Case Report

Mayer-Rokitansky-Kuster-Hauser syndrome and ovarian benign teratoma: a case report

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ABSTRACT

Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH) is an unknown congenital etiology disorder characterized by agenesis or hypoplasia of the Müller ductal system, including the upper vagina, uterus and fallopian tubes. The occurrence of an associated ovarian tumor is rare, with fewer than 20 cases reported to date according to the literature. We report the case of a 14-year-old girl, virgin, who had not yet seen her menarche, complaining of an abdominopelvic mass associated with pain. The ultrasound performed revealed a large left ovarian tumor and an absence of uterus. The indication of a laparotomy confirmed the ovarian mass and a complete absence of uterus associated with vaginal hypoplasia. The contralateral ovary was present, and of normal appearance. The pathological examination was in favor of a mature benign multissular teratoma. This is the first case described in our service. The mode of transmission of this entity appears to be autosomal dominant with low penetrance and variable expressivity, suggesting that the incidence of this syndrome is likely underestimated. With the development of techniques of medical assistance to procreation, maternity remains possible, particularly through gestational surrogacy.

Keywords: Agenesia, Mayer-Rokitansky-Kuster-Hauser, Ovarian tumor, Uterine hypoplasia

INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH) is a rare abnormality characterized by congenital aplasia of the uterus and vagina in women with normal development of secondary sex characteristics and a karyotype 44 XX. It is a rare disease, which is a common cause of primary amenorrhoea. The etiological basis of the syndrome has not yet been defined. Most cases are sporadic, although family groupings were reported, suggesting an alleged genetic origin of the syndrome. However, data on genetic and genomic alterations identified in patients with MRKHS, as well as the hereditary patterns observed, are consistent with a polygenic disorder. In addition, association with other malformations involving the kidneys, skeleton and ears is common. The occurrence of a gynecological tumor was reported in only a few cases of MRKHS syndromes. However, data on the association of a benign ovarian tumor are still limited. Authors report here a case of MRKH syndrome discovered during a laparotomy for a large ovarian tumor.

CASE REPORT

This is DH, a 14-year-old patient, who consulted at Angré teaching hospital in Abidjan for a large abdominopelvic mass associated with pelvic pain. The evolution was progressive over a period of about 6 months. She had no other associated sign apart from the effects related to compression. The interrogation found an absence of menarche. She never had sexual intercourse. The physical
examination performed in the patient noted a good general condition, a normal morphotype, the hemodynamic constants were normal. The examination of the abdomen revealed a large, firm, regular, painless mass, ranging from the pelvic region to the xiphoid appendage (Figure 1). Examination of the vulva did not objectify any abnormality; the hymen was intact. In addition, the pleuro-pulmonary and cardiovascular examinations were without particularity.

Figure 1: Abdominal-pelvic mass.

Figure 2: Pre-operative left ovarian tumor.

Figure 3: Absence of uterus.

Figure 4: Healthy contralateral appendage.

Figure 5: Adnexectomy.

Figure 6: Mixed ovarian tumor.
In front of this voluminous abdomino-pelvic mass, an ultrasound was made, and concluded to a voluminous left ovarian tumor (210 mm) of long axis. The requested tumor markers (ACE, CA125, CA19.9, alpha-fetoprotein) were normal, as was the conventional preoperative assessment. The intervention was planned, and intraoperatively, it was a large mixed tumor (fluid and solid) about 25 cm in diameter, developed at the expense of the left ovary (Figure 2). There was a complete absence of uterus (Figure 3). The ovary and contralateral tube (right appendage), were macroscopically healthy (Figure 4). Aspiration of the contents of the tumor allowed to collect 2 liters of citrine liquid. An ultrasound was made, and concluded to a voluminous left ovarian tumor (210 mm) of long axis. The requested tumor markers (ACE, CA125, CA19.9, alpha-fetoprotein) were normal, as was the conventional preoperative assessment. The intervention was planned, and intraoperatively, it was a large mixed tumor (fluid and solid) about 25 cm in diameter, developed at the expense of the left ovary (Figure 2). There was a complete absence of uterus (Figure 3). The ovary and contralateral tube (right appendage), were macroscopically healthy (Figure 4). Aspiration of the contents of the tumor allowed to collect 2 liters of citrine liquid. A left total adnexectomy was performed (Figure 5). The tumor after opening, was a mixed ovarian tumor (Figure 6). The operative follow-up was simple, the exit was decided at D4 postoperative with a post-operative appointment at D15. The histology performed revealed a benign pluritissular mature teratoma. A complementary radiological study of the urinary shaft did not reveal any associated malformations.

**DISCUSSION**

**Epidemiology**

The prevalence of MRKH syndrome is estimated at 1 per 4,000 female live births in the Caucasians. In addition, the etiology of MRKH syndrome remains unknown. The majority of cases appear to be sporadic, however, reports of familial cases of MRKH syndrome suggest a genetic component. In our patient, we found no family history of this pathology whose prevalence is unknown, and which remains a mystery in our country.

**Diagnosis**

The MRKH syndrome is a devastating diagnosis for a young woman to receive, which has considerable medical, psychological, social and reproductive implications. The diagnosis is often made in adolescence in the context of primary amenorrhoea with normal puberty. In our observation, the diagnosis was fortuitous following a laparotomy for a large ovarian tumor in a 14-year-old girl with no particular history. This condition, often suspected in the face of primary amenorrhoea, is usually confirmed radiologically (ultrasound or MRI) or by laparoscopy in patients for whom hormonal examinations and karyotype are normal. The MRKH syndrome was subdivided into 2 types: type I, with malformations of the Müller's canal which are in the form of a superficial vaginal dimple with uterine cervix, uterus and upper vagina absent and is not associated with other abnormalities. Type II with Müller’s canal agenesis similar to type I and various degrees of associated congenital renal malformations (renal agenesis and horseshoe kidney), skeletal abnormalities (scoliosis, spina bifida) and in rare cases of unilateral hearing defects, cardiac and extremities abnormalities. In our case, the patient was only 14 years old, and had not yet seen her menarche, which was not a concern for parents. MRKH syndrome was discovered during a laparotomy for ovarian tumor. The clinical and paraclinical investigations carried out did not reveal any other malformations. It was a type I. In the most recent publications on MRKH syndrome, we could find cases reported on leiomyoma, adenomyosis, but ovarian tumor is rare in MRKH syndrome and difficult to diagnose if it is not bulky. The incidence of the combination of

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**Table 1: Observations of MRKH associated with ovarian tumors.**

| Articles            | Country | MRKHS type | Ovarian tumor age at onset | Ovarian tumor histology | Ovarian tumor type | Ovarian tumor grade | Ovarian tumor stage (FIGO) |
|---------------------|---------|------------|---------------------------|-------------------------|-------------------|---------------------|-----------------------------|
| Takeuchi et al      | Japan   | NA         | 8                         | Germ cell               | Yolk sac tumor    | Na                  | I                           |
| Mishina et al       | Moldova | NA         | 35                        | Germ cell               | Dysgerminoma      | Na                  | I                           |
| Kavallaris et al    | Germany | II         | 48                        | Epithelial             | Mixed epithelial carcinoma | III                | III                         |
| Bae et al           | Korea   | II         | 31                        | Epithelial             | Serous papillary  | Na                  | III                         |
| Ko et al            | Korea   | II         | 37                        | Epithelial             | Serous carcinoma  | III                | III                         |
| Nusrath et al       | India   | II         | 65                        | Epithelial             | Endometrioid carcinoma | Na              | I                           |
| Juusela et al       | United States | I   | 72                        | Sex-cord stromal       | Bilat. Sex-cord stromal tumor | Na | I                           |
| Huepenbecker et al  | United States | NA | 64                        | Epithelial             | Serous carcinoma  | III                | III                         |
| Villa R et al       | Italy   | I          | 33                        | Epithelial             | Bilat. Serous carcinoma | I                 | II                         |
MRKH with ovarian tumors was not reported. However, a review of the literature demonstrated nearly a dozen reported cases of MRKH syndrome associated with ovarian tumors (Table 1). Benign tumors were predominant in most studies. This is the case in our observation, or it was a left ovarian benign multi-tissular teratoma which is a benign tumor often called ovarian dermoid cyst.

Treatment

Laparoscopy is the gold standard for the diagnosis and management of benign ovarian tumors. In the literature, in five cases described, associating an ovarian tumor with MRKH syndrome, management was performed under laparoscopy, which confirms the interest of this approach. In addition, conventional surgery by laparotomy is possible, especially in case of large solid or mixed tumor as was the case in our observation. In addition, the patient’s care remains holistic, taking into account the psychosocial aspects, vaginoplasty to improve sexual practice and the possibility of medical assistance, the cost of which is prohibitive in our country.

CONCLUSION

MRKH syndrome is a rare condition. The association with a voluminous ovarian tumor is even rarer, if not exceptional. The available data and knowledge on the etiopathogenic factors being still limited, it is impossible to define a supposed correlation between these two affections. The collection of new data and the publication of possible new cases could lead to a better knowledge of this association. With the rise of assisted reproduction techniques (ART), maternity remains possible.

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