Cloacal Dysgenesis Sequence (CDS) is rare and seen in only 1:50,000 to 250,000 pregnancies. This uncommon malformation is often not compatible 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 ambiguous genitalia.

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 oligohydramnios 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 exomphalos.

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.

REFERENCES