

Mayer-Rokitansky-Kuster-Hauser Syndrome: US Aid to Diagnosis¹

The Mayer-Rokitansky-Kuster-Hauser syndrome is composed of vaginal atresia with other variable Müllerian duct abnormalities such as bicornuate or septated uterus. The fallopian tubes, ovaries, and broad and round ligaments are normal. Unilateral renal and skeletal anomalies are associated in 50% and 12% of cases, respectively. Patients have a normal female karyotype and normal secondary sexual development. Previously, one had to rely on radiographic contrast studies and surgical exploration for accurate definition of the reproductive tract anatomy. The authors performed ultrasound (US) examinations on 12 patients, aged 5 days to 18 years, with the Mayer-Rokitansky syndrome. US allowed correct identification of the genitourinary anomalies found in these girls, including eight cases of unilateral renal agenesis; one absent, one rudimentary, and ten duplicated or obstructed uteri; eight duplicated or obstructed vaginas; and associated complications such as endometriosis. Eight of the patients had lower abdominal pain, often cyclical in nature. The findings demonstrate that high-resolution, real-time US in conjunction with water vaginography permits the anatomy of these complex anomalies to be defined.

Index terms: Genitourinary system, abnormalities • Genitourinary system, US studies, 8.1298 • Uterus, abnormalities • Vagina, abnormalities

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A SYNDROME involving absence of the vagina and other anomalies was first described by Mayer in still-born female infants in 1829 (1). Subsequently, Rokitansky in 1838 (2) and Kuster in 1910 (3) described a syndrome composed of vaginal atresia, rudimentary bicornuate uterus, and normal fallopian tubes, ovaries, and broad and round ligaments. There is a spectrum of uterine anomalies, ranging from a partial lumen to septate or bicornuate uterus with unilateral or bilateral obstruction. Unilateral renal (50%) and skeletal (12%) anomalies are associated. The patients have a normal female karyotype and secondary sexual development, with normal circulating levels of human chorionic gonadotropin (hCG), luteinizing hormone (LH), and follicle-stimulating hormone (FSH). The external genitalia are those of a normal female, although the introitus may end in a shallow, blind pouch. In patients with a patent, functional uterus, fertility might be expected to be normal.

Hauser and Schreiner emphasized the frequency of the disorder and the spectrum of associated abnormalities (4). Those females with imperforate anus or cloacal exstrophy may have vaginal stenosis, atresia, or a urogenital sinus. There may be septation of the uterus with or without atresia or obstruction, as well as associated renal anomalies in up to 50% of patients. Agnesis of one kidney or ectopia of one or both kidneys are most common. Less often, one may find a horseshoe-type kidney, abnormalities of the collecting systems, malfunctioning kidneys of unclear cause, or renal malrotation (5).

The cause of the syndrome is unknown. However, it is hypothesized that either an unknown toxic substance or a genetically induced event interferes with the developing Müllerian (paramesonephric) duct, the Wolffian (mesonephric) duct with its

ureteral bud, the metanephros, and the mesoderm that gives rise to the dorsal spine (5).

Before the advent of high-resolution, real-time ultrasound (US), one had to rely on excretory urography, cystography, voiding cystourethrography, vaginography (6), hysterosalpingography (7), and pneumopyelography (8) to help sort out the anatomy before surgery. It was often impossible to assess the components of an obstructed genitourinary system without laparoscopy (9) and/or exploratory surgery. Sonography (10-16) enables one to demonstrate anatomic detail, define the reproductive anomalies, evaluate renal and bladder abnormalities, and elucidate suspected spinal abnormalities (17). US, particularly when combined with water vaginography, may obviate the need for radiographic contrast studies or limit the number of studies done. More definitive therapeutic procedures may be planned in most cases without the use of invasive techniques. The objective of this paper is to illustrate the value of US in defining the wide spectrum of anomalies in the Mayer-Rokitansky syndrome.

PATIENTS AND METHODS

At the Children's Hospital of Philadelphia, we have seen 12 patients, aged 5 days to 18 years, with the Mayer-Rokitansky syndrome. Abdominal pain (cyclical or intermittent) was the initial symptom in eight of the 12. Two girls were being evaluated for urinary tract infection. The newborn infant had an imperforate anus and a ring 18 chromosome and was being evaluated for associated genitourinary anomalies. Five of the girls had a history of imperforate anus. All patients were examined with an Advanced Technology Laboratories (Bothell, Wash.) mechanical sector scanner equipped with a multifrequency transducer scan head (3.0, 5.0, and 7.5 MHz). A Siemens (Iselin, N.J.) articulated-arm gray-scale static B scanner was used in some cases. Routine renal and

pelvic US was performed. In four cases, water vaginography was performed to help elucidate the findings. In an additional patient, water vaginography was attempted but not accomplished because the vaginal introitus was covered by a flap of tissue.

RESULTS

The US and surgical findings are summarized in Table 1. US enabled the genitourinary anomalies to be correctly identified in all cases, except in patient 9. In this 14-year-old girl, there was a narrow right-sided cystic structure, the cause of which could not be determined from the sonogram. The patient had undergone multiple operative procedures before US examination, and it was difficult to correlate the sonographic and surgical findings. Eight of the patients underwent excretory urography, five underwent voiding cystourethrography, two underwent radiographic contrast vaginography, and one underwent computed tomography (CT). Patients evaluated during the past 2 years have undergone fewer radiographic contrast studies because of the increased confidence of the surgeons in the US findings.

In one 18-year-old patient (patient

1, Fig. 1), a 9.0 × 5.0-cm septated cystic mass was found in the right adnexal region in addition to the anomalies. This mass was proved at surgery to be a serous cystadenoma. US was especially valuable in those girls who had pain caused by obstruction of the uterus and/or vagina (patients 2-9, Figs. 2-5).

Water vaginography aided in perplexing situations. Patient 4 (Fig. 4) had right renal agenesis and duplication of the uterus. An elongated, slightly oval-shaped, mildly echogenic mass was observed on the right side of the pelvis at the level where one ought to visualize the vagina. It was difficult to distinguish whether or not this was a large coalesced mucus collection in a partially obstructed vagina or a vaginal mass. The appearance of the introitus was normal, and water vaginography was performed during real-time US observation. The catheter coursed to the left side of the abnormal vaginal area. Water contrast examination demonstrated a patent left vagina, with occasional wisps of air and water observed entering the right-sided vagina and outlining what appeared to be a collection of debris. We therefore hypothesized that there was a duplication of the vagina with an ob-

structed right vagina and a fenestration in the septum between the two compartments. Our findings were confirmed at surgery; a fenestration was found high in the vaginal septum. In patient 5 (Fig. 5), US revealed (in addition to left renal agenesis and a bicornuate uterus with an obstructed left horn) a large amount of heterogeneously echogenic material in the cul de sac that proved to be old blood and endometriosis surrounding the dilated left tube and ovary. In one infant with imperforate anus (patient 10, Fig. 6), a uterine duplication with fluid-filled, obstructed en-

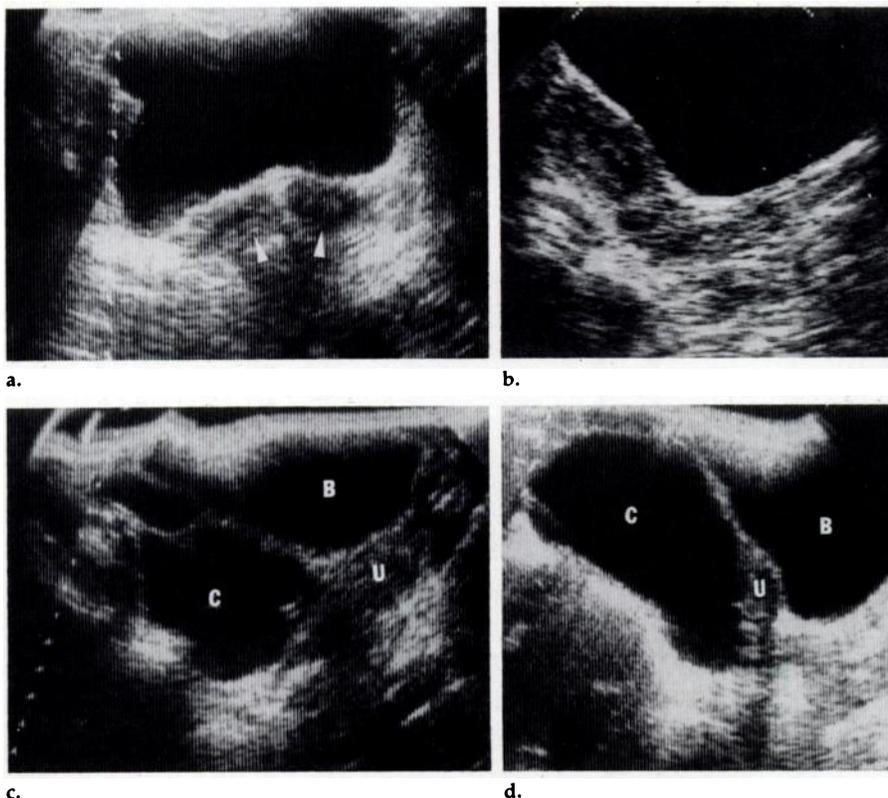


Figure 1. Patient 1. (a) Transverse scan of the pelvis shows two cervixes (arrowheads). (b) Sagittal scan of the left uterus. Transverse (c) and sagittal (d) images show a septated cystic mass (C) in the right adnexa posterior and lateral to the right uterus (U), found at surgery to be a serous cystadenoma. B = bladder.

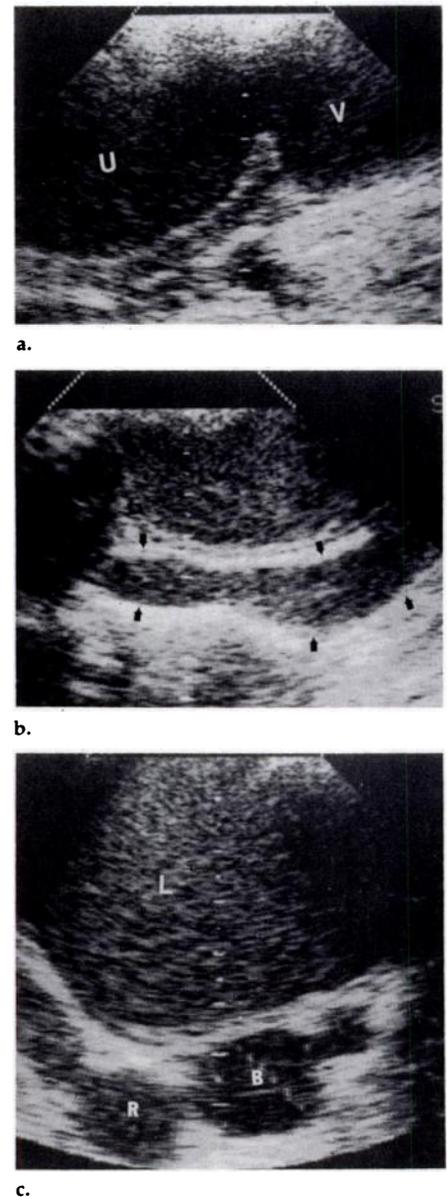


Figure 2. Patient 2. Sagittal scans of the dilated, obstructed left uterus (L) and vagina (V) (a) and the smaller, obstructed, rudimentary right uterus and vagina (arrows) containing echogenic material representing old blood (b). Transverse image (c) shows the blood-filled right (R) and left (L) vaginas, which empty into the bladder plate (B).

ometrial canals was unexpectedly found as we examined the urinary tract for anomalies (crossed, fused renal ectopia). We also found US useful in examining a 6-year-old girl (patient 11, Fig. 7) with a urinary tract infection, difficulty voiding, right renal agenesis, and a rudimentary uterus. A sacral anomaly was noted on the voiding cystourethrogram. Scanning over the sacral defect revealed a widened spinal canal, low-lying spinal cord, and an area of increased echogenicity, which was shown with

magnetic resonance (MR) imaging to represent a low-lying spinal cord. The cord was tethered at L-4.

DISCUSSION

The Mayer-Rokitansky-Kuster-Hauser syndrome is believed to be due to an arrest of development at some time prior to the 5th gestational week. The uterus, cervix, and upper three-fourths of the vagina are formed by the fused caudal ends of the Müllerian (paramesonephric)

ducts. The paired fallopian tubes are formed by the unfused upper ends. Müllerian duct development is dependent on the formation of the Wolffian (mesonephric) duct, which is the anlage of the ureteral bud. An event that interferes with normal development of the Müllerian duct, thus resulting in uterine and vaginal anomalies, may also affect the Wolffian duct and subsequent renal development. This would explain the high association of renal anomalies with the syndrome.

The skeleton, like the mesonephros, is derived from mesoderm. At about the same stage of development when the embryonic defect in the genitourinary tract occurs, the vertebrae develop from adjacent mesodermal cell concentrations and thus may also be damaged (1-5). Two-thirds of the skeletal anomalies involve the spine, including wedge vertebrae, fusions, rudimentary vertebral bodies, and supernumerary vertebrae. Patient 11 (Fig. 7a) had absence of the right and underdevelopment of the left lower sacral segments and coccyx. Other skeletal anomalies may be associated, including syndactyly (18), absence of a digit (19), and hypoplasia of the thenar eminence (20), but these anomalies do not appear to be related embryologically.

The Mayer-Rokitansky syndrome is the second most common cause of primary amenorrhea. Sonography is an excellent imaging modality in these patients. It avoids radiation and can image the entire genitourinary tract noninvasively using only water vaginography if indicated. Although specific radiographic studies would clarify some of the suspected abnormalities, they would all require the use of contrast material and ionizing radiation and cause patient discomfort. US is especially helpful in those patients who have obstructed components of the genital tract (e.g., uterus,

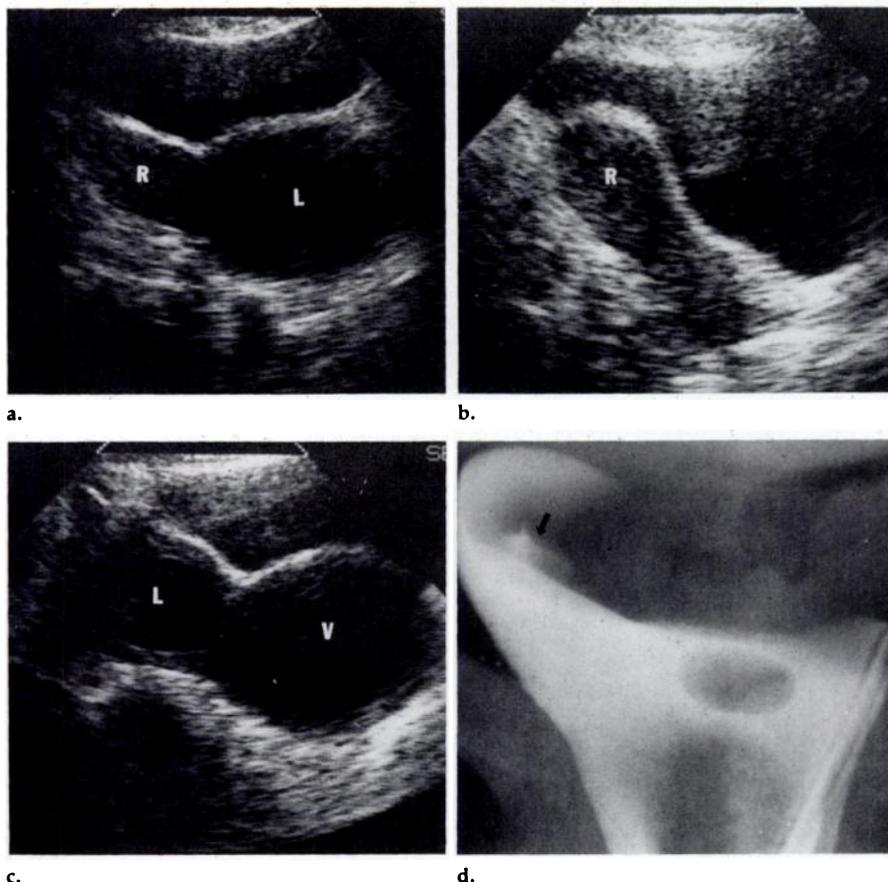


Figure 3. Patient 3. Transverse (a) and sagittal (b, c) scans of the pelvis show a normal right uterus (R in a, b) and a dilated, obstructed left uterus (L in a, c) and vagina (V in c). Preoperative vaginogram (d) shows mass effect on the vagina with displacement of the cervical os (arrow) to the right.



Figure 4. Patient 4. Transverse (a) and sagittal (b) images obtained during water vaginography show the catheter (arrows) in the normal left vagina. Right parasagittal scan (c) of the duplicated right uterus and vagina shows echogenic material within the obstructed vagina. V = debris in right vagina.

Table 1
Findings in 12 Patients with Mayer-Rokitansky-Kuster-Hauser Syndrome

Patient No.	Age	Clinical History	Renal Anomalies	US Findings	Surgical Findings
1	18 yr	Imperforate anus, vaginal stenosis	None	Complete duplicated uterus, 9 × 5-cm septated cystic right adnexal mass (Fig. 1)	Bicornuate uterus, septated vagina, serous cystadenoma
2	12 yr	Abdominal pain, cloacal exstrophy, imperforate anus, colostomy, ileal conduit	Mild bilateral hydronephrosis	Duplicated uterus, left hydrometrocolpos, obstructed rudimentary right uterus and vagina (Fig. 2)	Obstructed uterine didelphia, vaginas communicating into bladder plate
3	13 yr	Cyclic pain in left lower quadrant	Absent left kidney	Duplicated uterus and vagina, left hematometrocolpos (Fig. 3)	Uterine didelphia, distal left vaginal obstruction
4	16 yr	Monthly pain in right lower quadrant	Absent right kidney	Duplicated uterus, septated vagina, right hydrocolpos (Fig. 4)	Uterine didelphia, septated vagina, distal right vaginal obstruction
5	17 yr	Monthly lower abdominal pain	Absent left kidney	Duplicated uterine fundi with left hematometros, complex heterogeneous material in cul de sac (Fig. 5)	Bicornuate uterus with obstructed left horn, endometriosis
6	11 yr	Imperforate anus, lower abdominal pain, vaginal stenosis	Absent right kidney	Duplicated uteri, right hydrocolpos	Uterine didelphia, left hydrocolpos extending to right pelvis
7*	10 yr	Pain in right lower quadrant, pelvic mass	Absent right kidney	Duplicated uterus and vagina with right hydrometrocolpos	Uterine didelphia, right hydrometrocolpos
8	11 yr	Pain in right lower quadrant	Absent right kidney	Duplicated uterus, right hematometrocolpos	Uterine didelphia, right hematometrocolpos
9	14 yr	Abdominal pain, hematuria, urinary tract infection, imperforate anus	Horseshoe kidney with single ureter	Dilated obstructed rudimentary uterus	Normal right uterus, left hydrocolpos
10	5 d	Ring 18 chromosome, imperforate anus	Crossed fused ectopia	Duplicated uterus, hydrometros (left > right) (Fig. 6)	No surgery
11	6 yr	Urinary tract infection, difficulty voiding	Absent right kidney	Rudimentary uterus, sacral defect (Fig. 7)	Vaginal introitus covered by tissue, normal vaginal canal
12	4 yr	Urinary tract infection, malrotation	Absent right kidney	Absent uterus, ovaries present	Absent uterus, normal ovaries

* See reference 11.

vagina) in whom the obstruction can only be suspected based on mass effects or the inability to find an appropriate orifice.

In two of our patients (patients 1, 5), US permitted detection of an adnexal mass in addition to the vaginal and uterine duplications. At surgery, a serous cystadenoma was found in patient 1 and an endometrioma in patient 5. Pubertal women with obstructed genital structures have a higher incidence of backflow of menstrual blood through the fallopian tubes into the peritoneal cavity resulting in brownish deposits studding the pelvic organs and peritoneum, resembling endometriosis (7). Therefore, endometrioma should be suspected in girls with the Mayer-Rokitansky syndrome and an adnexal mass. A water enema examination may help clarify these confusing pelvic findings. Other unusual complications have been reported in women with the Mayer-Rokitansky syndrome, including a fibromyoma arising from a right uterine bud and inguinal or femoral hernias containing ovaries, fallopian tubes, or uterine remnants (21, 22).

In summary, we recommend real-

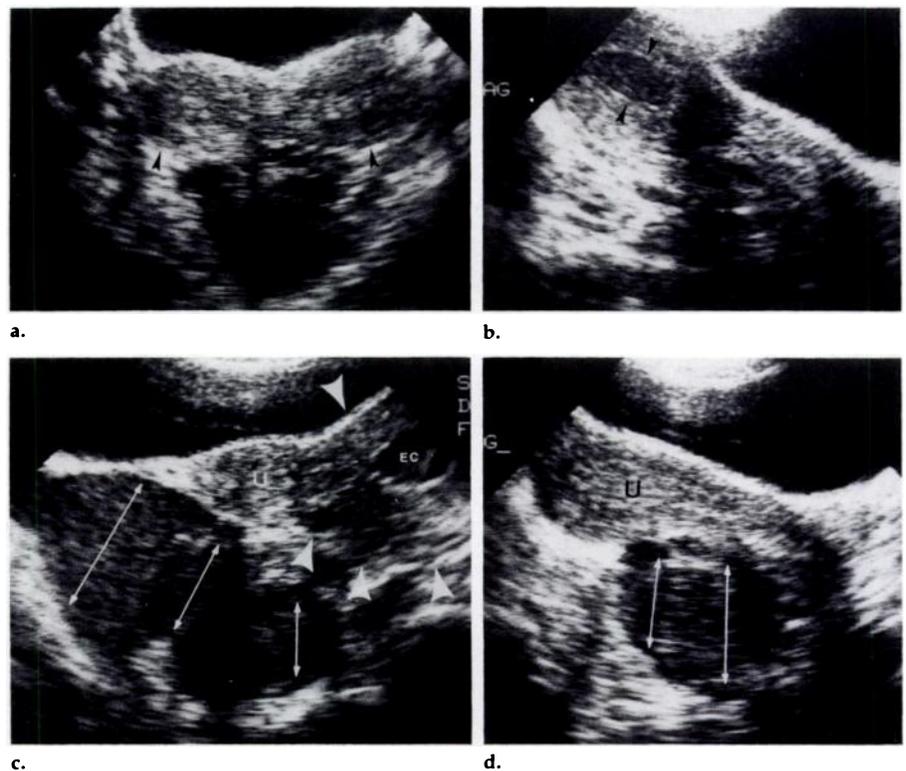


Figure 5. Patient 5. (a) Transverse scan of the pelvis shows two uterine fundi (arrowheads). (b) Left parasagittal scan shows echogenic material within the endometrial canal (arrowheads) of the left uterus. Transverse (c) and sagittal (d) images show heterogeneous echogenic material in the cul de sac representing old blood and endometriosis (double-headed arrows). U = right uterus, arrowheads = obstructed left uterus, EC = endometrial canal.



Figure 6. Patient 10. (a) Transverse scan of the right (R) and left (L) uteri shows the obstructed uterine horns (arrowheads) containing a fluid-debris level. Sagittal scans of the right (b) and left (c) uterus also show these findings.

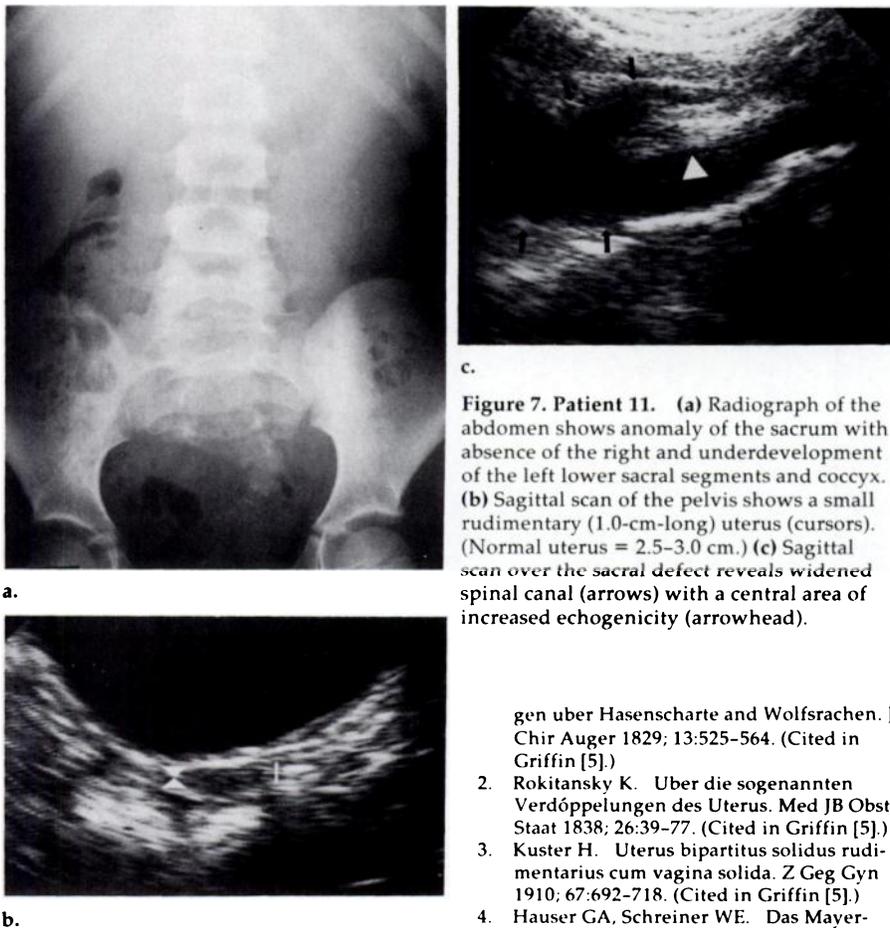


Figure 7. Patient 11. (a) Radiograph of the abdomen shows anomaly of the sacrum with absence of the right and underdevelopment of the left lower sacral segments and coccyx. (b) Sagittal scan of the pelvis shows a small rudimentary (1.0-cm-long) uterus (cursors). (Normal uterus = 2.5–3.0 cm.) (c) Sagittal scan over the sacral defect reveals a widened spinal canal (arrows) with a central area of increased echogenicity (arrowhead).

time US as the imaging modality for children clinically suspected of having genitourinary anomalies, prepubescent and postpubescent patients with lower abdominal or pelvic pain, and pubescent girls with severe dysmenorrhea unresponsive to medical therapy. ■

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