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Case Report

Late-Onset Pituitary Stalk Interruption Syndrome

Ali Mohamed D[1]*, Ahmed A[2], Fatima Zahra I[1], Motassim Billah N[1] and Nassar I[1]

[1]Department of Central Radiology, University Hospital Center Ibn Sina, Rabat, Morocco

[2]Department of Endocrinology and Metabolic Diseases, University Hospital Center Ibn Sina, Rabat, Morocco

***Corresponding author:** Daoud Ali Mohamed, Department of Radiology, University Hospital Center Ibn Sina, Rabat, Morocco

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Case Presentation

A 21-year-old woman of low socio-economic status and no schooling consults for delayed puberty and primary amenorrhea. Her history includes a home delivery with no reported neonatal suffering.

Examination revealed good general condition, normo-colored conjunctiva, statural delay at -3DS, absence of secondary sexual characteristics S1P1 of Tanner staging. Assessment of pituitary hormones showed collapsed GH and gonadotropin (LH and FSH) levels. Other hormones, including TSH/FT4 and ACTH, were normal.

In view of the low FSH and LH levels, pituitary MRI was performed and showed an interruption of the pituitary stem with hypoplastic antehypophysis was about 2mm and pituitary stalk was interrupted (Figure 1 and 2). The patient received hormone replacement therapy.

Discussion

Pituitary stalk interruption syndrome (PSIS) is a congenital pituitary anatomical defect. It is characterized by the triad of thin or interrupted pituitary stalk, absent or ectopic posterior lobe, and hypoplastic or aplastic anterior lobe. Moreover, the condition is considered rare. The diagnosis is confirmed based on clinical features, endocrine evaluation results, and findings on contrast-enhanced MRI, which is the technique used to assess pituitary shape, size, and microstructure in patients with PSIS. c. The age of the patients varies with an average age at the time of diagnosis is 9.4 ± 11.6 years [1]. However, the typical characteristics include permanent anterior pituitary hormone deficiencies during the pediatric age, which appear gradually and progress to panhypopituitarism during adulthood.

The incidence of breech delivery, cesarean section, and neonatal distress are high in the PSIS population [5]. Breech delivery leads to deformation of the head, which can result in injury to the pituitary stalk. Similarly, hypoxemia due to anoxia after birth may also cause injury of the pituitary stalk and pituitary [5]. Genetic mutations are thought to contribute to less than 5% of PSIS cases. Mutations detected were in HESX1, LHX4, SOX3, PROKR2, and OTX2 genes.

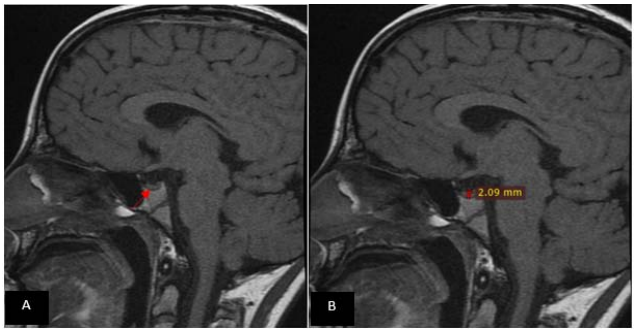

Figure 1: Pituitary magnetic resonance imaging (MRI) showed hypoplastic antehypophysis (red arrow) on sagittal T1 Flair (A and B).

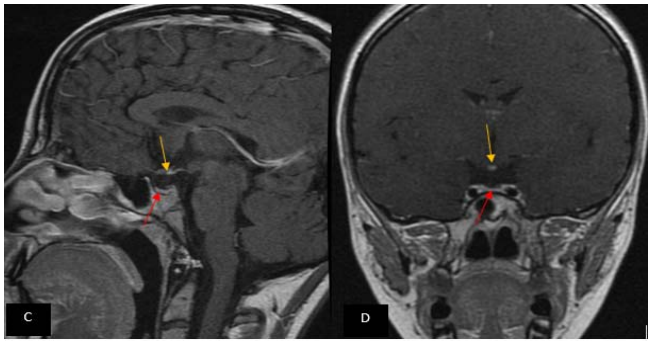

Figure 2: After injection (C and D), the pituitary stalk was interrupted (yellow arrow). The optic chiasm was not compressed, and the bilateral cavernous sinus had a clear structure.

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