Diaphragmatic hernia in a fetus presenting partial trisomy of chromosome 8 and partial monosomy of chromosome 15 secondary to a maternal translocation

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Objective
To report the diagnosis of congenital diaphragmatic hernia (CDH) using ultrasound and magnetic resonance imaging (MRI) in a fetus presenting partial trisomy of chromosome 8 and partial monosomy of chromosome 15 secondary to a translocation of maternal origin.

Methods
Case report.

Results
We report the case of a 34 years-old woman (G2P1). She was initially referred at 28 weeks of gestation with the diagnosis of ventriculomegaly and left diaphragmatic hernia. She had no first-trimester screening nor morphologic mid-trimester ultrasound. The examination performed at our institution confirmed these findings. The impression was that the stomach was within the chest. Moreover, a suspicion of an agenesis of the corpus callosum was raised. Fetal MRI was consistent with the sonographic findings and confirmed the agenesis of the corpus callosum. Fetal echocardiography showed no cardiac defects but the mediastinal shift. Fetal GTG-Banding karyotype through amniocentesis showed the presence of a structural abnormality in one of the chromosomes 15. The child was born at 37 weeks, weighing 2,675g and required immediate intubation. He did not present any external abnormality or dysmorphism. After progressing to hypoxemia, shock and severe pulmonary hypertension the neonate at one day of life. Chromosomal evaluation of the mother showed that she was a carrier of a balanced translocation between chromosomes 8 and 15 [46, XX, t(8;15)(q11.21;q13)]. This result allowed us to define the chromosomal alteration presented by the child: partial trisomy of chromosome 8 and partial monosomy of chromosome 15 secondary to a translocation of maternal origin [46, XY, +der(8)t(8;15)(q11.21;q13), -15 mat].

Conclusion
In our review of the literature, we did not find any cases of CDH associated with parental trisomies and/or monosomies of the chromosome segments involved in the structural alteration presented in our case. The identification of the balanced chromosomal abnormality in the mother was essential for future genetic counseling.