Polyhydramnios and genetic syndromes: predictive value of prenatal ultrasound

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Objective
To determine whether some specific prenatal ultrasound variables may discern among cases of polyhydramnios those that are associated to a genetic syndrome and/or a severe neuromuscular disease.

Methods
Retrospective study from 2005-2014 involving singleton pregnancies with polyhydramnios defined by a deepest vertical pocket (DVP) of amniotic fluid ≥ 8 cm after 20 weeks of gestation. The severity of polyhydramnios was stratified according to the DVP of amniotic fluid in mild (8.0 – 10.0 cm), moderate (10.1 and 12.9 cm) and severe (≥ 13.0 cm). The biophysical profile was performed to assess fetal movements. All neonates were followed-up for at least three months after birth. The multivariate association between prenatal parameters and the presence of genetic syndromes or neuromuscular diseases diagnosed postnatally was analyzed by binomial logistic regression.

Results
A total of 208 cases of polyhydramnios were identified during the study period. The mean gestational age at diagnosis was 30.1 (SD 3.4) weeks and the mean DVP of amniotic fluid was 9.3 (SD 2.1) cm. Postnatal investigations revealed a genetic syndrome or a neuromuscular disease with severe neurological sequelae in 27 (13.4%) newborns. Among these, GDM was diagnosed in 2 (7.4%) cases, 12 (44.4%) had major structural anomalies diagnosed prenatally and the remaining 13 (48.1%) had idiopathic polyhydramnios. In the multivariate analysis, significant and independent contribution for the prediction of genetic syndrome/severe neuromuscular disease were provided by severe polyhydramnios with a DVP ≥ 13.0 cm (p=0.015, OR 3.5 [95%CI: 1.3–9.5]), and reduction of fetal movements (p<0.001, OR 17.1 [95%CI: 4–74.1]).

Conclusion
The severity of polyhydramnios and the reduction of active fetal movements are prenatal predictors of a genetic syndrome, independent of the presence or absence of major structural defects and GDM.