A case report of Hypogonadotropic hypogonadism associated with empty sella.

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Abstract:  
We describe a 20 year old male who presented with complaints of failure to attain puberty, break in voice, underdeveloped genitalia and testicular atrophy. He had eunuchoid body proportions with a height of 150 cms and arm span of 157 cms, a lower segment of 84cms, a upper segment of 66cms. There was no anosmia or midline defects. MRI brain showed empty sella with ectopic posterior pituitary and CSF collection surrounding the gland. His laboratory investigations showed low luteinizing hormone (LH), follicle stimulating hormone (FSH) and testosterone levels. Conclusion - Uncommon presentation of isolated gonadotropin deficiency secondary to pituitary cause leading to delayed puberty.  
Keyword: Empty sella, gonadotropin, hypogonadism, delayed puberty.

Introduction:  
Empty sella syndrome is the pathological variant of radiologically verified empty sella. Empty sella syndrome may be associated with abnormal pituitary function. Low levels of growth hormone (GH) are the most common, but other pituitary hormones may be deficient. In this article, we are describing a patient with hypogonadism with isolated gonadotropin deficiency associated with empty sella.  
Case report:  
The patient was a 20 year old male who presented with complaints of failure to attain puberty, break in voice, underdeveloped genitalia and testicular atrophy. There was no history of parental consanguinity. He had no history of erection, ejaculation, or nocturnal emissions. There was no history of trauma, visual disturbances, loss of consciousness, or convulsions. He was the eldest among 2 siblings and there was no family history of similar complaints. No history of loss or gain in weight. No history of continuous drug intake in the past. He had eunuchoid body proportions with a height of 150 cms and arm span of 157 cms, a lower segment of 84cms, a upper segment of 66cms. There was no anosmia or midline defects. An MRI of brain showed empty sella with ectopic posterior pituitary and CSF collection surrounding the gland. There was no focal lesions in the gland or space occupying lesions.
His karyotype is 46,XY. The patient had a micropenis (length is 4 cms) and absent pubic hair. Testis size (Right- 2.2 X 1.7; Left - 2.0 X 1.5) with sensations preserved. Patient had scanty facial hair with absent axillary and chest hair. Vital signs and systemic examination were normal. His laboratory investigations were as follows (normal ranges for males in brackets): serum testosterone =62.45 ng/dl (241-827); LH- 0.2 IU/L (1.5-9.3); FSH- 0.2 IU/L (1.4-18.1); S.Prolactin-8.63 ng/ml (2.1-17.7) S. estradiol< 20 IU/L TSH, free T3, free T4, Growth hormone, Serum cortisol, Serum ferritin were within normal limits. A peripheral smear showed microcytic hypochromic anemia, platelets were adequate. A total and differential counts of white blood cells (WBCs) was also within normal ranges. Hemoglobin level was 11.3 g/dl. No signs of hemochromatosis. Patient is being treated with testosterone ecanoate 250 mg IM given every 3 weeks. He is being followed up regularly. He will be switched over to HCG injection 2000 U thrice a week once he develops secondary sexual characteristics and will be followed with periodical semen analysis.

Discussion:
Delayed puberty is defined as the lack of initial signs of sexual maturation by 13 years in girls and 14 years in boys. One of the causes is hypogonadotropic hypogonadism due to deficient secretion of gonadotropin due to hypothalamic or pituitary disorders. Classic kallmann syndrome and idiopathic hypogonadotropic hypogonadism are rare genetic syndromes that encompass the spectrum of isolated hypogonadotropic hypogonadism. Most patients have a GnRH deficiency as suggested by their response to pulsatile GnRH therapy. Hypothalamic–pituitary function is otherwise normal in most patients, and hypothalamic–pituitary imaging reveals no space occupying lesions. By definition either anosmia or hyposmia is present in patients with kallmann syndrome, in contrast to patients with
idiopathic hypogonadotropic hypogonadism, whose sense of smell is normal. In our patient features were not typical of kallmann syndrome though he had isolated gonadotropin deficiency. The cause of gonadotropin deficiency may be pituitary hypoplasia as suggested by imaging studies, which showed empty sella and pituitary hypoplasia. But the isolated deficiency of gonadotropins with other pituitary hormones being normal is the rare presentation in this patient, which could not be explained well by radiological findings alone. As empty sella is found in normal patients also, the isolated deficiency of gonadotropin observed in our patient could be due to other causes like genetic defects in GnRH secretion or action. GnRH could not be analysed in the present patient due to technical and financial reasons. Empty sella is a radiological diagnosis based on CT or MRI investigation. The radiological diagnosis does not mean a pathological situation in every instance. Many patients present without specific symptoms and the diagnosis is made by chance. Endocrine abnormalities are not a common occurrence. Hyperprolactinemia occurs rarely possibly due to stalk stretching or coincidental microprolactinomas. The growth hormone secretory reserve is often abnormal in these patients probably as a result of obesity. Occasionally thyrotropin and gonadotropin deficiency is noted. Rarely empty sella is associated with hormone excess possibly due to microadenoma within the compressed gland. Spontaneous CSF rhinorrhea and pseudotumour cerebri are two syndromes occasionally associated with an empty sella. Children with empty sella most commonly have growth hormone deficiency, although other pituitary hormone dysfunctions can occur. Secondary empty sella turcica is associated with iatrogenic events such as surgery, radiation, or both, or with non iatrogenic diseases such as infarction or infection of pituitary gland. Although testosterone therapy is sufficient for maturation and maintenance of secondary sexual characteristics in hypogonadal men, gonadotropins are required for stimulation of spermatogenesis. The prognosis for successful stimulation of spermatogenesis in men treated with HCG is good and sufficient to both initiate and maintain spermatogenesis in some patients even when FSH level is very low.

References:


