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The Triad - MRI Insights Into Short Stature and Delayed Puberty

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Case Description

A 15-year-old boy was brought to the endocrine clinic due to concerns about short stature and poor development of secondary sexual characteristics. He was an otherwise healthy child, born to non-consanguineous parents with a normal birth history and immediate postnatal period.

On examination, his height was plotted, and it was well below the lower limit of the mid-parental height range. His secondary sexual characteristics were poorly developed: testicular volume was 1ml on both sides with a pubertal stage Tanner 1. Visual field examination was normal. There were no facial features to suggest congenital syndromic conditions or midline defects. However, there was very clear midfacial hypoplasia. Biochemical evaluation including the Insulin Tolerance Test (ITT) revealed panhypopituitarism with involvement of hypothalamo-pituitary - adrenal, thyroid, gonadal axes and severe growth hormone deficiency. There was no evidence of posterior pituitary involvement. The patient was started on hormone replacement therapy.

Visual perimetry (60-4) was found to be normal. The x-ray of the hand with the wrist joint showed a delayed bone age compatible with 11 years. (Figure 1.)

The images of MRI-brain with pituitary cuts were as follows. (Figure 2.)

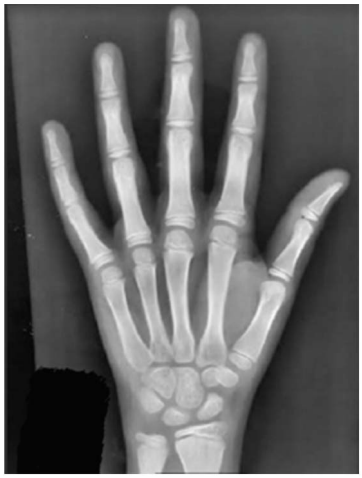

Figure 1

Name the abnormalities that can be identified in the above MRI images of this patient in the regions marked by the letters A & B and C.

A & B - Hypoplastic anterior pituitary gland, absent pituitary stalk and absence of normal posterior pituitary bright spot. (T2-Weighted MRI)

C - Ectopic posterior pituitary as indicative of bright spot in the floor of the third ventricle or hypothalamus. (T1-Weighted MRI)

What is the diagnosis?

Pituitary Stalk Interruption Syndrome (PSIS).

Pituitary Stalk Interruption Syndrome (PSIS) is a very rare disorder characterized by varying degrees of hypopituitarism with a typical triad of MRI features, including the hypoplastic or aplastic anterior pituitary gland, thinned or absent pituitary stalk **(A & B)** and the ectopic location of posterior pituitary gland indicated by the ectopic location of the bright spot. Here, it was at the base of the hypothalamus **(C)**. The pathogenesis is mainly related to genetic mutations and possible perinatal stress at the time of delivery, particularly during the delivery of breech presentation [1,2]. Some of the common genetic mutations involving in PSIS are PIT1, PROP1, HESX1, LHX3/4, PROKR2, TGIF and SOX3 [1,3].

The clinical presentations can vary based on the degree of pituitary involvement and the extent of hormonal deficiencies. There can be isolated or multiple hormonal deficiencies with preserved posterior pituitary function. Usually, the clinical features due to growth hormone deficiency and hypogonadotropic hypogonadism are the initial presentations as in this patient. Early diagnosis and prompt commencement of hormonal replacement therapy are crucial to improve the quality of life.

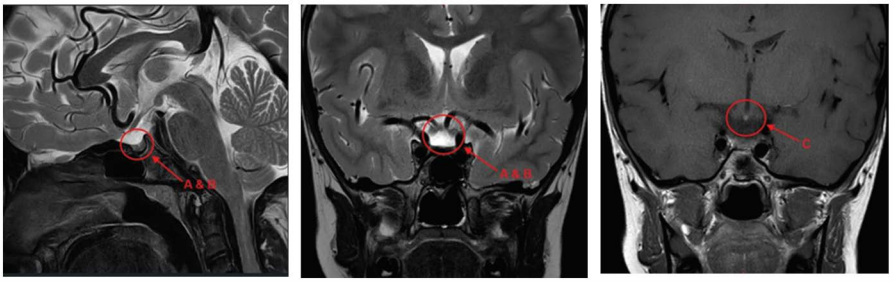

Figure 2

References

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