Prognostic significance of prenatal ultrasound in fetal arthrogryposis multiplex congenita (AMC)

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Objectives: Fetal arthrogryposis multiplex congenita (AMC) describes a heterogeneous disease entity characterized by multiple contractures affecting at least two different body areas. The aim of our study was to identify antenatal sonographic findings correlating with unfavorable prognosis.

Methods: This retrospective study included 41 pregnancies of suspected AMC diagnosed 1999-2017 at our tertiary referral center. Prenatal sonograms, pediatric charts and autopsy reports were analyzed.

Results: Pregnancy outcome data were verifiable in 34 out of 41 cases. Hydrops was present in 50% of postnatally deceased fetuses vs. 0% of the surviving 12 children. Absent stomach filling was found in 67% of the children with neonatal death. After subcategorization the limb-involvement-only-group, 8% showed hydrops vs. 100% in system anomaly group vs. 70% in neuromuscular dysfunction cohort (p=0.001).

Conclusions: In addition to disease-defining sonographic findings, those with prognostic significance could be identified. Hydrops, nuchal edema, scoliosis and absent stomach filling were associated with unfavorable outcomes implicating a neuromuscular etiology. This knowledge can help to predict the further course of the disease and support patient counseling.

[Graphs and images illustrating the findings]