Mayer Rokitansky Kuster Hauser (MRKH) syndrome with absent thumbs and big toes

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Abstract

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare developmental failure of Müllerian ducts. Principle clinical features of MRKH syndrome are primary amenorrhoea associated with congenital absence of vagina, uterine anomalies, normal ovaries, 46 XX karyotype with normal female secondary sexual characteristics and frequent association with renal, skeletal, and other congenital anomalies.

A case of a 3-year-old child with congenitally absent thumbs and big toes is reported herein; she was brought in with complaints of urinary incontinence. Radiological investigation (ultrasound and magnetic resonance imaging (MRI) scan) revealed absent uterus and vagina while both ovaries were normal. Intravenous urography (IVU) study showed bifid pelvicalyceal systems bilaterally. Karyotyping revealed a 46 XX female phenotype. Laparoscopy confirmed normal ovaries bilaterally and small unfused uterine buds lying beside both ovaries on each side of pelvis. Early diagnosis of MRKH syndrome is essential for timely planning of vaginal and (if possible) uterine reconstructive surgeries.

Keywords: Müllerian agenesis, Vaginal atresia, Uterine agenesis, Mayer-Rokitansky-Kuster-Hauser syndrome.

Introduction

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a rare developmental failure of Müllerian ducts, comprising of congenital absence of vagina (described by Mayer in 1829[1]) with uterine anomalies (described by Rokitansky in 1838[2] and Kuster in 1910[3]); ovaries are usually normal in these patients. The prevalence has been reported at 1 in 4000 to 1 in 5000 live births,[4,5] but considering the women presenting with primary amenorrhoea, the disorder is fairly common, constituting 15% of patients with primary amenorrhoea.[5,6]

Case Report

A 3-year-old female child was brought in with complaints of total urinary incontinence. Her general physical examination showed absent thumbs and big toes bilaterally (Figure-1)

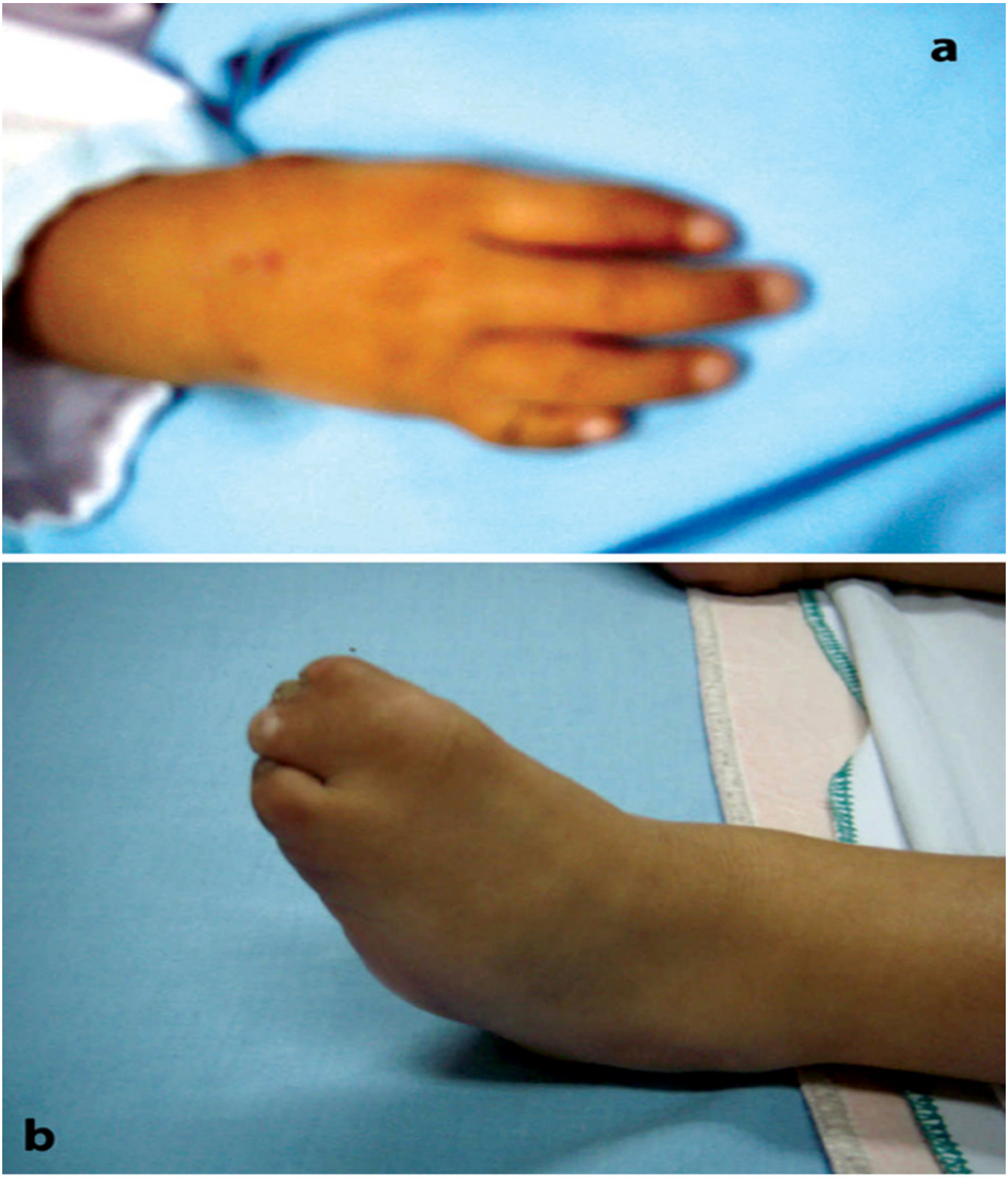

Figure-1: (a)Absent thumb and (b) absent big toe

.Rest of the physical examination was unremarkable. Perineal examination showed normal clitoris, and normal labia majora and minora. Wide urethral opening with urinary leakage in the introitus was noted. Vaginal opening was not seen. Normally-situated external anal opening was identified.

Complete blood picture and renal function tests were carried out and were found to be within normal

limits. During ultrasound examination of pelvis, the uterus was not identified while normal ovaries were noted bilaterally. Ultrasound examination of abdomen depicted normal size, shape and echogenicity of both kidneys. Rest of the abdominal ultrasound scan also appeared unremarkable. For confirmation of the uterine absence, a magnetic resonance imaging (MRI) examination was carried out (Figures-2, 3a, 3b)

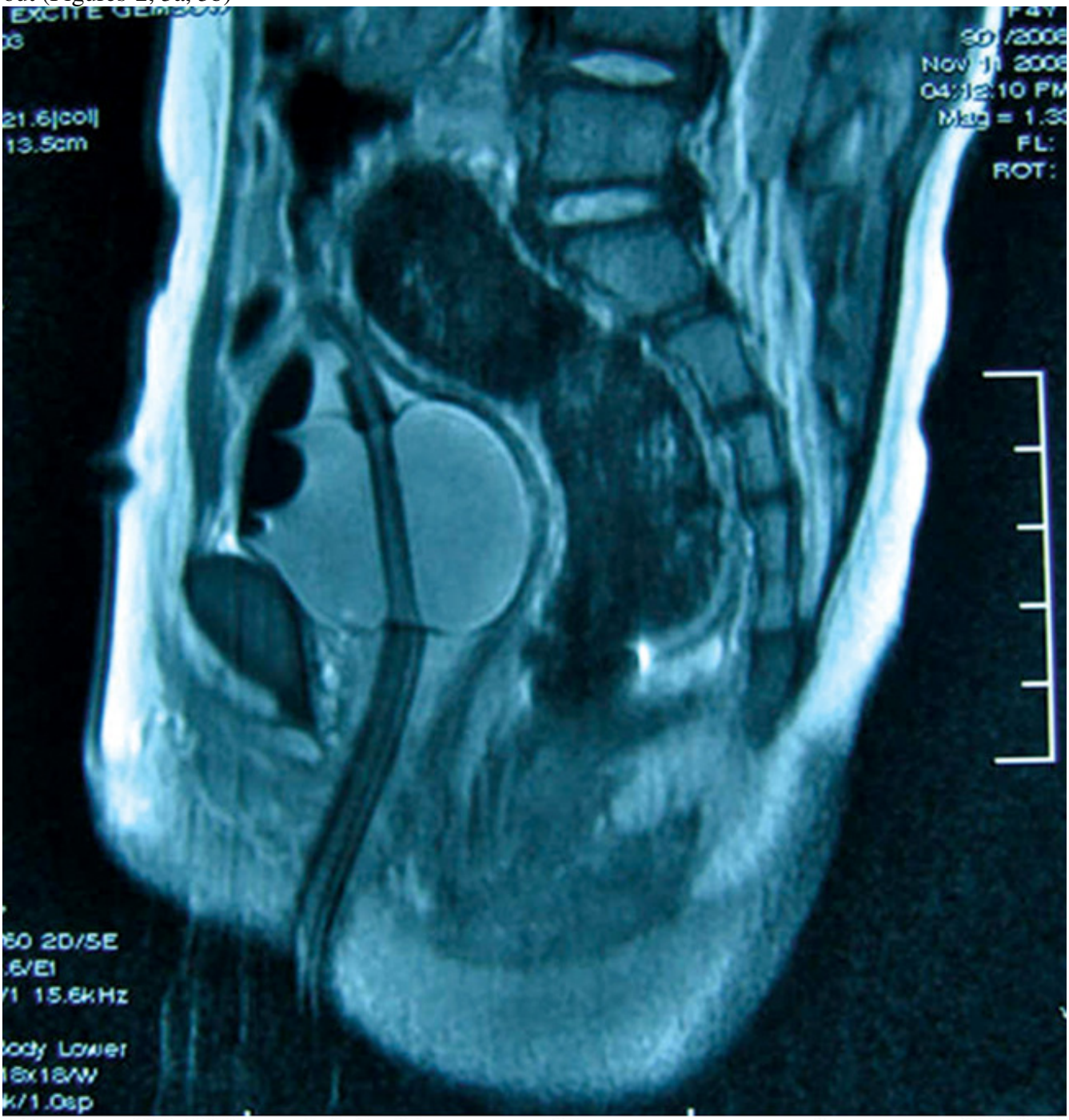

Figure-2: T2-weighted magnetic resonance images (MRI) of the pelvis. Mid-line sagittal section is showing normal urinary bladder anteriorly, rectum posteriorly and complete absence of the uterus and vagina.

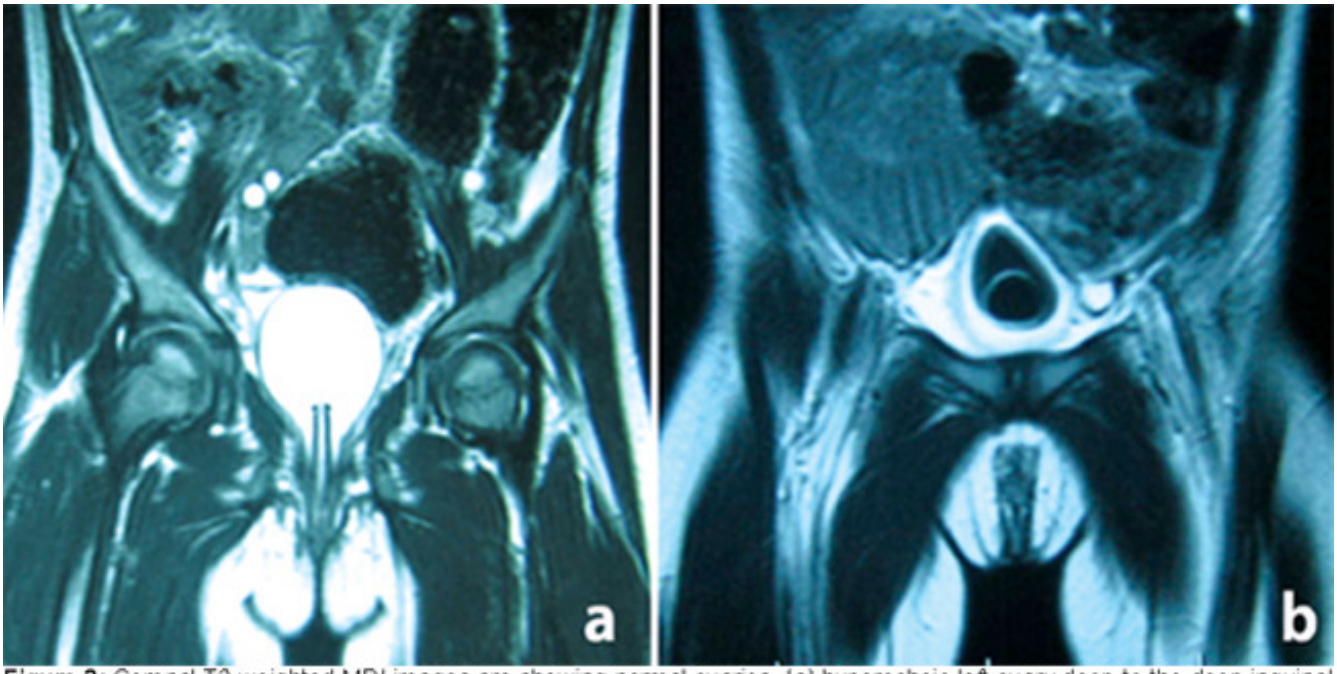

Figure-3: Coronal T2-weighted MRI images are showing normal ovaries, (a) hyperechoic left ovary deep to the deep inguinal ring, (b) two follicles are noted in right ovary which is lying inferiomedial to right iliopsoas muscle.

which confirmed the absence of the uterus and the vagina. Both ovaries had a normal appearance (isointense on TI-weighted images and hyperintense on T2-weighted images). The right ovary was normally placed and contained two follicles as well, while the left ovary was close to the deep inguinal ring and appeared solid (without follicles).

Intravenous urography (IVU) revealed bilateral bifid pelvicalyceal system. Karyotyping showed normal 46, XX female phenotype.

Laparoscopic findings (Figure-4A, 4B and 4C)

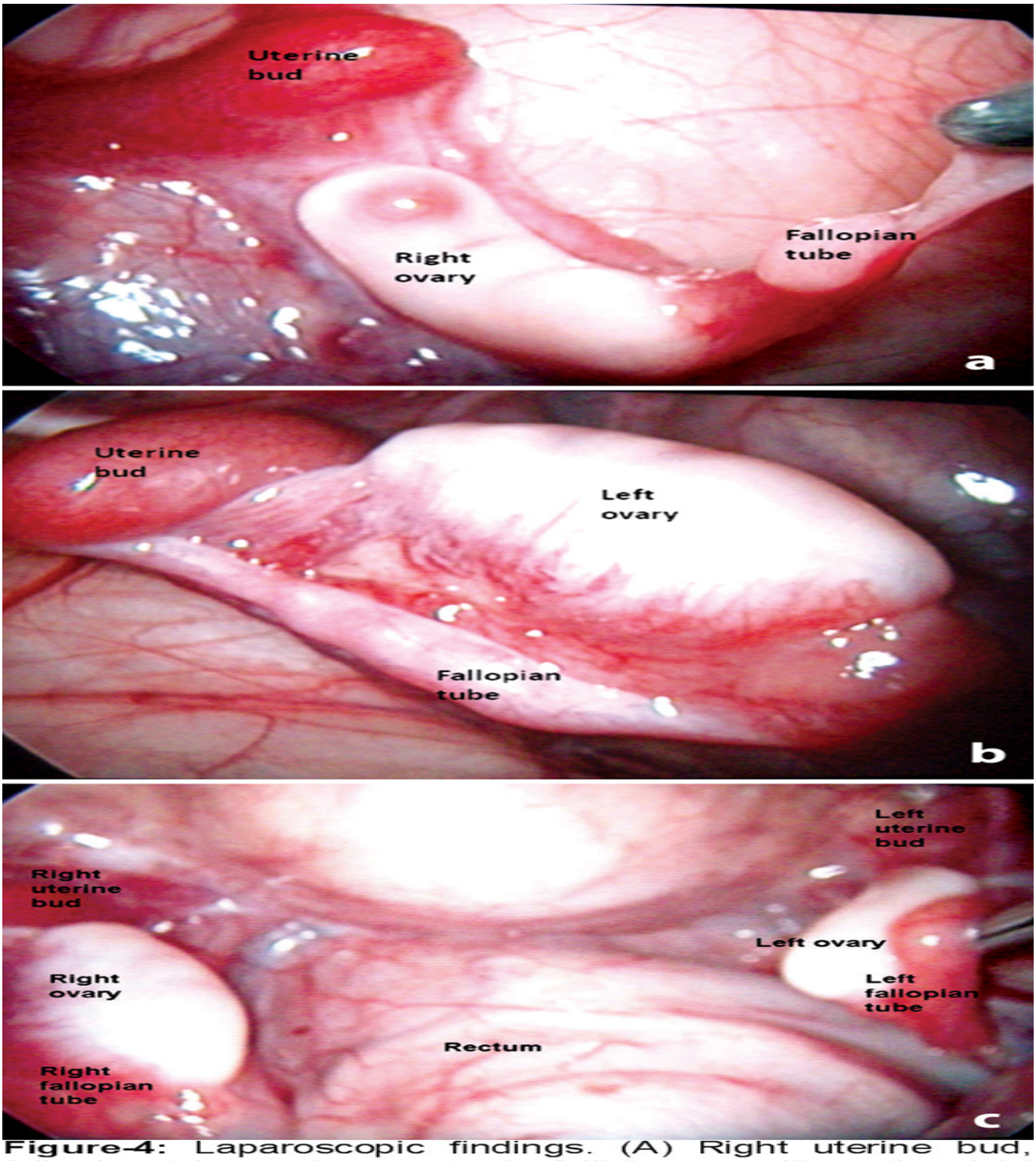

Figure-4: Laparoscopic findings. (A) Right uterine bud, fallopian tube and ovary (note follicle as well) on the right side of the pelvis. (B) Unfused left uterine bud, fallopian tube and ovary on the left side of the pelvis. (C) Superior view of the pelvis shows unfused uterine buds, fallopian tubes and ovaries lying on each side of the pelvis.

revealed about 1.2 x 0.5 cm sized right ovary with the fallopian tube and a small uterine bud on the right side of the pelvis. Presence of follicles in the right ovary was also confirmed. About 1.5 x 0.6 cm sized left ovary with fallopian tube and an unfused uterine bud were identified near deep inguinal ring on the left side. The vagina was absent.

Discussion

Absence of vagina and uterus (genital abnormalities) in the presence of normal ovaries and normal female karyotype with renal and skeletal anomalies has been recognized as Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH syndrome).

Principle clinical features of the MRKH Syndrome as described by James E. Griffin and et al[4] include:

* Primary amenorrhoea associated with congenital absence of vagina;
* 46 XX karyotype;
* Uterus that varies from anatomically complete (rudimentary bicornuate) to complete absence;
* Normal ovarian function and normal ovulation;
* Normal female secondary sexual characters like breast development, body proportion and body hair;
* And frequent association of renal, skeletal and other congenital anomalies. Abnormalities other than renal and skeletal anomalies have also been reported to occur in MRKH syndrome, including inguinal and femoral hernias, congenital heart diseases, cleft palate, situs inversus, bowel malrotation and anorectal malformation.[4,5]

A spectrum of Müllerian duct defects occurs in patients with MRKH syndrome. Ovaries and fallopian tubes are usually normal in these patients. Anomalies include vaginal agenesis or hypoplasia, accompanied by variable anomalies of uterus, urinary tract and the skeletal system. In 90% of the cases, complete vaginal agenesis is associated with uterine agenesis, while in 10% of the affected women, isolated vaginal agenesis may occur with an obstructed or non-obstructed rudimentary uterus.[4,5] Strübbe et al have classified typical or atypical forms of this syndrome as type A and type B.[7] Patients with type A have symmetric uterine muscular buds and normal fallopian tubes, while patients with type B have asymmetric uterine muscular buds, abnormally developed fallopian tubes, renal and skeletal abnormalities.[7] Uterine anomalies that can occur include unfused separate uterine buds, unicornuate uterus with communicating or non-communicating horn (which results in haematometra in the non-communicated horn), uterus didelphys, bicornuate uterus and septate uterus.[8]

Association between congenital absence of vagina and developmental abnormalities of urogenital system was recognised very early, and approximately one-third of the patients have renal anomalies.[2,3] Among these patients, 74% had either unilateral renal agenesis or ectopia of one or both kidneys, 5% had horseshoe kidneys, 13% had abnormalities of the collecting system, 5% had malfunctioning kidneys of uncertain cause, and renal malrotation.[4,9] Some affected patients may have lethal manifestation of bilateral renal agenesis.[4]

Skeletal abnormalities are recognised findings in MRKH syndrome.[2,3] The incidence is about 12%, among which 68% of the patients have spinal anomalies, 14% have limb anomalies and 10% have rib deformities.

Common spinal abnormalities that occur in this syndrome include scoliosis, spina bifida, transitional vertebral defects and vertebral segmentation defects including Klippel-Feil deformity, hemivertebra, butterfly vertebra and others.[4,10] Limb abnormalities include malformed arm or leg in 9% of the patients, syndactyly in 4%,[4] and absence of a thumb.

Embryologically, the fundamental abnormality in MRKH syndrome appears to be arising from the mesonephric kidney system, the development of which is interrupted during the 6th or 7th week of gestation. Wolffian ducts develop from the mesonephric kidney system (mesoderm), which leads to the development of uterus and upper vagina. Metanephric duct also develops from the mesonephric urinary bladder (in part).

Like the mesonephros, the skeletal system is also derived from the mesoderm, at approximately the same stage of development and at the same somite levels which are involved for the embryonic

development of urogenital tract, which explains the skeletal abnormalities.[4]

Another hypothesis that has been proposed by Schmid-Tannwald and Hauser[11] to explain the association between genital and renal anomalies in this syndrome is that the faulty gonadal differentiation due to production of Müllerian inhibiting factor induces the regression of Müllerian ducts.

Conclusion

Early diagnosis of MRKH syndrome is necessary for planning timely reconstructive vaginal, and if possible uterine, surgery. Awareness of the patient and related family members about the anomaly is essential so that they are relieved of the extreme psychological and mental stress that can arise from the situation when it is discovered while treating infertility after marriage. Ultrasound is the modality that leads to the diagnosis, but MRI is usually needed for confirmation of the uterovaginal anomalies and ovarian evaluation.

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