A case of caudal regression syndrome

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Objective
Our aim was to describe the prenatal findings of a fetus with the diagnosis of caudal regression syndrome.

Methods
We performed the description of the case, along with a review of the literature.

Results
A 22-year-old woman was referred to a reference fetal medicine hospital as result of a diagnosis of gestational syphilis and evidence of severe oligohydramnios by an ultrasound (US) evaluation at 17 weeks. It was her first gestation; the husband was 21 years old, healthy and non-consanguineous. There was no history of malformations or genetic disorders in the family. A 22-week US revealed a cystic image at the lumbosacral region measuring 1.3 x 0.9 cm (suggestive of myelomeningocele), dextrocardia with pericardial effusion, left-sided aortic arch and undifferentiated genitalia. The stomach, kidneys and bladder were not visualized. Fetal karyotype was normal (46, XY).

More recent US at 27 weeks confirmed the previous findings and was indicative of bilateral renal agenesis. Fetal development was in the 25th percentile for gestational age. Fetal magnetic resonance imaging (MRI) performed at 29 weeks confirmed the severe oligohydramnios and showed severe pulmonary hypoplasia, dextrocardia, pericardial effusion and bilateral renal agenesis. Genitalia couldn’t be identified. The MRI findings were suggestive of myelomeningocele measuring 2 cm in its biggest axis associated with an elongated 3.5 cm x 0.8 cm cystic collection. The vertebral column was tortuous, though not properly visualized below the level of the midline lesion. Additional findings were suggestive of bilateral lower limb hypoplasia. The couple decided to terminate the pregnancy. The fetus was stillborn at 33 weeks of gestation, weighing 1,475 g. Postnatal evaluation revealed a flattened face (Potter’s facies); flat nasal bridge; marked infraorbital rim; micrognathia; small, low-set and posteriorly-rotated ears with marked overlapping of the superior helixes; cloacal extrophy; ambiguous genitalia; absence of the anus and deformities of lower limbs, including club foot. The previously suspected myelomeningocele was in fact a terminal myelocystocele. Radiographic evaluation revealed several axial skeleton abnormalities, including hemivertebrae, butterfly vertebrae, fused ribs, bifid ribs and total sacral agenesis.

Conclusion
Caudal regression syndrome should be considered in cases of bilateral renal agenesis, especially when additional findings are present. The severe oligohydramnios secondary to the bilateral renal agenesis may hinder the evaluation through ultrasound. In these cases, MRI can serve as a complementary exam, assisting in the evaluation and diagnosis.