The first Brazilian case of Hydrolethalus syndrome
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Objective
To report a fetus with findings consistent with Hydrolethalus syndrome, which were confirmed postnatally. To review the literature.

Methods
Case report.

Results
The child is the first daughter of young, healthy and nonconsanguineous parents. Ultrasound evaluation, at second trimester, identified Dandy-Walker malformation, agenesis of the corpus callosum, microphthalmia, reduced thoracic diameter and length of limbs, cystic retrovesical lesion and polyhydramnios. Fetal echocardiography was normal. After confirmation of these findings through fetal magnetic resonance imaging (MRI), Hydrolethalus syndrome was suspected. The child was born by cesarean section at 35 weeks gestation: weight 2649g, length: 46cm head circumference: 33 cm, APGAR 3/5, ventilated. Physical examination identified a median cleft lip and palate, micrognathia, hypoplastic and malformed tongue, dysplastic and low-set ears, short neck with redundant skin, narrow chest, postaxial polydactyly of hands, contracture of wrists and fingers, hypoplasia of labia majora, anterior placed anus and broad/bifid hallux. The evaluation through GTG-Banding karyotype was normal (46, XX). Brain ultrasound confirmed the prenatal findings and identified dilation of the cerebral ventricles. Abdominal/pelvic ultrasound showed pielocalicinal dilatation and didelphic uterus with hydrometrocolpos. Whole body radiography showed narrow thorax with short ribs, underdeveloped vertebrae and shortening of the humeri. The child died due to acute respiratory failure at 25 days of life.

Conclusion
The clinical findings presented by the patient were consistent with the diagnosis of Hydrolethalus syndrome, a rare and lethal autosomal recessive genetic disease. We did not find cases in Brazilian literature. Most of the reported cases were from Finland, where the incidence is 1: 20,000 births. Our findings also stand out the importance of the multidisciplinary team in the evaluation of prenatal findings. This, by itself was able to provide important information for proper genetic counseling and adequate pediatric care for the neonate.