BMC Women's Health

Case report

Open Access

Congenital scoliosis associated with agenesis of the uterine cervix. Case report

Giorgio Carlomagno*†[1], Arturo Di Blasi†[2] and Matteo Della Monica†[3]

Address: [1]Università di Roma La Sapienza, Corso di Laurea in Ostetricia, Unità Operativa Ginecologia e Ostetricia, Azienda Ospedaliera "Gaetano Rummo", Via dell'Angelo 1, 82100 Benevento, Italy, [2]Unità Operativa Anatomia Patologica, Azienda Ospedaliera "Gaetano Rummo", Via dell'Angelo 1, 82100 Benevento, Italy and [3]Unità Operativa Genetica Medica, Azienda Ospedaliera "Gaetano Rummo", Via dell'Angelo 1, 82100 Benevento, Italy

Email: Giorgio Carlomagno* - giorgio.carlomagno@uniroma1.it; Arturo Di Blasi - arturo.diblasi@ao-rummo.it; Matteo Della Monica - matteo.dellamonica@ao-rummo.it

* Corresponding author †Equal contributors

Published: 30 June 2004

BMC Women's Health 2004, **4**:4 doi:10.1186/1472-6874-4-4

Received: 26 April 2004
Accepted: 30 June 2004

This article is available from: http://www.biomedcentral.com/1472-6874/4/4

Abstract

Background: Alterations in the normal sequence of development of müllerian ducts lead to a wide spectrum of reproductive tract abnormalities. A rare form of lack of development, regarding a short tract of the müllerian ducts, leads to the isolated agenesis of the uterine cervix. Anomalies identified among patients with müllerian agenesis include skeletal deformities (i.e., scoliosis of the spine and Klippel-Feil anomaly).

Case presentation: A 46 years old woman presenting cyphoscoliosis and very low stature (120 cm – 3,93 feet), came to our observation for acute pelvic pain; she also reported primary amenorrhoea associated with cyclic pelvic pain. Clinical and imaging evaluation, evidenced a blind vaginal duct of normal length, left cystic adnexal mass, and enlarged uterus with hematometra. FSH, LH, 17β estradiol and CA-125, karyotype and radiographic study of limbs and vertebral column were also evaluated.

At laparotomy, a left ovarian cyst was found. Uterus ended at the isthmus; under this level a thin fibrous tissue band was found, joining the uterus to the vagina. Uterine cervix was replaced by fibrous tissue containing some dilated glands lined with müllerian epithelium. Karyotype resulted 46, XX. The described skeletal deformity, were consistent with Klippel-Feil syndrome.

Conclusion: We report a case of congenital scoliosis associated with müllerian agenesis limited to uterine cervix, association thus far seen only among patients with Mayer-Rokitansky-Kuster-Hauser syndrome (utero-vaginal agenesis). This case report supports the necessity to evaluate, for accompanying müllerian anomalies, all cases of congenital structural scoliosis in view of the possibility for many müllerian development abnormalities, if timely diagnosed, to be surgically corrected.

Background

The müllerian tract is the embryologic structure that develops into the internal female genitalia. Bilateral müllerian ducts grow medially and caudally to contact and fuse then abuts with the urogenital sinus. Alterations in the normal sequence of development leads to the wide spectrum of reproductive tract abnormalities classified as lack of development or lateral or vertical fusion defects.

The complete agenesis of uterus and vagina is known as Mayer-Rokitansky-Kuster-Hauser syndrome. A rare form of lack of development, regarding only a tract of the müllerian ducts, leads to the isolated uterine cervix agenesis [1,2]. Patients affected by this rare, "non communicating", abnormality have a functional uterus, but from menarche on, the blood is trapped: they therefore present as cases of primary amenorrhoea with cyclic pelvic pain, due to hematometra distending the uterine cavity. In most cases these patients are identified and diagnosed in the early adolescent period (12–17 years), but, since silent bleeding can occur for months to years before causing symptoms, other cases are diagnosed later, when lamenting primary amenorrhoea [3-5]. Furthermore müllerian duct development abnormalities may be seen in association with skeletal deformities like congenital scoliosis of the spine and Klippel-Feil anomaly [6-8].

Case presentation

We report the case of a 46 years old woman seen in emergency for acute pelvic pain. The patient's history evidenced primary amenorrhoea, never investigated before, and associated with cyclic pelvic pain. There was not any history of in-utero DES exposure. The patient reported a right adnexectomy, done 30 years before to remove an ovarian cyst. She did not save any record of this laparotomy, done when she was a 17 years old adolescent with primary amenorrhoea. She had been married and sexually active, but sterile; possible causes of sterility were never investigated. Physical examination evidenced low stature (120 cm – 3,93 feet), short neck and thoracic cyphoscoliosis; breast development and other sexual characters were normally present. Pelvic clinical examination evidenced a normal vagina, ending in a "cul de sac" where uterine cervix was absent; above this level an enlarged uterus was palpable. The left ovary was enlarged due to a cystic mass. Ultrasound and computed tomography scan imaging confirmed the presence of a left cystic adnexal mass (58 × 49 mm) without evidencing any urinary tract anomalies. Uterine enlargement (96 × 78 mm) was caused by two leiomyomata and hematometra distending the uterine cavity.

Attention was given also to the skeletal abnormalities through radiographic study of limbs and vertebral column. Genetic work up included karyotype and radiographic evaluation of skeletal deformities; FSH, LH, 17β estradiol, and CA-125 where also evaluated. Karyotype resulted 46, XX. FSH, LH, 17β estradiol, and CA-125 where within range of normality. Radiographic study of skeletal deformities evidenced dysmorphism of carpal bones and radium epiphysis, and narrow angle cyphoscoliosis with fusion of cervical and upper thoracic vertebrae.

Due to the patient's age, the presence of an ovarian cyst greater than 50 mm, and the symptomatic uterine leiomyomatosis, a destructive surgical approach was proposed. The patient gave informed consent to abdominal hysterectomy and left oophorectomy.

At laparotomy a left ovarian cyst 50 × 50 mm (simple serous cyst at frozen section) was found and removed. Uterus, enlarged (100 × 85 mm) both because of leiomyomatosis and hematometra, ended at the isthmus. Under this level, after blunt dissection of bladder and rectum, a thin fibrous tissue band, 30 mm long 40 mm large, was found, joining uterus and vagina. On both sides of this band, a thin (4 × 2 mm) tubular structure distended by fluid content, was present. No findings related to endometriosis were observed at laparotomy. The uterus was removed sectioning the fibrous band just above the level of the blind vaginal cuff.

Gross examination of uterus (Fig. 1) evidenced a "non communicating" uterine cavity, filled with menstrual blood, and two intramural leiomyomata (30 mm each) among a diffusely hypertrophic miometrium. The cervix was absent, being in its place two nodular structures 1 cm in diameter joined by fibrous tissue. Microscopically, these cervical appendages presented intramural glandular structure often cystic, without an endocervical canal. These structures were lined by different types of epithelium; some cystic spaces were filled with mucous fluid and lined by flattened mucus secreting cells, as in Nabothian cysts (Fig. 2); glandular ducts and cysts lined by mono and bilayer non mucinous epithelium were also present; finally large dilated ducts with micro papillary intraluminal budding of pseudo stratified epithelium were present. All these epithelial aspects likely represented a diffuse glandular hyperplasia within a lack of development of müllerian ducts at the uterine cervix level. In histological evaluation ectopic presence of endometrial glands and stroma was not evidenced.

Discussion

The skeletal deformities described above – short neck, short stature, vertebral fusion (fig. 3), thoracic cyphoscoliosis -, were consistent with Klippel-Feil syndrome, without malformation of the temporal bones and ossicles and deafness [6].

Klippel-Feil syndrome is a rare disorder caused by failure of the division of the bones in the cervical (neck) section of the spinal column during embryonic development. Common signs are short neck, impaired movement of the head and neck especially from side to side and a low hairline at the back of the neck. The extent to which individuals are affected by these features can vary widely. The association of absent vagina and Klippel-Feil syndrome

Figure 1
Uterus: non communicating cavity, hematometra and two leymiomata are visible. One of the two cervical appendages is visible in the right upper side. A serous cyst (lined with mesothelial cells) is in the low left side.

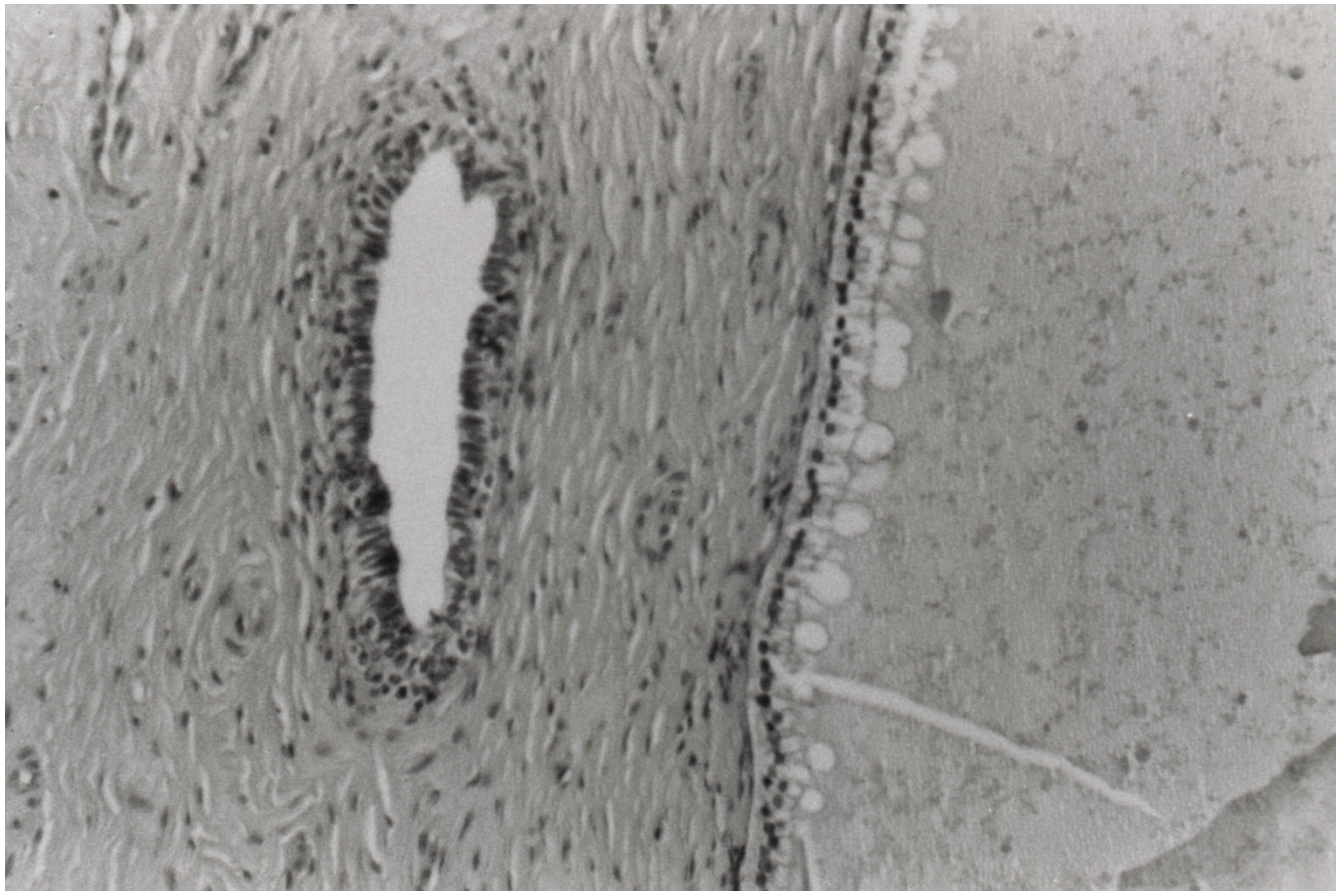

Figure 2
A glandular structure in the cervical appendage lined by flattened mucus secreting cells

was first reported by Baird [6]. Fisher [8] first reported the case of a woman with the müllerian agenesis known as Mayer-Rokitansky-Kuster-Hauser syndrome, typical form (no renal deformities), associated with scoliosis of the thoracic and lumbar spine, an association thus far seen only among patients with atypical form (renal deformities also present) of the syndrome [9]. We report the first case, at our knowledge, of Klippel-Feil syndrome associated with a müllerian agenesis limited to the uterine cervix.

Unfortunately the patient was misdiagnosed when surgically treated for ovarian cyst at the age of 17, and thoroughly investigated only presently. Her age, and associated pathology, indicated a destructive surgical treatment. In a different setting, seen the small distance separating the two structures, a reconstructive surgery could have been planned, in order to anastomose vaginal cuff to uterine cavity. Waiting for surgical treatment, and in order to give relief of symptoms, menstrual outflow obstruction causing pain and mass effect to these patients, could eventually be treated with gonadotropin-releasing hormone agonist or oral contraceptives.

Conclusions

Attention should be paid to the diagnostic management of cases of congenital structural scoliosis and they should be evaluated for accompanying müllerian anomalies. Well before primary amenorrhoea occurs, as in present case, different imaging diagnostic tools (ultrasounds, computed tomography magnetic resonance imaging) may be of help. The vagina can be easily inspected by vaginoscopy, and give direct evidence of such malformations. Müllerian development abnormalities, if timely diagnosed may be corrected and functional and reproductive potential can be recovered.

Competing interests

None declared.

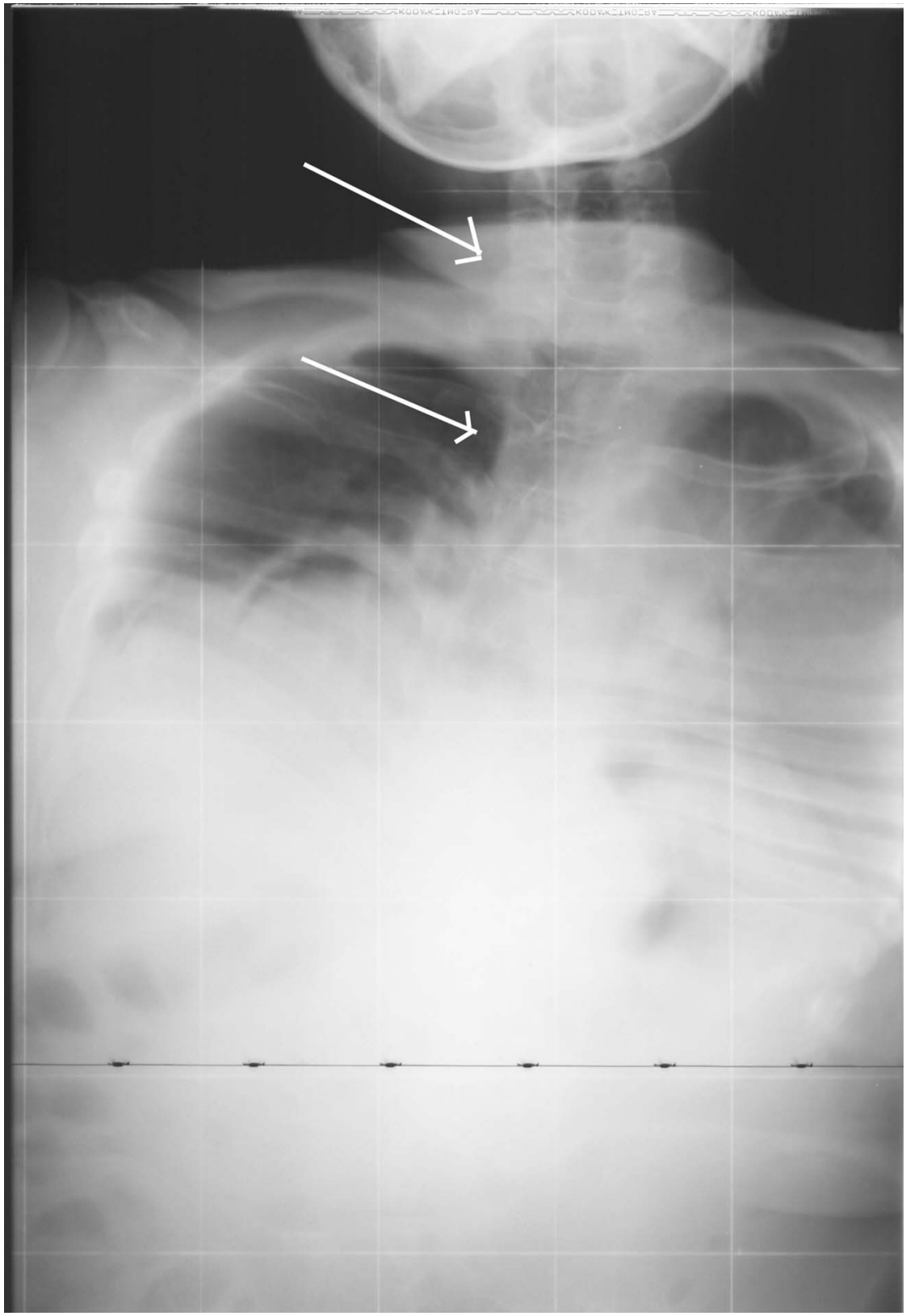

Figure 3
X-rays of the neck in antero-posterior view: arrows indicate vertebral X ray of cervical spine fusion.

Authors' contributions

All authors conceived of the study, and participated in its design and coordination. All authors read and approved the final manuscript.

Acknowledgements

Written consent was obtained from the patient or their relative for publication of study.

References

1. American Fertility Society: **Classification of adnexal adhesions, distal tubal occlusion secondary to tubal ligation, tubal pregnancy and Müllerian anomalies and intrauterine adhesions.** *Fertil Steril* 1988, **49:**944-955.
2. Bruni V, Dei M: *Ginecologia dal periodo neonatale all'età evolutiva. Firenze, S.E.E* 2000.
3. Letterie GS: **Combined congenital absence of the vagina and cervix. Diagnosis with magnetic resonance imaging and surgical management.** *Gynecol Obstet Invest* 1998, **46:**65-7.
4. Gidwani G, Falcone T: *Congenital Malformations of the Female Genital Tract: Diagnosis and Management: Hardbound* Lippincott Williams and Wilkins; 1999.
5. Spence J, Gervaize P, Jain S: **Uterovaginal anomalies: diagnosis and current management in teens.** *Curr Women's Health Rep* 2003, **3:**445-50.
6. Baird PA, Lowry RB: **Absent vagina and Klippel-Feil anomaly.** *Am J Obstet Gynec* 1974, **118:**290-1.
7. Chawla S, Bery K, Indra KJ: **Abnormalities of urinary tract and skeleton associated with congenital absence of vagina.** *Br Med J* **5500:**1398-400. 1966, Jun 4
8. Fisher K, Esham RH, Thorneycroft I: **Scoliosis Associated With Typical Mayer-Rokitansky-Küster-Hauser Syndrome.** *South Med J* 2000, **93:**243-6.
9. Strubbe EH, Thijn CJP, Willemsen WNP, Lappohn R: **Evaluation of radiographic abnormalities of the hand in patients with the Mayer-Rokitansky-Küster-Hauser syndrome.** *Skeletal Radiol* 1987, **16:**227-231.

Pre-publication history

The pre-publication history for this paper can be accessed here:

http://www.biomedcentral.com/1472-6874/4/4/prepub