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Endoped Abstract

Pituitary Stalk Interruption Syndrome: Baseline Characteristics and Long-Term Evolution of Two Pediatric Cases with Rare Presentation

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Background and Aims

Pituitary stalk interruption syndrome (PSIS) is characterized by the presence of a thin or absent pituitary stalk in association with a hypoplastic or aplastic anterior pituitary or an ectopic neurohypophysis [1,2]. The phenotype associated with PSIS has a high incidence of various combinations of hormonal deficiencies, sometimes associated with extra-pituitary birth defects [1]. The aim of our study is to illustrate the heterogeneity of PSIS through a complete description of the clinical, biological and imagistic differences between our cases.

Case Reports

We describe two rare pediatric cases. The first case was a 6-year-old boy admitted to our department with polyuro-polydipsic syndrome and underdeveloped external genitals. He had a history of neonatal micropenia and a family medical history of panhypopituitarism and midline malformations. Physical examination revealed normal stature (height: 117 cm, −0.4 SDS), obesity (BMI: 25.2 Kg/mp, over 97th percentile), micropenia (stretched penile length: 3 cm) and non-palpable testes. Initially, his urine output was 5000 mL/24 h, with a low urine specific gravity of 1005–1010, and no increase after water deprivation test, with an appropriate response after desmopresin administration. He presented normoglycemia. Serum gonadotropin (FSH, LH) and testosterone levels were low, with no response after GnRH-analogue stimulation test. IGF-1 levels were low-normal. His Copeptin value was decreased. Pituitary MRI showed anterior pituitary hypoplasia, absence of the pituitary stalk and neurohypophysis.

The second case was a 4-year-old girl referred to our service for growth retardation. She had a history of neonatal prolonged jaundice and intermittent seizures, for which she was prescribed phenobarbital for 2 years. Her physical examination described short stature (height: 90.5 cm, −3.3 SD), underweight (BMI: 15.87 Kg/m^2, 15–25th percentile), head dysmorphic features: medial epicanthal fold, depressed nasal bridge, anteverted nares, ogival palate. She had a bone age of 2 years. The initial evaluation diagnosed severe GH deficiency, and during the next 3 years of follow-up she developed ACTH, TSH and FSH-LH deficiencies. MRI examination indicated anterior pituitary hypoplasia, absence of pituitary stalk and ectopic neurohypophysis.

Discussion

Central diabetes insipidus is rarely reported in patients with PSIS (0–4%) [1,3]. Although GH deficiency was found almost ubiquitous in PSIS, our first patient had no growth retardation. Normal linear growth despite abnormal GH secretion was also reported [4]. Neonatal cholestasis was linked to ACTH deficiency, which can also be a cause of recurrent hyponatremia [1]. There were no signs of ACTH deficiency or hyponatremia in our second patient in the neonatal period. Few reports showed cases of PSIS that were diagnosed with acute epileptic seizures accompanied by hyponatremia [5]. The rapidly progressive evolution to combined pituitary deficiency observed in PSIS cases [4] with neonatal features was also recognized in our patients.

Conclusions

The phenotype associated with PSIS can be highly heterogeneous. It is important to increase the awareness of PSIS, especially in the neonatal period when the features of PSIS can be easily overlooked. We also emphasize the importance of patient follow-up as an essential part of an adequate management.

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