Isolated Growth Hormone Deficiency and Amenorrhea – Case Report

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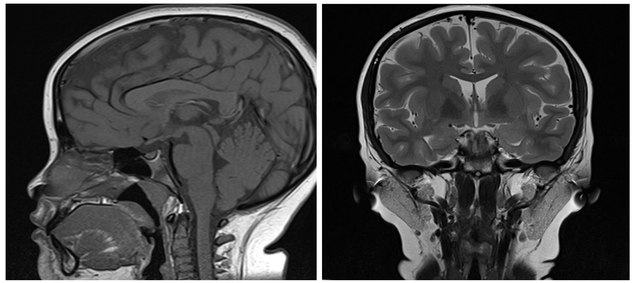
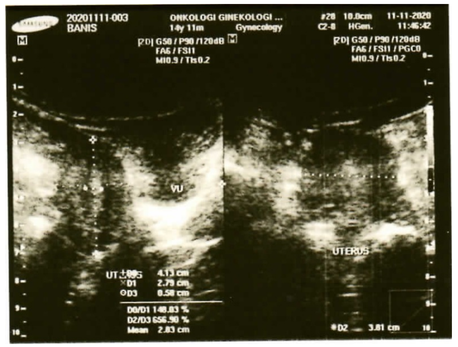
Objective: Hypopituitarism may affect a single or multiple hormones. Its phenotype depends on the affected hormone(s) deficiency. Growth hormone (GH) deficiency is the most common. We wish to highlight its diagnosis and management challenges in a developing country.

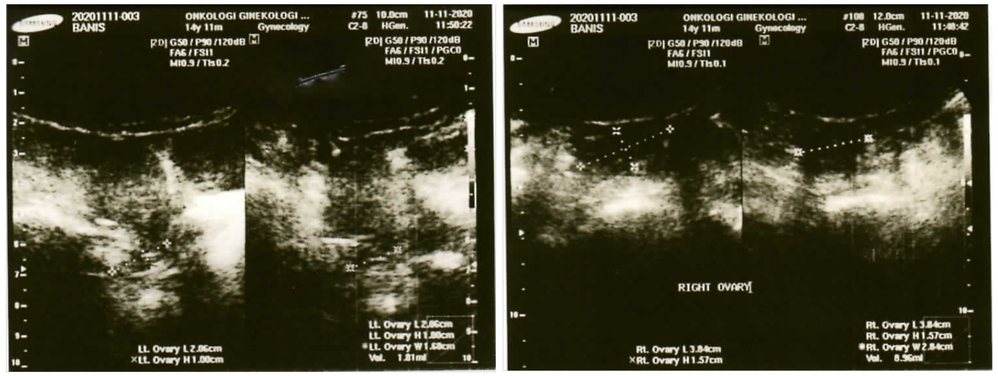
Case Report: A 14-year-old came to the pediatric gynaecology clinic with a chief complaint of inability to menstruate. Menarche was at 12, lasted 7 days, soaking 2 pads/day which discontinued shortly after. Thelarche was at 12 and her breast is currently at Tanner stage 4. Her axillary and pubic hairs are at Tanner stage 1. Her height was 120 cm, weight 34.8 kg, height for age z-score <-3. Her midparental height is 156 ± 8.5 cm. Her mother noted that she was shorter than her peers. No abnormalities on abdominal examination and external genital examination. Her lab results were normal for estradiol, luteinizing hormone, follicle-stimulating hormone, and prolactin levels. Bone age was suitable for her age. Magnetic resonance imaging (MRI) revealed pituitary gland hypoplasia (5.3 mm). She had undergone a hormonal panel testing 3 years prior with abnormally low GH level but normal cortisol and thyroid hormone levels. Her karyotype is 46, XX. She was diagnosed with isolated growth hormone deficiency (IGHD) with delayed puberty. She was subsequently treated with medroxyprogesterone tablets once daily, after which menstruation restarted. However, due to her economic background, she discontinued her medication and amenorrhea recurred. She also declined further genetic tests citing the above reason.



MRI: pituitary gland hypoplasia (5.3 mm)



Abdominal ultrasound showing uterus with homogenous density 4,02 cm x 2,61 cm x 2,60 cm. Endometrial line (+) with 8,0 mm in diameter, body to cervix ratio 2:1.



A B

Abdominal ultrasound showed the left ovary (A) with a size of 2.51 cm x 1.47 cm and the right ovary (B) with a size of 2.43 cm x 1.41 cm.

Discussion: Amenorrhea present after a brief menarche should alert gynaecologists of a possible multi-hormone disorder with an underlying structural abnormality. A battery of hormonal tests examining all the pituitary hormones should be undertaken. IGHD may be due to a structural abnormality, such as pituitary gland hypoplasia and this finding should alert the clinician of other possible midline defects. Unfortunately, economic reasons prevented the patient from consenting to genetic tests to identify the underlying IGHD mutations. Similarly, they also prevented her from adequate hormonal treatment with it not covered under the current Indonesian national health insurance.

Conclusion: IGHD is a common childhood disorder which may present with a gynaecological complaint, such as amenorrhea. Hormonal tests should be undertaken, along with MRI and genetic tests to identify the specific hormonal deficiency. GH supplementation is the treatment of choice. Early diagnosis and hormonal treatment enhance her reproductive prognosis.

Disclaimer: This abstract has been presented previously at the Annual Update in Paediatric and Adolescent Gynaecology; Joint RCOG/BRITSPAG Meeting on 21-22 March 2022