Introduction

Pituitary stalk interruption syndrome (PSIS) is a rare (incidence, 0.5/1,000,000 births) congenital abnormality of the pituitary. It is characterized by the triad:

• a very thin or interrupted pituitary stalk
• an ectopic (or absent) posterior pituitary (EPP)
• hypoplasia or aplasia of the anterior pituitary

Typical features are tertiary hypothyroidism, hyperprolactinemia and other pituitary hormone deficiencies. However, in rare cases hyperprolactinemia have also been reported.

Aim

In this report, we describe the clinical and hormonal characteristics of a patient with PSIS who was referred for short stature and primary amenorrhea.

Case Report

A 16-year-old girl was referred for short stature and primary amenorrhea. She has been well with no chronic medical conditions, no hospitalizations, and no surgeries. Parents were not related. She was born at term after an uneventful pregnancy by normal vaginal delivery with head presentation. Her birth weight was 3000 g. She did not have a history of birth asphyxia. There was no family member with short stature, delayed puberty or amenorrhea.

On physical examination:
Height: 150.2 cm (-1.49 SDS),
Weight: 70.6 kg (1.63 SDS),
Target height: 161 (0.17 SDS),
Tanner's stage: M3P3A+, no dysmorphic features.

Results

Karyotype: 46,XX

- FSH: 6.11 mIU/ml
- LH: 2.95 mIU/ml
- E2: <5 pg/ml
- LHRH test:
  - peak LH: 9.3 mIU/mL
  - peak FSH: 8.5 mIU/mL
- PRL: 1.66 ng/ml (4.79-23.3)
- β-hCG: negative

- TSH: 5.25 uIU/ml (0.51-4.30)
- fT4: 0.97 ng/dl (0.98-1.63)
- Cortisol: 13.4 mcg/dl
- IGF-1: 78.2 ng/ml (226-903)

GH stimulation tests
- Clonidine peak GH: 0.14 ng/ml
- L-DOPA peak GH: 0.22 ng/ml

Brain MRI:
- Anterior pituitary hypoplasia
- Ectopic posterior pituitary
- Absent pituitary stalk

Central hypothyroidism, hyperprolactinemia, normogonadotropic amenorrhea, growth hormone deficiency

Pituitary Stalk Interruption Syndrome

Conclusion

The exact cause of PSIS is still unknown. Perinatal injuries, defective organogenesis or rare mutations of HESX1, LHX4, OTX3 and SOX3 are proposed to be the cause of PSIS in familial cases. Despite the fact that PSIS is a rare disorder, which typically presents with hyperprolactinemia, it should always be considered in the differential diagnosis of a patient with normogonadotropic amenorrhea and/or hyperprolactinemia.