Congenital Pituitary Stalk Deformity

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Case Presentation:

A 20 year old female with a history of diabetes mellitus presents with primary amenorrhea and normal secondary sexual characteristics. No associated headaches, visual changes, galactorrhea, or hirsuitism. She denies sexual activity, never used OCPs or progesterone, and has no history of brain trauma. Gynecological exam revealed Tanner stage IV with normal external genitalia. Internal exam was declined. Pregnancy test was negative. Laboratory work showed FSH 3.9, estradiol <18, LH 3.7, Prolactin 7.3, free testosterone <12. Thyroid studies, IGF-1, DHEA, 17-(OH) progesterone, and 24-hour urine cortisol were normal. A pelvic ultrasound showed normal ovaries and uterus. Radiographic bone age was that of an 18 year old. Karyotyping resulted in 46 XX. This patient was diagnosed with hypogonadotrophic hypogonadism. An MRI brain was done to look for an etiology (see images). What is the diagnosis?

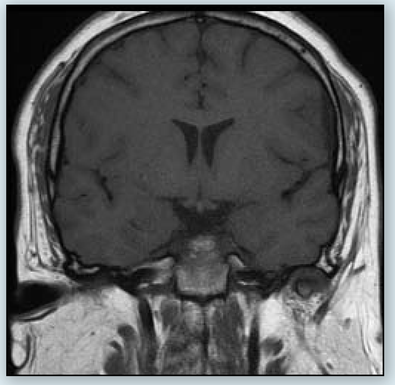

Figure 1. T1 pre contrast.

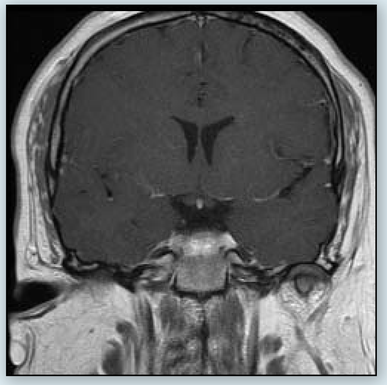

Figure 2. T1 post contrast.

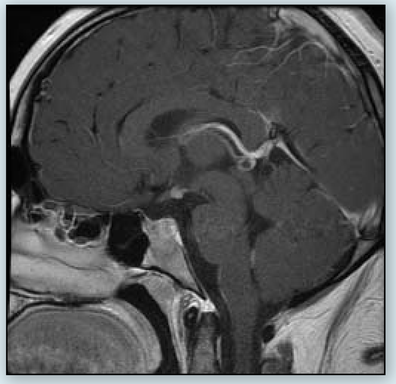

Figure 3. Lateral view.

Answer:

The imaging displays hypoplasia of the pituitary gland without any visible stalk. There is a focus of enhancing tissue at the midline of the median eminence, posterior to the optic chiasm. This may reflect ectopic posterior pituitary tissue. Findings, along with clinical history, conclude that this patient has pituitary stalk transection syndrome with probable ectopic posterior pituitary gland. This syndrome involves a small anterior pituitary gland, absent or ectopic high signal intensity of posterior pituitary, with absence or hypoplasia of the infundibulum.[1,2,3] The mechanism of abnormality is unclear, but proposed theories are birth trauma, perinatal anoxia, and disordered embryogenesis.[2] It is important to rule out acquired causes such as weight loss, anorexia nervosa, stress, heavy exercise, or hyperprolactinemia. The deficiencies vary, but gonadal insufficiency can be seen in adults.[4] This patient developed hypogonadotrophic hypogonadism with low FSH and LH, resulting in low estrogen production from a congenital structural deformity of her pituitary stalk.

References:

1. Kulkami, Chinmay. Pituitary stalk transection syndrome: Comparison of clinic-radiological features in adults and children with review of literature. Indian J Radiol Imaging. 2012 Jul-Sep; 22(3): 182-185.
2. Rottembourg, Diane, et al. Gonadotrophic status in adolescents with pituitary stalk interruption syndrome. Clinical Endocrinology. 2008; 69: 105-111.
3. Pinto, G, et al. Pituitary Stalk Interruption Syndrome: A Clinical Biological-Genetic Assessment of Its Pathogenesis. Journal of Clinical Endocrinology and Metabolism. 1997; 82(10): 3450-3454.
4. Moore, W. Tabb, and Richard C. Eastman. "Acromegaly; Prolactin and Gonadotropin-secreting adenomas; Hypopituitarism." Diagnostic Endocrinology, 2nd edition, Ed. Stephanie Manning, James F. Shanahan. Missouri: Mosby, 84-92. Print.