Coexistence of pituitary macroadenoma and Mayer-Rokitansky-Küstner-Hauser syndrome

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ABSTRACT

The Mayer-Rokitansky-Küstner-Hauser (MRKH) syndrome is characterized by congenital aplasia of the uterus and upper part (2/3) of the vagina in women showing normal development of secondary sexual characteristics with a normal 46, XX karyotype. We report a case of a 30-year-old woman who presented with complaints of headache. Pituitary macroadenoma and MRKH syndrome were diagnosed. To the best of our knowledge, pituitary macroadenoma has not been reported in association with MRKH syndrome. However, no genetic links between MRKH syndrome and pituitary macroadenoma have been observed. Thus, the association may be incidental rather than causal.

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1. Introduction

Mayer-Rokitansky-Küstner-Hauser (MRKH) syndrome is characterized by congenital aplasia of the uterus and upper part (2/3) of the vagina in women showing normal development of secondary sexual characteristics with a normal 46, XX karyotype. The incidence of this anomaly is estimated to range from 1:4000 to 1:5000. Pituitary adenomas, clinical as well as subclinical, are common. However, they are rarely accompanied by congenital anomalies. Here, we report the first case of pituitary macroadenoma associated with MRKH syndrome.

2. Case report

In July 2007, a 30-year-old woman presented with a one-month history of headache. She had not attained menarche. She had no family history of congenital disorders or exposure to teratogenic agents during gestation. External examination revealed completed puberty with normal female sexual characteristics (Tanner stage 5, pubic hair and breast development) and normal external genitalia. Neurological examination revealed bitemporal hemianopsia. The basal prolactin level was 693.1 ng/mL. Besides prolactin, all other endocrinological findings were within the normal range. Radiological evaluation revealed no skeletal deformity. The patient had a normal 46, XX karyotype. Cranial magnetic resonance imaging (MRI) revealed a homogeneously enhancing, large, lobulated intrasellar mass in the intra- and suprasellar region, measuring 43 mm in craniocaudal diameter. The mass extended into the 3rd ventricle, interpeduncular fossa, and left cavernous sinus (Fig. 1). Transrectal ultrasonography (US) revealed a 2.5 cm small hypoplastic uterus and a 6 cm bilobulated cystic mass. According to the US findings, MRKH was suspected and a pelvic MRI examination was performed to confirm the diagnosis. MRI showed a small hypoplastic uterus and a thin, hyperintense, lined endometrium (Fig. 2). Bilateral ovaries were observed. A 6 cm bilobulated cystic mass was noticed, and it was suspected to be hydrosalpinx. These findings confirmed MRKH syndrome. No other anomalies, frequently reported in MRKH patients, were observed.

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in our patient. We recommended a transsphenoidal adenectomy (TSA) for the pituitary tumor, but the patient refused the operation and was administered bromocriptine mesylate instead. After 1 month of medication, the prolactin level returned to normal, and a cranial MRI after 6 months of medication revealed that the size of the pituitary tumor had decreased by approximately 40%. In February 2008, the patient was admitted to an emergency room with severe headache and diagnosed with pituitary apoplexy. TSA was then immediately conducted. The histological diagnosis was a pituitary adenoma. The patient’s condition has improved, except for the bitemporal hemianopia.

3. Discussion

There appear to be two subtypes of MRKH: the typical (also called type I or isolated) and the atypical (type II). The occurrence of type II is more frequent. Isolated utero-vaginal aplasia is referred to as Rokitansky sequence or type I MRKH syndrome. Incomplete aplasia along with or without other associated malformations, is generally referred to as type II MRKH syndrome. Other associated malformations include renal (unilateral agenesis, ectopia of kidneys, or horseshoe kidney) and skeletal (in particular vertebral) malformations (Klippel-Feil anomaly; fused vertebrae, mainly cervical; scoliosis), hearing defects, and, more rarely, cardiac and digital anomalies (syractyly, polydactyly). Cases of polycystic ovaries and ovarian tumors have been described in women with normal 46, XX karyotypes. Our patient was diagnosed with type II MRKH syndrome although there were no other associated malformations. Previously the syndrome had been considered as a sporadic anomaly, but an increasing number of familial cases now support the hypothesis of a genetic cause. In familial cases, the syndrome appears to be transmitted as an autosomal dominant trait with incomplete penetrance and variable expressivity. This suggests the involvement of either mutations in a major developmental gene or a limited chromosomal imbalance. However, the etiology of MRKH syndrome still remains unclear. Treatment of vaginal aplasia, which involves construction of a neovagina, can be provided to allow sexual intercourse. As psychological distress is very important in young women with MRKH, it is essential for the patients and their families to attend counseling before and throughout treatment.

To the best of our knowledge, pituitary macroadenoma has not been reported in association with MRKH syndrome. However no genetic links between MRKH syndrome and pituitary macroadenoma have been observed. Thus, the association may be incidental rather than causal.

References