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Case Report

CASE REPORT

MURCS (Müllerian duct aplasia–renal agenesis–cervicothoracic somite dysplasia): a rare cause of primary amenorrhoea

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Abstract

The agenesis of the Müllerian duct is the second most common cause of primary amenorrhoea after Turner syndrome. The abnormal development of Müllerian duct often associates with the urinary tract and skeletal abnormalities. MURCS (Müllerian duct aplasia–renal agenesis–cervicothoracic somite dysplasia) association is a unique and rare developmental disorder with four common features of uterine hypoplasia or aplasia, renal agenesis or ectopy, vertebral anomalies and short stature. We report a case of young female with primary amenorrhoea. She had well-developed secondary sexual characteristics along with multiple congenital developmental abnormalities such as the absence of uterus, ectopic kidney, cervical vertebral fusion, hemivertebrae, scoliosis, cervical rib, facial asymmetry and growth retardation. Our case highlights the rarity and clinical importance of this syndrome. For the evaluation of primary amenorrhoea in a female with well-developed secondary sexual characteristics, congenital anomalies should be ruled out before hormone and karyotype analyses.

INTRODUCTION

Turner syndrome is the most common cause of primary amenorrhoea followed by Müllerian duct agenesis. The causes of primary amenorrhoea with well-developed secondary sexual characters are few such as Mayer–Rokitansky–Küster–Hauser (MRKH), MURCS association, VACTERL and Goldenhar syndrome. Developmental failure of Müllerian duct structures sometimes associates with the urinary tract and skeletal abnormalities. MURCS (Müllerian duct aplasia–renal agenesis–cervicothoracic somite dysplasia) association is a rare developmental disorder with four common malformations specifically described as uterine hypoplasia or aplasia, renal agenesis or ectopy, vertebral anomalies and short stature (<152 cm) [1, 2].

CASE REPORT

A 28-year-old female presented with short stature, recurrent abdominal pain and neck pain from teenage. She never underwent menarche (primary amenorrhoea). There was no such type of illness in family members. Physical examination revealed facial asymmetry, short neck, high arched palate, scoliosis and short stature (height 146 cm). Her secondary sexual characters were well developed with tanner A2P5 B5. The remaining examinations were unremarkable (Figs 1–3).

Haemogram and biochemical and hormonal analyses were normal. X-ray spine showed a fusion of C2–C3, C4–C5–C6 vertebrae, hemivertebrae, scoliosis of thoracic vertebrae and cervical rib. Ultrasound of the abdomen showed bilateral normal gonads,

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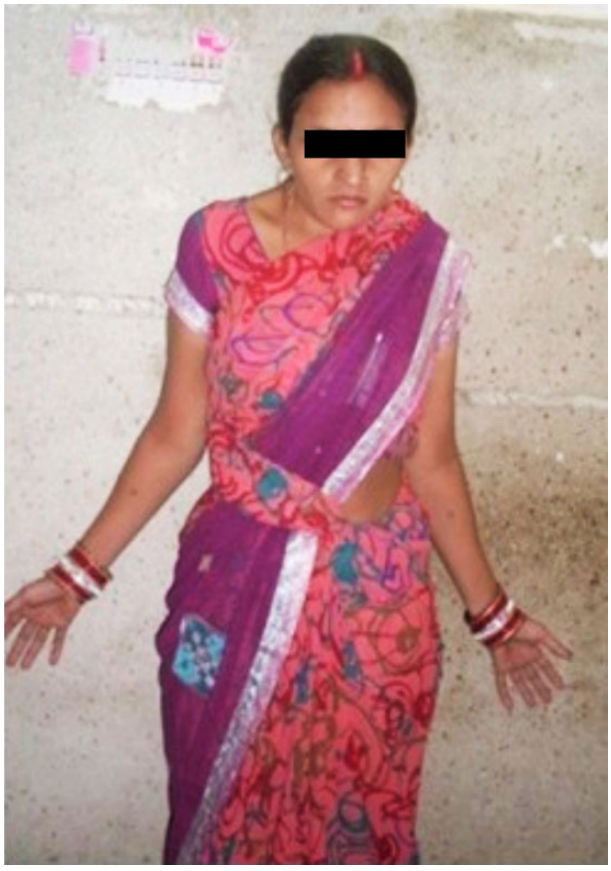

Figure 1: Image of female shows short neck and facial asymmetry.

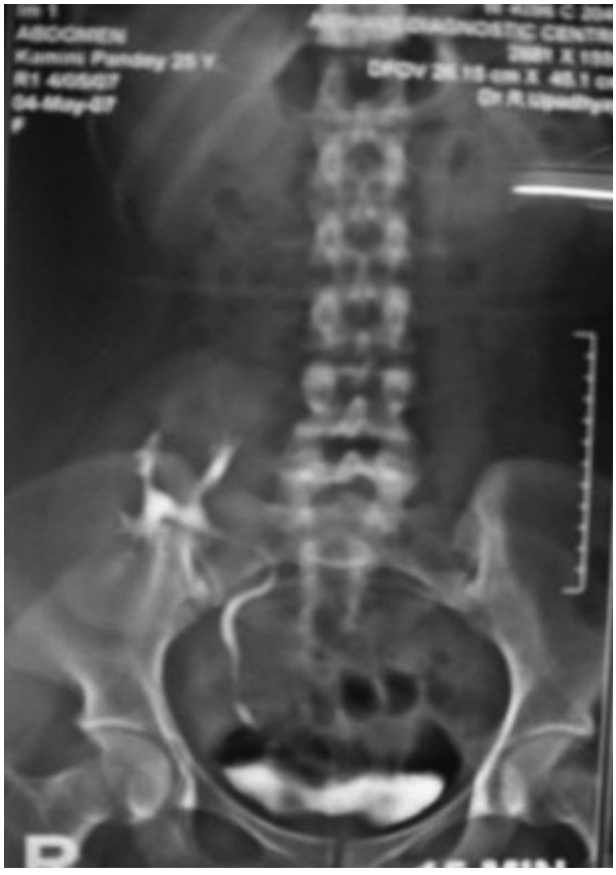

Figure 2: X-ray abdomen with an intravenous pyelogram shows ectopic kidney lying in the paralumbar region. The intravenous pyelogram showed non-functional left kidney. Hemivertebrae is present.

non-visualization of uterus and fallopian tubes; both kidneys (right—9.9 × 3.46 cm, left—3.7 × 1.74 cm) were ectopic lying in the paralumbar region. The intravenous pyelogram showed non-functional left kidney. She was counselled and prognosis of disease was explained.

DISCUSSION

MURCS syndrome is a rare developmental disorder characterized by primary amenorrhoea in a female with well-developed secondary sexual characteristics and other urological and somatic abnormalities [1, 2]. Once believed to sporadic, an increasing number of familial cases now supports the disease as an autosomal dominant trait with incomplete penetrance. So, the family members in addition to index case should be enquired of and investigations for renal and skeletal abnormalities are recommended. The family history was negative in our patient. The cause of this syndrome is still unknown [3]. A mutation in WNT4 was reported in a patient with absent Müllerian structures and mild hyperandrogenaemia. Our patient had clinicoradiological features of MURCS association such as aplasia of the uterus and fallopian tubes, ectopic kidney and cervicothoracic vertebral abnormality. The urological abnormalities include unilateral renal agenesis (23–28%), ectopia of one or both kidneys (17%), renal hypoplasia (4%), horseshoe kidney and hydronephrosis [4, 5]. Apart from the classical presentations of MURCS, other clinical features are facial asymmetry, cleft lip and palate, micrognathia, abnormal external ear, deafness, Sprengel's shoulder, rib abnormalities, maldevelopment of upper limb and gastrointestinal abnormalities [1–6].

The closest differential diagnoses of MURCS association are Goldenhar syndrome, VACTERL association and Turner syndrome. Goldenhar syndrome is characterized by the abnormal congenital development of mandible, nose, lip, soft palate and ear [7]. Urogenital abnormalities are rare in Goldenhar syndrome. VACTERL anomaly is characterized by vertebral defects, limb abnormalities, renal anomalies, anal atresia, tracheoesophageal fistula and cardiac defects. It is rarely associated with genital malformation and vertebral anomalies are more common in caudal region. Turner syndrome is associated with primary ovarian failure and undeveloped secondary sexual characters.

The pathogenesis of MURCS association is not known. At the end of the fourth week of foetal life, the blastemas of cervicothoracic somite, arm buds and pronephric buds are in close proximity to each other. MURCS association may be due to teratogenic effect at this time, which affects the relations within these blastemas [8]. The prognosis of such case varies and is probably dependent on the extent and severity of renal abnormalities.

To conclude, for the evaluation of primary amenorrhoea in well-developed secondary sexual characters, congenital anomalies should be ruled out before hormone and karyotype analysis. Our case highlights the rarity and clinical importance of MURCS association.

AUTHORS' CONTRIBUTIONS

All the authors contributed to prepare this study.

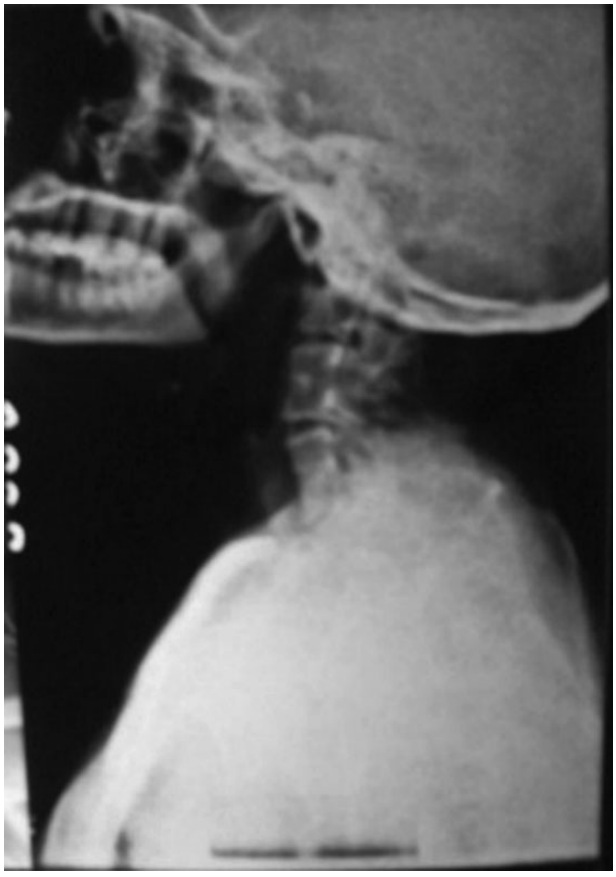

Figure 3: X-ray spine shows a fusion of C2–C3, C4–C5–C6 vertebrae.

CONFLICT OF INTEREST STATEMENT

None declared.

FUNDING

No funding support was received for this article.

ETHICAL APPROVAL

We have followed the ethical norms and have taken proper informed consent from the patient and relatives. Our patient participated voluntarily and did not suffer any harm. We confirm that all the research meets the ethical guidelines, including adherence to the legal requirements of the study country.

CONSENT

We have obtained written, informed patient consent for publication of the report and any accompanying images.

GUARANTOR

S.K. is a guarantor of this study.

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