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MURCS association and anorectal malformation: Case report of a female newborn

Sofia Morão [a,*], Fernando Chaves [b], Daniel Virella [b], Fátima Alves [c], Rui Alves [a], João Pascoal [a]

[a] Pediatric Surgery Department, Hospital Dona Estefânia, Centro Hospitalar Lisboa Central, Portugal
[b] Neonatal Intensive Care Unit, Hospital Dona Estefânia, Centro Hospitalar Lisboa Central, Portugal
[c] Pediatric Surgery Department, Urology Unit, Hospital Dona Estefânia, Centro Hospitalar Lisboa Central, Portugal

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ABSTRACT

MURCS association is rare, first described by Duncan et al. in 1979, including Müllerian duct aplasia, renal aplasia and cervicothoracic somite dysplasia. Levitt and Peña described in 2007 a classification of syndromic anorectal malformation (ARM) that associates these two entities. The reported case is the first one described in neonatal period. We describe a case of a female newborn with suspected diagnosis of anorectal and renal malformations. Physical and radiologic investigation revealed agenesis of sacrum and coccyx, tethered cord, left multicystic renal dysplasia, absence of vaginal orifice and hymen, normally placed urethral orifice and abnormal anal opening at the vaginal introitus as a rectovestibular type fistula. Also, she had right uterine, tube and ovary agenesis with a normal 46, XX female karyotype. A left diversing colostomy was done in first day of life and four months later, was performed a posterior sagittal anorectoplasty (PSARP), with intra-operative identification of a duplication of the distal rectum (related with caudal regression syndrome type 2). There were no complications in postoperative period. A staged management strategy is a viable option avoiding further complications in an already poor prognosis situation.

1. Introduction

MURCS association, first described by Duncan et al., in 1979, is a rare and nonrandom constellation of findings that includes Müllerian duct aplasia, renal aplasia and cervicothoracic somite dysplasia [1]. Anorectal malformations (ARM) comprise a wide spectrum of diseases that involve the distal anus and rectum as well as the urinary and genital tracts [2,3]. Levitt and Peña described in 2007 a classification of syndromic ARM that associates these two entities [3]. As it is extremely rare, high suspicion is essential for diagnosis and surgical orientation. We present a case diagnosed in neonatal period which poses several concerns regarding growth, urinary and bowel control.

2. Case report

A 40-year-old G3P3 presented at 39 weeks and 3 days gestation with premature rupture of membranes 36 h before delivery with clear amniotic fluid. Fetal ultrasound at 34 weeks of gestation suggested presence of multicyst left kidney. Because of suspected fetal distress, a caesarian delivery was performed. After delivery, the neonate needed non-invasive ventilation with quick recovery of heart rate. Apgar scores were 6 at 1 min but 8 at 5 min. The neonate had female phenotypic appearance and weighted 2580 g. Physical examination at that point revealed absence of anal orifice so it was decided to transfer her to a tertiary care pediatric hospital. On presentation, the neonate was hemodynamically stable. The lateral abdominal x-ray revealed agenesis of sacrum and coccyx (Fig. 1).

Renal and pelvic ultrasound suggested presence of tethered cord and showed left multicystic renal dysplasia (Fig. 2).

Transfontanelar ultrasound didn't reveal any associated malformations. The echocardiogram demonstrated patent ductus arteriosus, tricuspid and pulmonary regurgitation. After observation by a pediatric surgeon, who confirmed the presence of ARM

* Corresponding author. Pediatric Surgery Department, Hospital Dona Estefânia, Centro Hospitalar Lisboa Central, Rua Jacinta Marto, 1169-045, Lisboa, Portugal.
E-mail address: sophia.morao@gmail.com (S. Morão).

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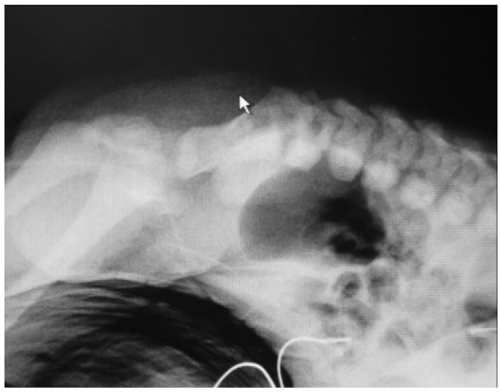

Fig. 1. Lateral x-ray.

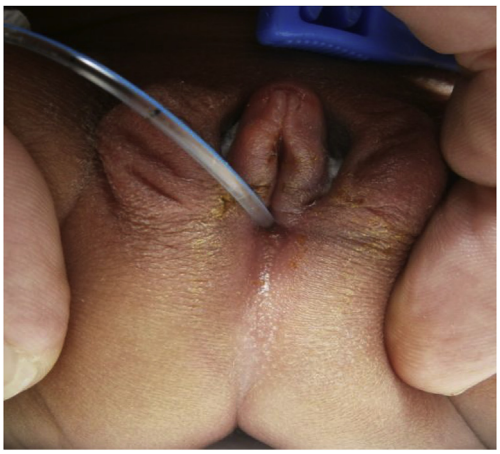

Fig. 3. Perineal inspection.

and it was decided to perform a left diversing colostomy. Under general anesthesia, the genital inspection, performed before the procedure, detected the absence of normal vaginal orifice and hymen, normally placed and permeable urethral orifice, and finally, an abnormal anal opening at the vaginal introitus as a variation of a rectovestibular type fistula (Figs. 3 and 4).

Intraoperatively, it was observed right uterine, tube and ovary agenesis with left structures intact (Figs. 5–7). Further investigation showed a normal 46, XX female karyotype, right grade I vesicoureteral reflux (VUR) in cystography (according to the International Reflux Committee classification) (Fig. 8), left kidney with 2% function in MAG3 renogram. The distal pressure colostogram revealed the presence of an abnormal reflux of contrast to the vaginal lumen (Fig. 9).

At four months of age, it was performed a PSARP procedure with interoperative identification of a 2-cm-long tubular duplication of rectum (Figs. 10–12). There were no complications in the postoperative period. At the most recent appointment, the child is one year old and is growing well; she is under anal dilatation scheme. Colostomy is working properly and she wears diapers (Fig. 13).

3. Discussion

In females, fallopian tubes, uterus and upper third of vagina develop from Müllerian ducts while lower two thirds of vagina and vaginal vestibule develop from lower part of definitive urogenital sinus [4]. Malformations of the uterus or vagina are attributed to abnormalities of fusion or regression of the caudal ends of the müllerian ducts.

In this case, we have right ovary, fallopian tube and hemi-uterus agenesis which excludes Mayer-Rokitansky-Küster-Hauser syndrome (vaginal agenesis, uterine malformation but normal tubes and ovaries). Moreover, right müllerian structures agenesis with left renal dysplasia excludes Herlyn-Werner-Wunderlich syndrome (müllerian ducts malformations with ipsilateral renal anomaly).

MURCS association is named when nonrandom combination of malformations (müllerian, renal and cervicothoracic) is formed together. The etiology of MURCS association is not known. The karyotype was normal in all investigated cases, including ours [1,5]. The presented case has all the classical findings described by Duncan et al. in 1979, except for vertebral anomalies which are seen in lower part of column. However, as it is mentioned on literature, identification of one component of the MURCS association suggests the presence of the other associated anomalies which may not be noticed until later in life. The child described in this paper has only 13 months of age. Duncan hypothesized about a close spatial relationship at the end of the fourth week of fetal life between cervical-upper thoracic somites, arm buds, and pronephric ducts. An error at this stage would cause the maldevelopment of vertebrae and kidneys and organ systems originating from müllerian ducts [1].

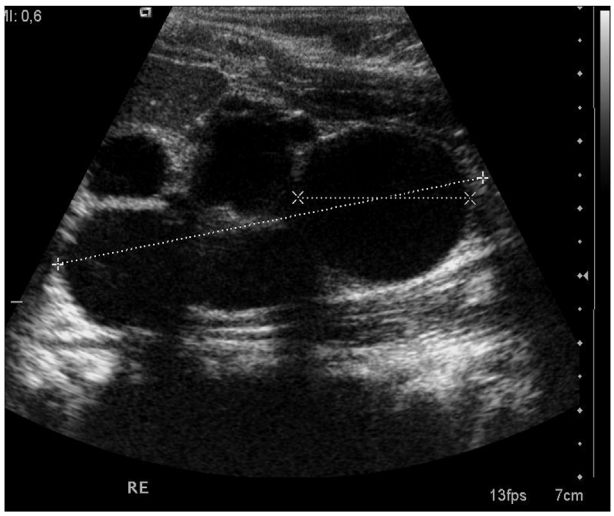

Fig. 2. Renal ultrasound.

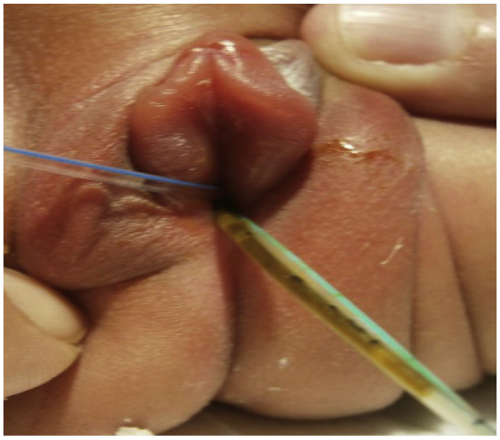

Fig. 4. Perineal inspection.