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Rare case of pituitary stalk interruption syndrome diagnosed in an adult

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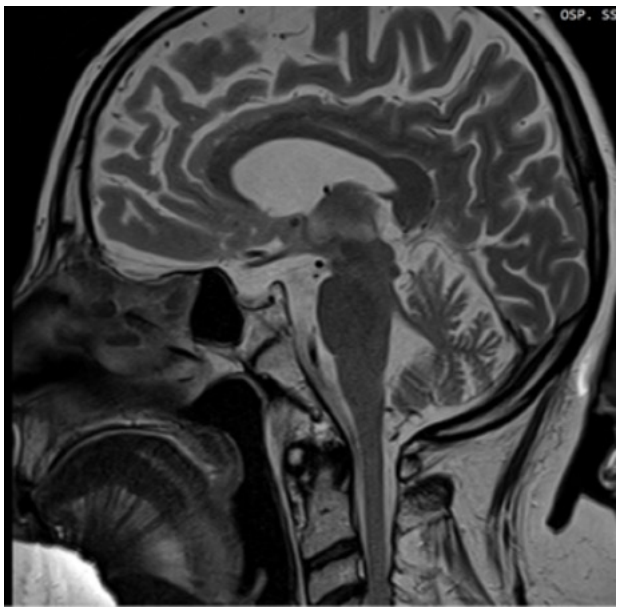
Introduction

Pituitary stalk interruption syndrome (PSIS) is a rare disorder with a reported incidence of 0.5/100,000 live births. Cases are usually sporadic, rarely familial (5%) and the aetiology remains elusive. PSIS is characterized by the combination of specific findings in magnetic resonance imaging (MRI) including an absent or ectopic posterior pituitary, absent or interrupted pituitary stalk and anterior pituitary hypoplasia but sometime this triad can be incomplete and the age at diagnosis ranges from newborn to adult. The consequences of PSIS are a series of anterior pituitary deficiencies that may be isolated or may be associated with other hormonal deficiencies. PSIS that presents later in life appears gradually and generally progresses in adulthood to panhypopituitarism. The diagnosis mainly depends on the deficiency of hormones and a typical abnormality of the pituitary gland as revealed by magnetic resonance imaging (MRI).

Case presentation

A 55-year-old male was admitted for acute respiratory failure.
Remote pathological history: paraplegia secondary to polio; hypothyroidism; permanent atrial fibrillation; chronic pulmonary heart; rib cage malformation; cholelithiasis; previous pericarditis.
During the hospital stay, the patient received intravenous therapy (diuretics and steroids) and oxygen therapy with progressive clinical improvement.
However, due to the presence of underdeveloped sexual characteristics on physical examination, the patient was subjected to further diagnostic examinations.



Diagnostic examination

Hematochemical examinations: pituitary hormone deficiency (FSH, testosterone, somatomedin C igf-1)
Scrotal ultrasonography: testicles retained in the inguinal canal, of partially preserved morphology, hypoplastic, hypoechogenic with bilaterally inhomogeneous echogenicity
DEXA bone mineralometry of the femur and spine: examination suggestive of severe osteoporosis
pituitary MRI : finding of adenohypophysis clearly smaller than normal (cranial-caudal diameter 1-2 mm); absent pituitary peduncle; presence of ectopic neurohypophysis. alterations compatible with pituitary stalk interruption syndrome.

DXA Results Summary:

Region	Area (cm^2)	BMC (g)	BMD (g/cm^2)	T-score	PR (%)	Z-score
L1	10.19	3.90	0.383	-5.7	38	-5.2
L2	14.52	8.16	0.562	-4.8	51	-4.3
L3	16.03	8.37	0.522	-5.3	47	-4.8
L4	16.71	6.22	0.372	-7.0	32	-6.5
Total	57.44	26.64	0.464	-5.7	43	-5.2

IGF1 (ng/ml)	27,4	(N.V. 54-167)
FSH (mUI/ml)	0,18	(N.V. 1-8)
LH (mUI/ml)	0,24	(N.V 2-12)
PRL 9,6 (ng/ml)	9,6	(N.V. 1,6-19)
Testosterone (ng/ml)	<20	(N.V. 220-1730)
TSH (mUI/ml)	0,065	(N.V 0,4-4,7)
FT4 (pg/dl)	0,69	(N.V. 0,7-2)

Treatment

Treatment was based on substituting hormones with lifelong poly-hormonal replacement therapy.
Glucocorticoid replacement was the initial treatment, followed by testosterone gel. Also calcium tablets, calcitriol, and denosumab were used for the management of osteoporosis. Nevertheless, due to the onset of SARS-CoV-2 pneumonia, the patient died and could not be followed up.

Conclusions

PSIS is a rare condition whose aetiology remains unknown. However, this disease might be attributed to gene–environment interactions.

The early diagnosis of PSIS is based on MRI findings. Then, prompt hormonal replacement therapies are provided. The diagnosis of this patient was delayed, due to his poor conditions, and the prognosis was difficult to predict. Therefore, the understanding of PSIS in clinical practice, particularly early diagnosis and timely treatment among newborn or children, should be assured.