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CASE REPORT Open Access

Pituitary stalk interruption syndrome (PSIS) presenting in a Jordanian male patient with short stature and delayed puberty: a case report

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Abstract

Background Pituitary stalk interruption syndrome is a rare congenital pituitary anatomical defect manifested with wide and various clinical presentations. Short stature and delayed puberty are present in most cases and may be combined with extra pituitary malformations. Magnetic resonance imaging is considered the key factor for reaching the definite diagnosis as it reveals the different radiological presentations of this syndrome.

Case presentation A 17-year-old male patient from Jordan was presented to the radiology department for pituitary MRI. The patient presents with multiple pituitary hormonal deficiency, short stature, and under-developed secondary sexual characteristics. The bone age of the patient was significantly less than the chronological age. MRI pituitary gland showed hypoplasia of the anterior pituitary, absence of the pituitary stalk and ectopic posterior pituitary gland located posterior to the optic chiasm. These findings were consistent with pituitary stalk interruption syndrome.

Conclusions Considering uncommon incidence of pituitary stalk interruption syndrome, magnetic resonance imaging of the pituitary should be considered while examining a patient with pituitary deficiency for prompt diagnosis and treatment. Hormone replacement medication is the primary treatment for pituitary stalk interruption syndrome and should be started as soon as feasible. Thus, early diagnosis and monitoring of individuals are crucial.

Keywords Pituitary stalk interruption syndrome (PSIS), MRI, Hypopituitarism, Short stature, Case report

Background

Pituitary stalk interruption syndrome (PSIS) is a rare congenital anatomical defect of the pituitary gland with an estimated incidence of 0.5/100,000 live births and with male predominance [1–3]. PSIS is characterized by the triad of thin or absent pituitary stalk, ectopic or absent posterior pituitary, and hypoplasia or aplasia of the anterior pituitary [4].

First reported case of PSIS was in 1987 by Fujisawa et al. [5]. While the etiology and pathogenesis of PSIS are not yet fully understood, high association with birth trauma leading to perinatal pituitary injury has been reported, considering breech or footling presentation, dystocia and cesarean section [3, 4, 6]. In addition, abnormal early pituitary development caused by various genetic mutations has been reported in less than 5% of cases [1, 7, 8].

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Magnetic resonance imaging (MRI) is the key factor for reaching the definite diagnosis as it reveals the different radiological presentations of PSIS, concerning the anterior lobe (absent, hypoplastic, normal), the posterior pituitary lobe (absent, ectopic along the stalk, ectopic at the hypothalamus base, or normal in the sella turcica) and/or the stalk (interrupted, absent, thin, or normal) [1, 9].

PSIS has wide, various clinical presentations. Growth hormone (GH) deficiency, however, is the most common feature with 100% prevalence. Other pituitary hormonal deficiencies include ACTH, LH, FSH, TSH. Prolactin, on the other hand, can be either low or increased suggesting dopaminergic pathway disconnection [2–4, 9]. Extra pituitary malformations associated with PSIS have been also reported including midline defects in the central nervous system such as septal agenesis, microcephaly, hydrocephalus, partial corpus callosum agenesis, optic nerve hypoplasia, or Arnold Chiari malformations [8, 10–12].

Case presentation

We report a 17-year-old male patient referred to the radiology department for the evaluation of the pituitary gland. The patient is a known case of short stature and delayed puberty. Birth history reports no pre-, peri- or post-natal complications. The patient was delivered by normal vaginal delivery with cephalic presentation. During his early preschool years (4–5 years), his parents noticed that he is shorter than his colleagues. At age of 6, the patient had surgery for the treatment of cryptorchidism and started to be followed up by a pediatrician, where he was discovered to have combined anterior pituitary hormonal hypo-function. At age of 9, he was started on GH replacement injection, which continued for about 8 years, this was interrupted for 6 months due to financial problems. The patient was on HRT for the thyroid gland (Thyroxine 100 mg) at age 12 years for about 1 year.

On physical examination, patient's height was 158 cm (< 5th Percentile), weight 57 kg (at 25th percentile), had small genitalia, no secondary sexual characteristics, no gynecomastia.

Laboratory data showed normal complete blood count, serum electrolytes, and normal levels of TSH (3.9 IU/ml). The patient had increased prolactin levels (21.98 ng/ml), and decreased free T4 (0.653 ng/dl), LH (0.499 mIU/ml), testosterone (< 0.025 ng/ml), and FSH (0.688 mIU/ml). Bone age of the patient was 9 years when chronological age was 14 (Fig. 1).

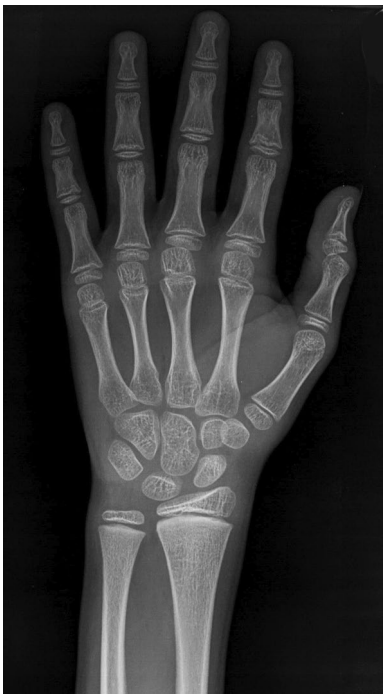

Fig. 1 Left hand frontal X-ray for a 14 years old male demonstrates that the bone age is about 9 years

MRI findings

MRI pituitary gland showed hypoplasia of the anterior pituitary, absence of the pituitary stalk and ectopic posterior pituitary gland located posterior to the optic chiasm, no associated brain abnormalities (Fig. 2).

MRI protocol

All MRI tests were carried out using a closed magnet, 1.5 T Philips Achieva MRI. A dedicated head coil was used. The sella turcica was imaged using thin slices (2 mm), T1 weighted (T1W), and T2 weighted (T2W) images in the coronal and sagittal planes. T1W sequences were repeated after administration of intravenous gadolinium with a dose of 0.1 mmol/Kg.

Conclusions

Despite the rare incidence of PSIS, MRI pituitary should be considered in patients presenting with pituitary hormonal hypo-function to ensure proper diagnosis and treatment.

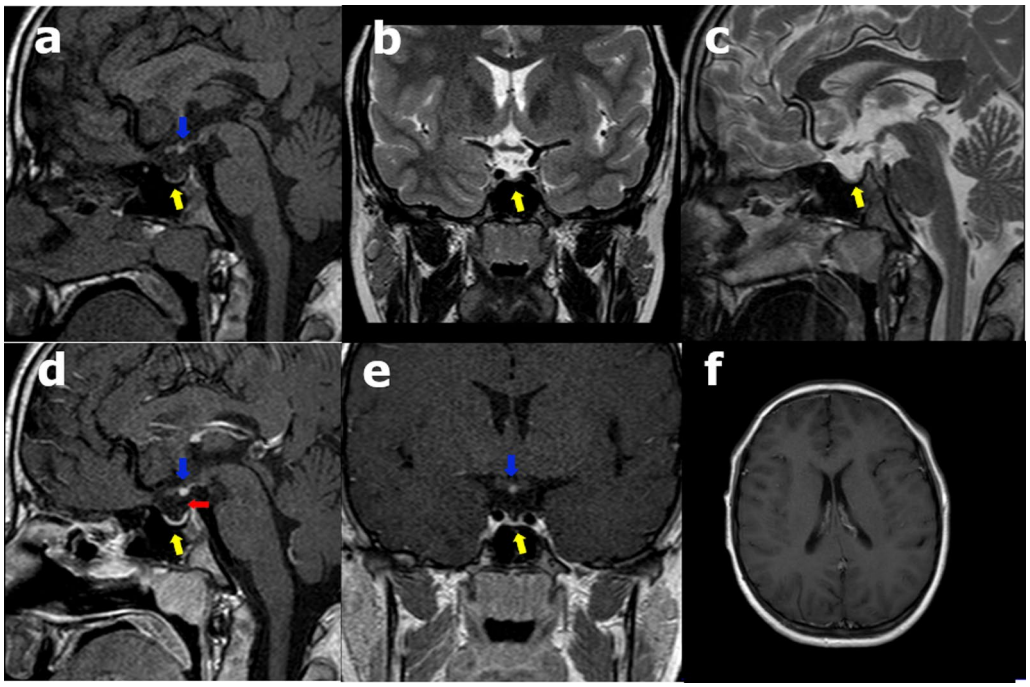

Fig. 2 MRI findings in a 17-year-old male patient presenting with short stature, delayed puberty, and hypopituitarism. **a** Sagittal non-contrast T1W image of the pituitary gland showing small anterior pituitary (yellow arrow), absent pituitary stalk, and ectopic posterior pituitary (blue arrow) seen as hyperintense focus posterior to the optic chiasm. **b** Coronal T2W image of the pituitary gland showing small anterior pituitary (yellow arrow). **c** Sagittal T2W image of the pituitary gland showing small anterior pituitary gland (yellow arrow) with absent pituitary stalk. **d** Sagittal post-contrast (IV Gd) T1W image of the pituitary gland showing homogenous enhancement of the hypoplastic anterior pituitary (yellow arrow) and homogenous enhancement of the ectopic posterior pituitary gland (blue arrow) and absent pituitary stalk (red arrow). **e** Coronal T1W post-contrast (IV Gd) of the pituitary gland showing homogenous enhancement of the hypoplastic anterior pituitary (yellow arrow) and homogenous enhancement of the ectopic posterior pituitary (blue arrow). **f**) Axial post-contrast (IV Gd) T1W brain MRI showing normal septum pellucidum and normal brain midline structures, no hydrocephalus, normal corpus callosum

Abbreviations

PSIS	Pituitary stalk interruption syndrome
MRI	Magnetic resonance imaging
GH	Growth hormone
ACTH	Adrenocorticotropic hormone
LH	Luteinizing hormone
FSH	Follicle-stimulating hormone
TSH	Thyroid stimulating hormone
HRT	Hormone replacement therapy

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Author contributions
LQM is the corresponding author and contributed to data collection, diagnosis and MRI findings. LQM and AMR were involved in manuscript writing. All authors read and approved the final manuscript.

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Availability of data and materials
All data generated or analyzed during this study are included in this published article.

Declarations

Ethics approval and consent to participate
Written informed consent was obtained from the patient's family for publication of this case report and accompanying images, and the study is approved by the ethics committee at Princess Basma Teaching Hospital.

Consent for publication
Case publication was approved; consent form was signed by the patient's family.

Competing interests
The authors declare that they have no conflicts of interest.

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References

1. Reynaud R, Albarel F, Saveanu A, Kaffel N, Castinetti F, Lecomte P, Brauner R, Simonin G, Gaudart J, Carmona E, Enjalbert A, Barlier A,

Brue T (2011) Pituitary stalk interruption syndrome in 83 patients: novel HESX1 mutation and severe hormonal prognosis in malformative forms. Eur J Endocrinol 164:457–465. https://doi.org/10.1530/EJE-10-0892

2. Bar C, Zadro C, Diene G, Oliver I, Pienkowski C, Jouret B, Cartault A, Ajaltouni Z, Salles JP, Sevely A, Tauber M, Edouard T (2015) Pituitary stalk interruption syndrome from infancy to adulthood: clinical, hormonal, and radiological assessment according to the initial presentation. PLoS ONE 10:1–12. https://doi.org/10.1371/journal.pone.0142354
3. Guo Q, Yang Y, Mu Y, Lu J, Pan C, Dou J, Lv Z, Ba J, Wang B, Zou X, Yang L, Ouyang J, Yang G, Wang X, Du J, Gu W, Jin N, Chen K, Zang L, Erickson BJ (2013) Pituitary stalk interruption syndrome in Chinese people: clinical characteristic analysis of 55 cases. PLoS ONE. https://doi.org/10.1371/journal.pone.0053579
4. Vergier J, Castinetti F, Saveanu A, Girard N, Brue T, Reynaud R (2019) Pituitary stalk interruption syndrome: etiology and clinical manifestations. Eur J Endocrinol 181:R199–R209. https://doi.org/10.1530/EJE-19-0168
5. Fujisawa I, Kikuchi K, Nishimura K, Togashi K, Itoh K, Noma S, Minami S, Sagoh T, Hiraoka T, Momoi T (1987) Transection of the pituitary stalk: development of an ectopic posterior lobe assessed with MR imaging. Radiology 165:487–489. https://doi.org/10.1148/radiology.165.2.3659371
6. Wang CZ, Guo LL, Han BY, Wang AP, Liu HY, Su X, Guo QH, Mu YM (2016) Growth hormone therapy benefits pituitary stalk interruption syndrome patients with short stature: A retrospective study of 75 HAN Chinese. Int J Endocrinol. https://doi.org/10.1155/2016/1896285
7. Brauner R, Bignon-Topalovic J, Bashamboo A, McElreavey K (2020) Pituitary stalk interruption syndrome is characterized by genetic heterogeneity. PLoS ONE 15:1–19. https://doi.org/10.1371/journal.pone.0242358
8. Karaca E, Buyukkaya R, Pehlivan D, Charng WL, Yaykasli KO, Bayram Y, Gambin T, Withers M, Atik MM, Arslanoglu I, Bolu S, Erdin S, Buyukkaya A, Yaykasli E, Jhangiani SN, Muzny DM, Gibbs RA, Lupski JR (2015) Whole exome sequencing identifies homozygous GPR161 mutation in a family with pituitary stalk interruption syndrome. J Clin Endocrinol Metab 100:E140–E147. https://doi.org/10.1210/jc.2014-1984
9. Pham LL, Lemaire P, Harroche A, Souberbielle JC, Brauner R (2013) Pituitary stalk interruption syndrome in 53 postpubertal patients: factors influencing the heterogeneity of its presentation. PLoS ONE. https://doi.org/10.1371/journal.pone.0053189
10. Wang C-Z, Guo L-L, Han B-Y, Su X, Guo Q-H, Mu Y-M (2017) Pituitary stalk interruption syndrome: from clinical findings to pathogenesis. J Neuroendocrinol. https://doi.org/10.1111/jne.12451
11. Kulkarni C, Moorthy S, Pullara S, Rajeshkannan R, Unnikrishnan A (2012) Pituitary stalk transection syndrome: comparison of clinico-radiological features in adults and children with review of literature. Indian J Radiol Imaging 22:182–185. https://doi.org/10.4103/0971-3026.107179
12. El Chehadeh-Djebbar S, Callier P, Masurel-Paulet A, Bensignor C, Méjean N, Payet M, Ragon C, Durand C, Marle N, Mosca-Boidron AL, Huet F, Mugneret F, Faivre L, Thauvin-Robinet C (2011) 17Q21.31 microdeletion in a patient with pituitary stalk interruption syndrome. Eur J Med Genet 54:369–373. https://doi.org/10.1016/j.ejmg.2011.03.001

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