



Case Report

# Antenatal Diagnosis of Cloacal Dysgenesis Sequence by Fetal Magnetic Resonance Urography: Is It Incompatible with Fetal Life?

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## Abstract

Cloacal dysgenesis sequence (CDS) is a lethal and rare congenital malformation; usually requiring termination of gestation. As prenatal ultrasonography remains a method with doubtful results, we used Fetal Magnetic Resonance Urography (FMRU) for the evaluation of a fetus at 18 weeks of gestation, suspected to cloacal anomalies. Prenatal imaging of the patient; showed smooth perineum, bilateral hypoplastic kidneys, dilated urinary bladder and absence of anal, urethral and vaginal openings; while prenatal sonographic investigation merely showed a mesenteric cyst. Gestation was culminated and no vesicocolon fistula was detected in the abortus. In the current study, FMRU provided larger number of anomalies and showed to be more sensitive in evaluation of urogenital anatomy compared to the results obtained from sonography.

**Keywords:** prenatal diagnosis, fetal magnetic resonance urography (FMRU), cloacal dysgenesis syndrome (CDS).

## Introduction

Cloacal dysgenesis sequence (CDS) is an infrequent but distinctive malformation with a wide spectrum of clinical manifestations, which occurs in about 1 out of 50,000 live-born infants (Warne, Chitty & Wilcox 2002) that results in termination of pregnancy in most of the cases (Sahinoglu et al. 2004). It is important to reveal a short review of embryology to understand the pathogenesis

of CDS. Embriologically, by the sixth week of gestation, the cloaca is divided into a urogenital sinus disposed in the anterior part and a separate hindgut in the posterior section. These divisions occur by proliferation of folds originated from the caudal part of urorectal septum. After the separation of the urogenital sinus from the cloaca, the urethra is developed from the caudal end of the sinus. If the development is arrested at any stage, a wide range of urogenital anomalies such as

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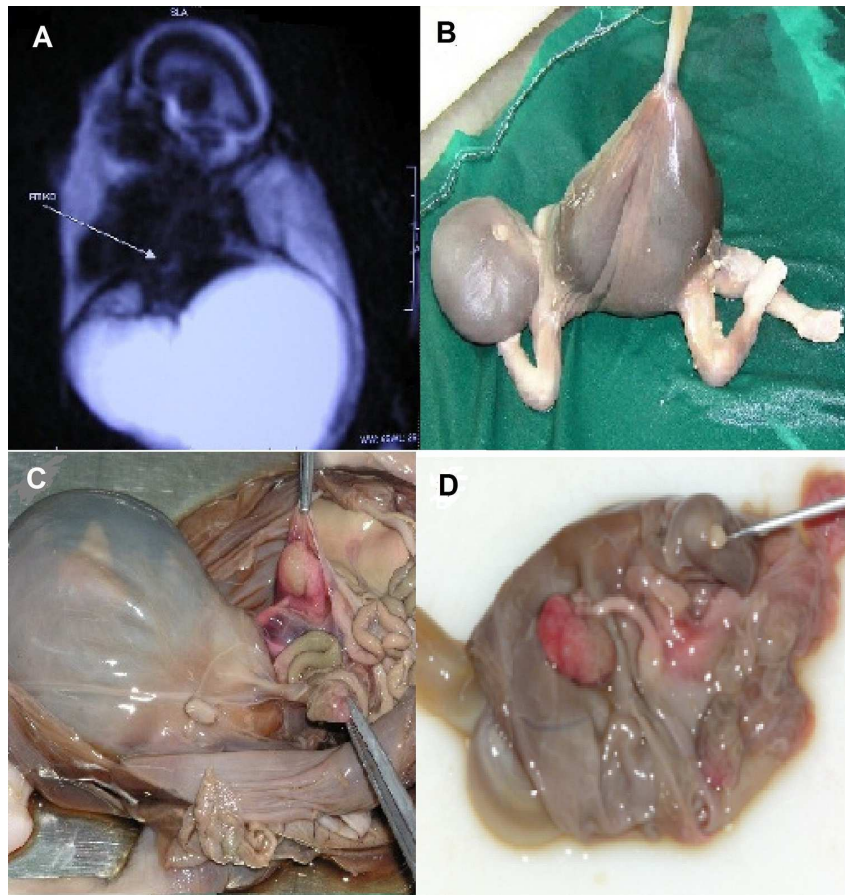
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CDS will occur (Achiron et al. 2000). This defect is characterized by the absence of anal, urethral and vaginal openings with flat perineal surface and a phallus-like structure which is usually accompanied with numerous anomalies such as malformed kidneys, urinary tract obstruction, oligohydramnios, urethral atresia or agenesis, pulmonary hypoplasia, renal agenesis and bladder aplasia (Pauli 1994). It can also induce different anomalies such as pulmonary hypoplasia, thoracic compression, enterolithiasis, vertebral, cardiac and gastrointestinal defects. Prenatal recognition of CDS is usually difficult because of the fact that some anomalies may not be significantly recognized by ultrasonography. Bladder diverticula, hydronephrosis, tubular ectasia, ureterocele, renal cystic disease, multicystic dysplastic kidneys (MCDK), atresia of the ureter and other urogenital complications can be verified by the utilization of FMRU (Cerwinka & Damien 2008). This case presentation shows a prenatally diagnosed fetus with CDS confirmed with detailed FMRU.

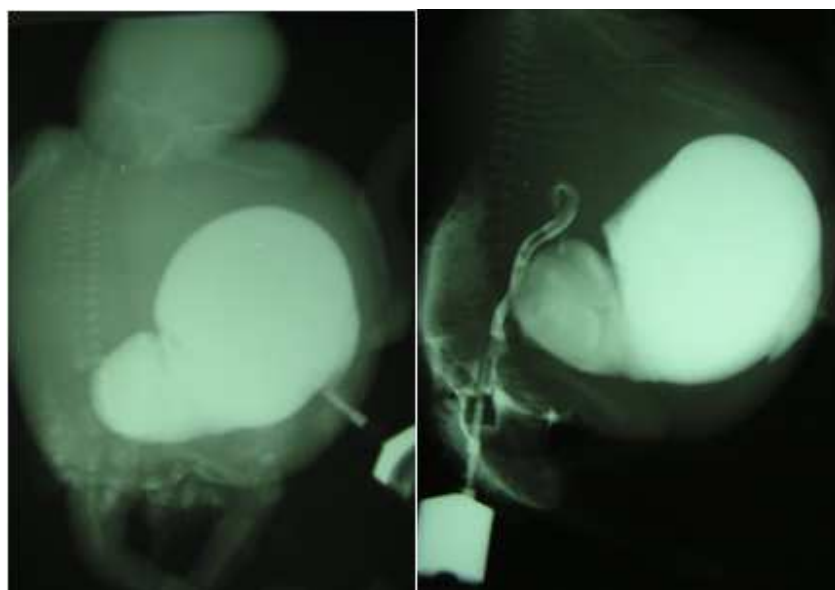
### **Case Presentation**

A fetus with ambiguous external genitalia was detected in a 24-year-old woman with FMRU investigation at 18 weeks of gestation.

Ultrasound at 16 weeks of gestation revealed a bladder solid mass without any concomitant anomaly, However, FMRU examination specifically detected anomalies including, bilateral hypoplastic kidneys, right cystic kidney, markedly enlarged urinary bladder accompanied with dilatation of upper urinary tracts and no urethra. The imaging findings included evidence of lower urinary tract obstruction without a fistula between the GI and GU tracts and admixing of urine and meconium. FMRU signals in the distal bowel were altered due to the meconium/urine mixing. In fact, a large bladder cystic mass had filled the abdominal cavity of the fetus, producing pressure effect to diaphragm. In addition, moderate oligohydramnios was noted. FMRU results made diagnosis of CDS most likely. After the examination of fetus by three perinatologists, it was decided to terminate the gestation after obtaining parental consent. All the imaging findings in combination with ambiguous genitalia were the clue that our case was afflicted by CDS. In post termination autopsy dysplastic left kidney and complete bladder neck obstruction due to urethral atresia were observed (Figure 1). Furthermore, post termination cystogram and colon imaging was performed that confirmed urethral atresia (Figure 2).



**Figure 1: A. Transverse FMRU of the Fetus Demonstrating Right Kidney with Transverse of Dilated Fetal Bladder and Urethral Agenesis B-D. Post Termination Autopsy of the Fetus with Absence of Urethral, Vaginal and Anal Openings, Depicting Right and Dysplastic Left Kidneys with a Total Bladder Neck Obstruction and a Striking Dilated Bladder**



**Figure 2: Post Termination Cystogram and Colon Imaging Confirmed Urethral Atresia**

### Discussion

The etiology of CDS is not exactly clear. Both genetic and environmental factors can induce this complex malformation, but there is no definitive gene identified in producing this combination of multiple developmental abnormalities (Liang, Ioffe & Sun 1998). The consumption of doxylamine succinate and etretinate (a long-acting retinoid in mice); as environmental agents, have been suggested for its occurrence (Liang, Ioffe & Sun 1998). In the past, it was considered that CDS occurred only in females; but it has been recognized that male fetuses can also be affected by this abnormality (Pauli 1994). A major characteristic of this malformation concludes absence of a median perineal raphe (Liang, Ioffe & Sun 1998). Proximal urethral dilation, oligohydramnios, enormously enlarged bladder, bilateral hydronephrosis, megacystitis, bilateral MCDK, hydroureter, various degrees of cystic dysplasia of kidneys and reduction of amniotic fluid index are the consequences of the urinary tract obstruction (Sahinoglu et al. 2004). As the time of urinary obstruction prolongs, the severity of kidney disease and pulmonary hypoplasia increases and the volume of amniotic fluid reduces, that

can notably influence the neonatal survival (Sahinoglu et al. 2004). Pulmonary hypoplasia is the most frequent complication of CDS which may occur if oligohydramnios develops before 24 weeks of gestation as the result of urinary tract obstruction. In order to facilitate the prenatal counseling, the affected organs should be examined precisely by the application of more sensitive diagnostic tools. In some situations, termination of gestation is recommended; but the survival of fetuses in neonatal period and the management of these cases should also be considered. Delivery of such cases may be complicated and should be performed at a center with available perinatal, neonatal and surgical services.

One of the elements determining survival of a fetus with cloacal anomalies is the degree of renal development (Liang, Ioffe & Sun 1998). There has been a striking development in diagnostic protocol of FMRU within the last seven years (Vegar-Zubovic, Kristic & Lincender 2011). Urological pathologies can be noticeably detected and differentiated by the application of FMRU (Vegar-Zubovic, Kristic & Lincender 2011), which is a non invasive and feasible method in diagnosis of dilated urinary system, in both neonates and

fetuses. Even in cases with obvious communication between genitourinary and gastrointestinal tracts; in which the diagnosis by ultrasonography is challenging, FMRU can be used as a practical method with shorter scanning time. Most of the urological abnormalities such as ureteropelvic junction (UPJ) stenosis, posterior urethral valve (PUV) or mega ureter can be sensitively detected by the application of FMRU. This meticulous diagnostic tool can play a crucial role in fetuses faced with complex anomalies. In addition, some of the coexisting anomalies can't be detected in ultrasonography examination because of late referral (Sahinoglu et al. 2004). For these reasons, FMRU is regarded as a valuable tool in specification of urinary tract disorders. Although most of malformations are diagnosed in the last trimester of pregnancy (Gupta et al. 2010), the critical period for diagnosing the abnormalities and evaluating the possible need for termination of pregnancy is before 24 weeks of gestation.

In conclusion, we prenatally diagnosed almost all the specific details of CDS by the application of FMRU. Since this congenital malformation is accompanied with several complications that will result in neonatal death in most of the cases, its accurate prenatal diagnosis, its differentiation from other conditions with better prognosis can be crucial to the families. FMRU can be a finer diagnostic tool to characterize the defects identified by ultrasound in pregnant women confronted with possible CDS fetuses. However, more awareness of this technique would be of benefit in confirming cases with prenatal assumed diagnosis of CDS.

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