

Isolated pituitary stalk duplication

Pituitary stalk duplication

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Abstract

Pituitary duplication is a rare anomaly with about 40 cases reported to date. The duplication of the pituitary stalk with a single gland is an exceptionally rare manifestation with only a few case reports in the current literature. Although incidentally discovered cases of pituitary duplication have been reported, patients with this disorder usually present with endocrine disturbance(s) regarding the adenohipophyseal axis and/or with accompanying pathologies such as median cleft face syndrome, craniocervical bony abnormalities, tuberomammillary/ pituitary mass(es) or various ocular and vascular disorders. Cross-sectional imaging modalities play a key role in the comprehensive evaluation of these patients. Here, we present a 12-year-old boy who presented with growth retardation and showed pituitary stalk duplication without a duplicated gland or associated findings.

Keywords

Pituitary Duplication; Magnetic Resonance Imaging; Pituitary Diseases

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Introduction

The pituitary gland and stalk develop from the primordium which can be distinguished on the 22nd day of gestation. The gland forms as a single structure from neural ectoderms that are tightly adhered to each other prior to the pituitary evagination. Although various mechanisms have been proposed to explain why and how pituitary duplication occurs in some cases, the exact cause remains unknown [1]. The duplication may occur at different embryonic stages, and the scope of the presentation may vary depending on the stage at which the duplication occurs. Several cases of pituitary gland duplication have been reported to date. And the vast majority of these cases were accompanied by additional abnormalities such as tuberomammillary or pituitary mass, various craniofacial midline defects or some ophthalmological pathologies [1-3]. However, cases of pituitary stalk duplication with a non-duplicated pituitary gland are extremely rare in the current literature [4-6]. Here, we present a 12-year-old boy with isolated pituitary stalk duplication who presented with growth retardation.

Case Report

A 12-year-old boy with a complaint of short stature was admitted to the hospital. His height was 119 cm, weight was 21 kg and bone age was 8 years. He was the only child of non-consanguineous healthy parents and the family history was unremarkable. He was delivered at term following an uneventful pregnancy. Except for the developmental delay, his medical history was unremarkable. No dysmorphic facial features were noted on physical examination. Neurological and ophthalmological examinations were normal. Endocrine laboratory tests revealed low serum growth hormone (GH) and insulin-like growth factor 1 (IGF-1) [22 ng/ml (normal range 93-567 ng/ml)] levels. Insulin tolerance test -as a GH stimulation test- revealed a low GH [2.1 µg/L (normal>5 µg/L) level. A slightly increased serum prolactin level [3.8 ng/mL (normal range 4.8-23.3 ng/mL)] was also noted. Thyroid function tests, luteinizing hormone (LH), follicle-stimulating hormone (FSH), cortisol and electrolytes were all within normal ranges. Brain magnetic resonance imaging (MRI) revealed marked anterior pituitary hypoplasia. Pituitary stalk duplication and the downward extension of the third ventricle into the pituitary fossa via the split in the hypothalamus were also evident in

the sagittal T1-weighted magnetic resonance image (Figure 1). Coronal T1- and T2-weighted magnetic resonance images nicely delineated the duplicated pituitary stalks and the split in the hypothalamus (Figure 2). No additional brain malformation was detected on MRI. Computed tomography (CT) of the head and cervical vertebrae revealed no cranio-cervical bony pathology.

Based on the clinical, laboratory and imaging findings, the patient was diagnosed with GH deficiency as the result of anterior pituitary hypoplasia and duplicated pituitary stalk. A GH replacement therapy was started and close monitoring of the endocrine functions was planned. Informed consent for publication was obtained from the patient.



Figure 1. A 12-year-old boy who presented with the complaint of short stature. Sagittal T1-weighted magnetic resonance image shows marked anterior pituitary hypoplasia. The duplicated pituitary stalks (red arrows) and the downward extension of the third ventricle into the pituitary fossa via the split in the hypothalamus (green arrow) are also evident.

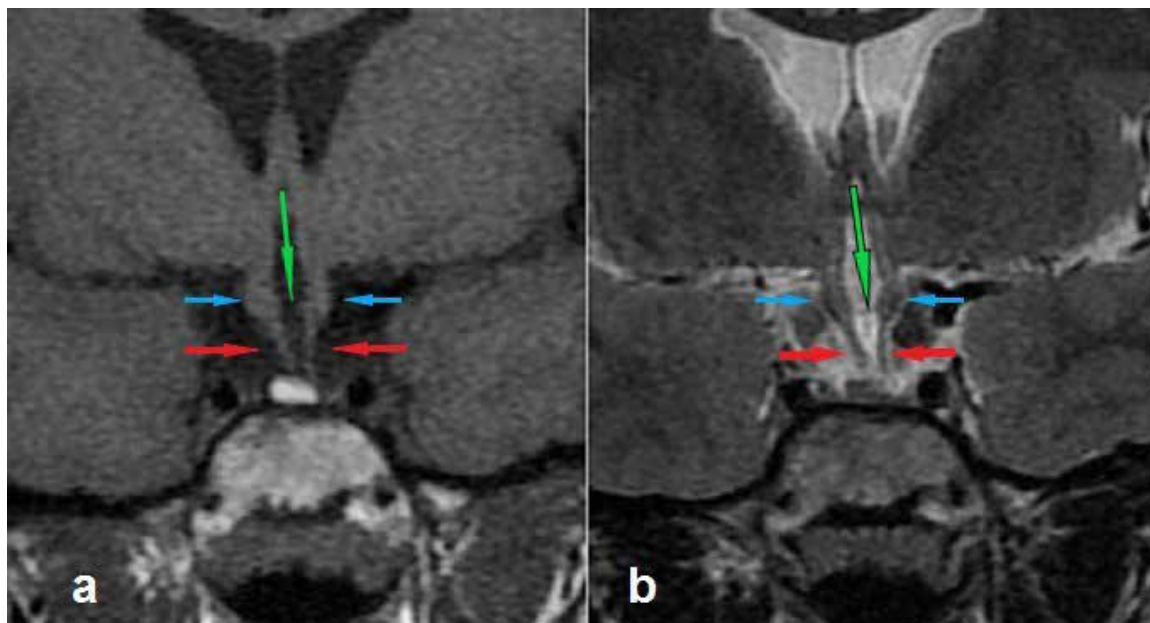


Figure 2. A 12-year-old boy who presented with the complaint of short stature. (a) Coronal T1-weighted magnetic resonance image shows duplicated pituitary stalks (red arrows), a split in the hypothalamus (blue arrows) and the passage in the hypothalamus where the third ventricle extends into the pituitary fossa (green arrow). (b) Coronal T2-weighted magnetic resonance image shows duplicated pituitary stalks (red arrows), a split in the hypothalamus (blue arrows) and the passage in the hypothalamus where the third ventricle extends into the pituitary fossa (green arrow).

Discussion

Pituitary duplication is a rare anomaly with about 40 cases reported to date [2]. And the duplication of the pituitary stalk with a single gland is an exceptionally rare manifestation with only a few case reports in the current literature [4-6]. The duplication of the pituitary gland and stalk seems to be two forms of the same disorder which are in continuation with each other and differ in terms of the embryonal stage at which the neural development interrupted. Although many factors such as prenatal exposure to teratogens, the inclusion of a dermoid in the midline, or various genetic disorders have been accused of giving rise to this rare embryonal disorder, the exact cause has not yet been clarified [1-6]. Genetic causes of pituitary developmental anomalies, including pituitary duplication, have extensively been studied over the last few years [6-8]. Recently, ROBO1 novel heterozygous variants have been reported in patients with pituitary stalk interruption syndrome [8]. Further, Scala et al. reported a case of familial ROBO1 deletion associated with duplication of the pituitary stalk, anterior pituitary hypoplasia, and ectopic posterior pituitary [7]. The pituitary imaging findings and endocrine laboratory results of the patient reported by Scala et al. were very similar to those of the patient we currently present. However, in the case reported by Scala et al., both the patient and the patient's father showed facial dysmorphic features unlike in our case. Unfortunately, because the patient's family did not give consent, genetic counseling could not be performed in our case. However, as recommended by Scala et al., investigation of ROBO1 haploinsufficiency in patients with congenital pituitary anomalies would add much to the existing information on the etiological mechanism of the pituitary duplication.

Although incidentally discovered cases of pituitary duplication have been reported [3], patients with this disorder usually present with endocrine disturbance(s) regarding the adenohipophyseal axis and/or with accompanying pathologies. According to the previous reports, median cleft face syndrome, skull base or cervical bony clefts or duplications, agenesis or hypoplasia of corpus callosum, sellar duplication, widened optic chiasm, tuberomammillary or pituitary mass, moya moya disease and morning glory disc anomaly can accompany pituitary duplication [1-7]. Cross-sectional imaging modalities play a key role in the comprehensive evaluation of these patients. Craniofacial and cervical bony disorders can be demonstrated in detail, using multiplanar CT [3]. And MRI is the method of choice for the assessment of the brain, orbita and vascular pathologies [9]. Using conventional MRI sequences, we clearly demonstrated the hypoplastic adenohipophysis and double pituitary stalks. Brain MRI and craniocervical CT studies revealed no additional pathologies in our patient. To our knowledge, this is the first case report of pituitary stalk duplication without a duplicated gland or associated findings.

The management of the patients with pituitary duplication varies according to the clinical presentation, hormone profile and the presence or absence of the additional pathologies. Rapid hormone replacement therapy and close monitoring of endocrine functions are critical in patients presenting in childhood with hormone deficiencies and developmental delay [7]. In addition, detailed neurological, ophthalmologic, and neuroradiological evaluation and follow-up are necessary to avoid missing a concomitant pathology in these patients [1].

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and

its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

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Conflict of interest

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