Mayer-Rokitansky-Küster-Hauser Syndrome: Distinction Between Two Forms Based on Excretory Urographic, Sonographic, and Laparoscopic Findings

OBJECTIVE. The purpose of this study was to discriminate typical (type A) from atypical (type B) Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome (congenital absence of vagina and uterus) and determine their association with renal anomalies and ovarian disease.

MATERIALS AND METHODS. The excretory urographic, sonographic, and laparoscopic findings in 91 patients with MRKH syndrome were compared retrospectively. Symmetric muscular buds and fallopian tubes were diagnostic of type A, and asymmetric muscular buds or abnormally developed fallopian tubes were diagnostic of type B.

RESULTS. On the basis of laparoscopic findings, type A was diagnosed in 40 patients (44%) and type B was diagnosed in 51 patients (56%). Renal anomalies were found in 34 (37%) of the 91 patients, all of whom had type B syndrome. Renal agenesis and a pelvic kidney were the most common findings in the upper part of the urinary tract. Ovarian abnormalities were seen in 14 patients (15%), all of whom had type B syndrome. Sonography did not allow discrimination between types A and B in patients with normal kidneys (17/51 = 33%), but it provided important information in patients with associated cyclic abdominal pain, in cases of diagnostic dilemma, and in patients with associated renal anomalies.

CONCLUSION. Discrimination between type A and type B of MRKH syndrome is important because associated renal and ovarian abnormalities occur only in type B. Laparoscopy is still needed to discriminate between these two forms. Sonography is useful for diagnosing cyclic abdominal pain and associated renal anomalies.

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Congenital absence of the uterus and vagina, Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, is a rare disorder. The prevalence has been reported as one in 4000–5000 female births [1–5]. Patients with MRKH syndrome have a 46,XX karyotype and normal secondary sex characteristics. The external genitalia appear normal, but only a shallow vaginal pouch is present. Ovarian function is normal [4–6]. The typical form of the syndrome is characterized by the absence of both the vagina and the uterus. Only symmetric uterine remnants (the muscular buds), normal fallopian tubes, and normal ovaries are present [1, 4, 5, 7–10] (Fig. 1).

In 1977, on the basis of laparoscopic findings in 10 patients, Schmid-Tannwald and Hauser [8] described abnormalities of the internal genitalia that differed from the typical form of MRKH syndrome. These differences included asymmetric uterine remnants (aplasia of one or both muscular buds, and when both muscular buds were found, one muscular bud was larger than the contralateral one) and abnormalities of the fallopian tubes (hypoplasia or aplasia of one or both tubes). In seven of these 10 patients, congenital renal abnormalities were seen. They also noticed ovarian disease in 10 patients: cystic ovaries, or anomalies in form or position of the ovaries. They called their findings the atypical form of MRKH syndrome.

We retrospectively reviewed the sonographic, excretory urographic, and laparoscopic findings in 91 patients with MRKH syndrome to determine imaging features.
that can be used to distinguish typical from atypical forms and to correlate the association of congenital renal anomalies with these two entities.

Materials and Methods

From 1982 to 1990 at the University Hospital Nijmegen in the Netherlands, MRKH syndrome was diagnosed in 91 patients between 15 and 45 years old (mean age, 25 years). The diagnosis was based on findings at physical examination and laparoscopy in all patients. Signs and symptoms included primary amenorrhea (78%), sterility (9%), and dyspareunia (13%). None of the patients had a medical history that suggested urologic disease (pain, hypertension, pyelonephritis, urinary abnormalities) or testicular feminization. In retrospect, no indication of MRKH syndrome could be found in the relatives of these women. Nine patients (10%) also complained of concomitant cyclic abdominal pain. On the basis of laparoscopic findings, a distinction was made between typical (type A) and atypical (type B) forms; patients with type A had symmetric muscular buds and normal fallopian tubes (Fig. 1); patients with type B had asymmetric muscular buds (aplasia of one or both buds or, when both buds were found, one bud smaller than the other one), or abnormally developed fallopian tubes (hypoplasia or aplasia of one or both tubes) (Fig. 2).

In all patients, excretory urograms were obtained in order to exclude congenital urinary tract abnormalities. Sonography was performed in 25 patients because of cyclic abdominal pain (nine) or because of difficulty in diagnosis. All abdominal and pelvic sonograms were obtained with standard real-time equipment (3.5-MHz sector scanner) and analyzed for the presence of uterus and renal abnormalities.

We retrospectively reviewed the excretory urograms, sonograms, and laparoscopic findings. Renal agenesis was diagnosed on the basis of findings on excretory urograms, that is, no opacification of the urinary tract on one side, a normal or enlarged kidney on the other side, and no other abnormalities. Renal hypoplasia was diagnosed if the kidney was very small but otherwise similar to a normal organ [11].

Results

On the basis of laparoscopic findings, type A MRKH syndrome was diagnosed in 40 patients (44%) and type B was diagnosed in 51 patients (56%). The abnormal laparoscopic findings included aplasia of one muscular bud (11 cases), aplasia of both muscular buds (four cases), one muscular bud smaller than the other (seven cases), aplasia or hypoplasia of one fallopian tube (eight cases), aplasia or hypoplasia of both fallopian tubes (seven cases), fallopian tube aplasia or hypoplasia and muscular bud aplasia (10 cases), and unicorne uterus with hematometra (four cases).

Laparoscopy showed additional abnormalities of the ovaries in 14 patients (15%), all of whom had type B syndrome. These findings included inguinal hernia containing an ovary (six cases), no descent of ovary (five cases), agenesis of one ovary (one case), and streak ovaries (two cases).

Anomalies of the upper urinary tract were seen on excretory urograms in 34 patients (37%), all of whom had type B MRKH syndrome (34/51, 67%). The renal abnormalities included renal agenesis (17 cases), pelvic kidney (eight cases), renal agenesis and contralateral pelvic kidney (five cases), renal hypoplasia (three cases), and horseshoe kidney (one case).

In 21 of 25 patients (five type A; 20 type B) examined with sonography, agenesis of the uterus was confirmed. In four patients with cyclic abdominal pain, a unicornuate uterus with hematometra was found. In five other patients with cyclic abdominal pain and type B syndrome, symptoms were attributed to the laparoscopic finding of a small amount of endometrial tissue inside the bud(s); the pain resolved after the tissue was surgically removed. This abnormality was not detected on sonograms in any case. In all 25 patients, sonography confirmed the results of excretory urography. Eight of these patients had renal agenesis, a pelvic kidney, or both.

We compared congenital anomalies of the urinary tract with laparoscopic findings of the internal genitalia, with respect to the side on which they occurred. Unilateral anomalies of the urinary tract were found to be associated with ipsilateral uterine bud agenesis or fallopian tube dysplasia (16 cases), contralateral uterine bud agenesis or fallopian tube dysplasia (three cases), or bilateral uterine bud agenesis or fallopian tube dysplasia (nine cases). Bilateral anomalies of the urinary tract were found to be associated with unilateral uterine bud agenesis or fallopian tube dysplasia (two cases) and bilateral uterine bud agenesis or fallopian tube dysplasia (four cases).
These results show that three of the 19 patients with unilateral urinary tract anomalies had uterine bud agenesis or fallopian tube dysplasia on the other side.

Discussion

MRKH syndrome is defined by the congenital absence of the vagina and the uterus. Instead of a normal uterus, patients with MRKH syndrome have bilateral nonfunctioning rudimentary uterine anlagen in the form of small, noncanonicalized, muscular (myometrial) buds (müllerian duct remnants) [1–6]. In 6–10% of cases, however, endometrial tissue or even variable development of the uterus with hematometra may be present, resulting in cyclic abdominal pain [4, 7, 12–14]. Associated congenital anomalies of the upper urinary tract are reported to occur in 30–40% of all cases, and the most common are renal agenesis and pelvic kidney [2–7, 15].

Both typical and atypical forms of this disease have been described. In 1982, this distinction was noted by Ghirardini and Segre [9] in six patients. In 1988, Heidenreich [10] found a combination of congenital renal anomalies next to asymmetric buds or abnormally developed fallopian tubes in 15 of 51 patients with MRKH syndrome. However, he did not define his findings in a typical or atypical form. In the present series of 91 patients, more than half (51) of the patients with the clinical MRKH syndrome had the atypical form. We therefore suggest that the typical and atypical forms of the disease be designated “type A” and “type B,” respectively. Various frequencies have been reported for anomalies of the urinary tract, especially agenesis or ectopia of kidneys, in the general population. Fore et al. [16], using autopsy reports, reported a frequency of one in 920 to one in 1850 (0.1–0.5%), which is in agreement with other reports summarized by Felding [17].

In our group of 91 patients with MRKH syndrome, 34 (37%) had associated congenital anomalies of the urinary tract, and all had type B syndrome (34/51, 67%), which confirms the results of Schmid-Tannwald and Hauser [8]. No specific data are known on what percentage of women with an absent kidney might have MRKH syndrome. In 40–50% of patients with renal agenesis, an associated genital anomaly has also been found [17, 18].

Schmid-Tannwald and Hauser [8] proposed a hypothesis to explain the association between genital and renal anomalies in MRKH syndrome. Faulty gonadal differentiation can occur, with consequent production of müllerian inhibiting factor, which induces regression of the müllerian ducts, as normally seen in males. Depending on the onset of production of müllerian inhibiting factor, and the asymmetry in production by the two gonads, the development of the müllerian ducts would stop at various stages. This theory could explain the uterine asymmetry. In 16 of the 19 patients with unilateral renal anomalies, the uterine bud agenesis or fallopian tube dysplasia was ipsilateral; in the other three patients with unilateral uterine tract anomalies, the uterine bud agenesis or fallopian tube dysplasia was contralateral. This is variably reported by other authors [4, 7, 8, 10, 19].

All patients with associated ovarian abnormalities had type B MRKH syndrome. We agree with Ghirardini and Segre [9] that the hypothesis of Schmid-Tannwald and Hauser [8] could explain this association. This theory could also explain the inguinal hernia containing an ovary as was seen in six patients, because of the production of a müllerian inhibiting factor [8].

The theory of Schmid-Tannwald and Hauser is difficult to prove because biopsies of the ovaries were not performed in our patients and our study did not focus on detecting and isolating the müllerian inhibiting factor. More studies need to be done, especially with respect to the ovaries, to confirm the suggestion of Schmid-Tannwald and Hauser that patients with type B MRKH syndrome may have a very slight form of female pseudohermaphroditism [8–10].

Review of the patient's medical history and a simple gynecologic examination usually are sufficient to diagnose MRKH [1, 4, 7]. Laparoscopy was performed in our study to differentiate between type A and type B of the disease. Sonography did not show differences between these two types when no urinary abnormality was shown. In those cases in which a diagnostic dilemma exists, sonography may be useful to confirm uterine agenesis [4, 20]. In 6–10% of cases, however, endometrial tissue in the muscular buds or even a unicorne uterus with hematometra may cause associated cyclic abdominal pain. Sonography has been recommended for diagnosis of these abnormalities [20, 21], which were seen in nine patients (10%) in our study. Four of these nine patients had a unicorne uterus with a hematometra, which was correctly diagnosed on the basis of sonographic findings. In five other patients, sonograms did not show the small amount of endometrial tissue in the muscular buds, which was later found at laparotomy. The difficulties in detecting small remnants of endometrial tissue on sonograms has been reported [20].

In our study, all anomalies of the upper urinary tract were seen exclusively in patients with type B MRKH syndrome. Sonography provided the same information as excretory urography and provided additional information in cases of hematometra. We did not consider the role of MR imaging in discriminating between the two forms of MRKH syndrome in this study. It is not certain whether these two forms can be differentiated as accurately on MR images as at laparoscopy, but the recent study of Fidele et al. [22] might set a starting point. In small series of patients, MR imaging was useful for showing small amounts of endometrial tissue in patients who have cyclic abdominal pain [12, 22, 23]. More prospective studies in representative groups of patients are needed to establish the definite role of MR imaging in patients with MRKH syndrome.

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REFERENCES