Cloacal Dysgenesis Sequence: A Second Trimester Loss

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INTRODUCTION

Cloacal Dysgenesis Sequence (CDS) is rare and seen in only 1:50,000 to 250,000 pregnancies[1]. This uncommon malformation is often not complatible with long term survival and results in early oligohydramnios resulting in lethal pulmonary hypoplasia. Cloacal Dysgenesis Sequence commonly presents postnatally with a flat perineum, absent urethral, vaginal, and anal openings, renal anomalies and ambigious genetalia.

METHODS

Sequential fetal ultrasound monitoring from 13 weeks gestation with fetal postmortem following in-utero Demise at 20+3 weeks gestation.

CASE PRESENTATION

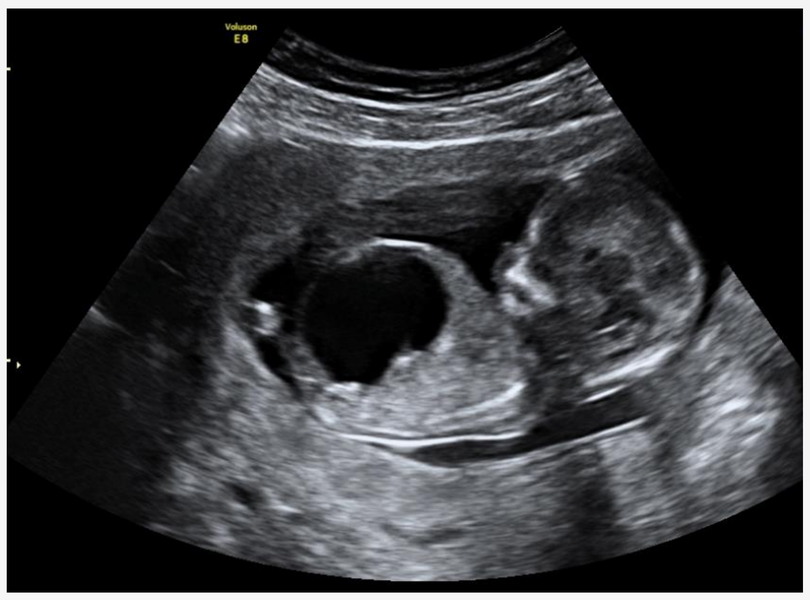
We present the case of a 33-year-old para 0+3 with three previous unexplained first trimester losses. An early anatomy scan at 13+5 weeks gestation showed megacystis with exomphalos and normal nuchal translucency. Chorionic villous sampling confirmed a normal karyotype 46, XY. At 16 weeks gestation oligohydramnious was noted with normal growth velocity, megacystis with bilateral megaureter and talipes. At 20+5 weeks gestation in utero fetal demise was diagnosed. A subsequent postmortem carried out confirmed a diagnosis of a Cloacal Dysgenesis Sequence with exomplalos.

DISCUSSION

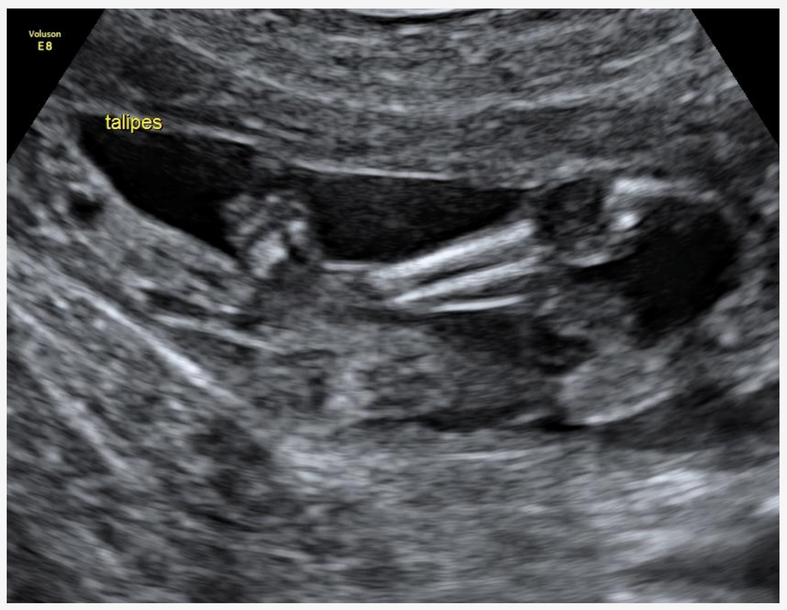
Cloacal Dysgenesis Sequence developes during the seventh week and results in a single perineal opening with communciation between the genital, urinary and gastrointestianl structures. This congenital malformation is usually seen in associataion with other malformations such as vertebreal, genitourinary and pulmonary malformations. Though mainly lethal, there have been few reported cases of survival, all of which however had a fetal urinary outlet, thus preventing oligohydramnios and pulmonary hypoplasia[1]. Diagnosis of CDS can be made antenatally by transabdominal ultrasound, as in the case described above. Oligohydramnios with associated megacystis, hydronephrosis and a perineal mass are characteristic ultrasound findings.
Karyotyping usually reveals normal female karyotype[2].
It is important to distinguish CDS from malformations such as fetal obstructuve uropathies which could potentially benefit from intrauterine surgical intervention and are associated with a better overall prognosis.

CONCLUSIONS

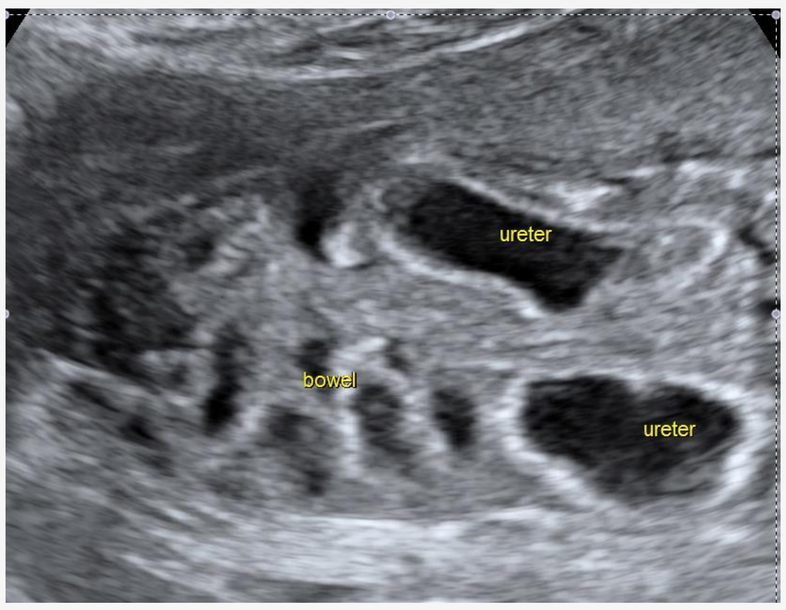
Cloacal Dysgenesis Sequence is a rare malformation with only an estimated 80 cases described in the literature. Survival after one year of age is extremely rare. Prenatal diagnosis is difficult, but Cloacal Dysgenesis Sequence should be considered in the differential diagnosis of megacystis in the first trimester.



13 weeks, Megacystis



16 weeks, Talipes



18 weeks, Bilateral Megaureter

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2. Chen CP, Chern SR, Lee CC, Town DD. Isochromosome 18q in a fetus with congenital megacystis, intra-uterine growth retardation and cloacal dysgenesis sequence. Prenat Diagn 1998;18(10):1068 - 74.