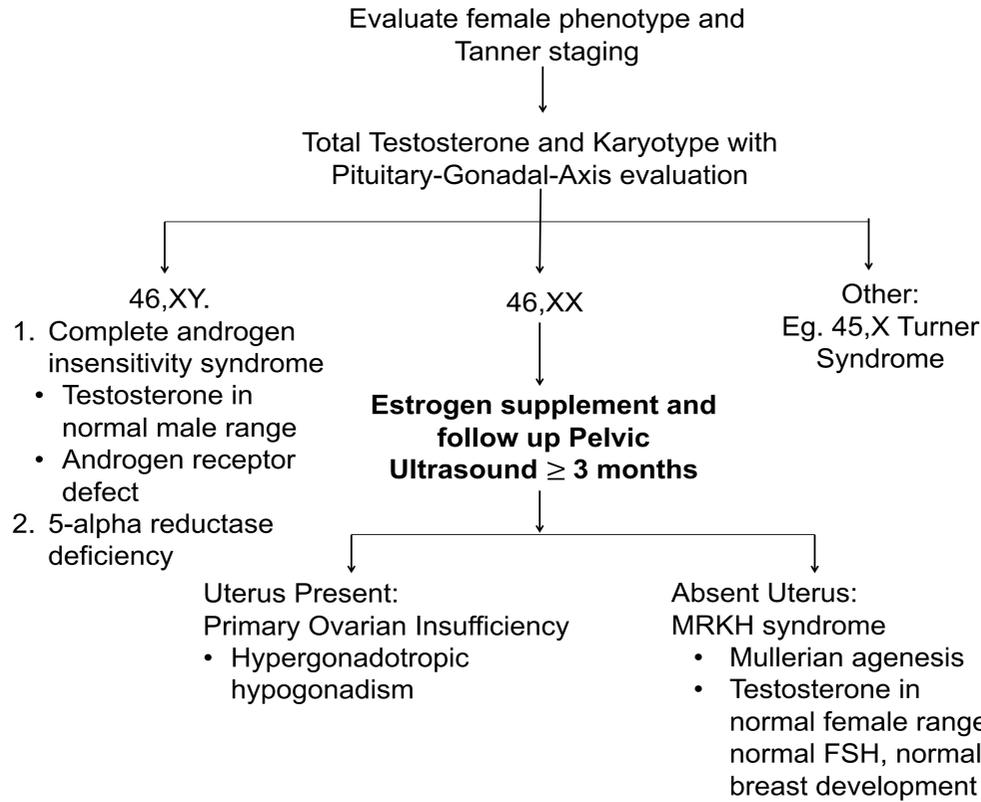


Treatment and Result

- Conjugated estrogen (0.625mg) for 3 months with calcium supplement
- Follow up pelvic ultrasound 3 months later: **uterus visible**
- Add medroxyprogesterone (5mg) with estrogen monthly



Primary Amenorrhea with Absent Uterus



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 \geq 3 months
- Misdiagnosed infertility can cause psychological burden and limit future fertility options
- Monitor for low bone marrow density

Selected References

- 1.S.Salardi LFO, E. Cacciari. Pelvic ultrasonography in premenarcheal girls: relation to puberty and sex hormone concentrations. Archives of Disease in Childhood. 1985; 60: 120-5.
- 2.Akierman SV, Skappak CD. Turner Syndrome and apparent absent uterus: a case report and review of the literature. J Pediatr Endocrinol Metab. 2013; 26: 587-9.
- 3.Bannink EM, van Sassen C. Puberty induction in Turner syndrome: results of oestrogen treatment on development of secondary sexual characteristics, uterine dimensions and serum hormone levels. Clin Endocrinol (Oxf). 2009; 70: 265-73.
- 4.Berglund A, Burt E. A critical assessment of case reports describing absent uterus in subjects with oestrogen deficiency. Clin Endocrinol (Oxf). 2019; 90: 822-6.
- 5.Bialka A, Gawlik A. Coexistence of Mayer-Rokitansky-Kuster-Hauser Syndrome and Turner Syndrome: A Case Report. J Pediatr Adolesc Gynecol. 2016; 29: e35-8.
- 6.Michala L, Aslam N. The clandestine uterus: or how the uterus escapes detection prior to puberty. BJOG. 2010; 117: 212-5.
- 7.Bousfiha N, Errahay S. Gonadal Dysgenesis 46, XX Associated with Mayer-Rokitansky-Kuster-Hauser Syndrome: One Case Report. Obstetrics and Gynecology International. 2010; 2010: 1-3.
- 8.Mulayim B, Celik NY. Pubertal failure and primary amenorrhea with uterine hypotrophy due to hypogonadotropic hypogonadism. J Pediatr Adolesc Gynecol. 2009; 22: e1-3.
- 9.Rousset P, Raudrant D. Ultrasonography and MRI features of the Mayer-Rokitansky-Kuster-Hauser syndrome. Clin Radiol. 2013; 68: 945-52.
- 10.Yollin E, Jonard S. Delayed puberty with extreme uterine hypotrophy: do not conclude too early to the absence of the uterus. Gynecol Obstet Fertil. 2006; 34: 1029-35.

