

Cytogenetic analysis during fetoscopic laser surgery treatment of twin-to-twin-transfusion syndrome

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Objective

Fetoscopic laser surgery (FLS) for twin-twin transfusion syndrome (TTTS) offers prenatal cytogenetic diagnostic opportunities without increasing procedure related risk. We sought to evaluate the incidence of prenatal cytogenetic variations via karyotype and/or chromosomal microarray analysis (CMA) on amniotic fluid samples obtained at FLS in monochorionic twins.

Methods

All patients undergoing FLS at our institution are offered genetic counseling and karyotyping or CMA on amniotic fluid obtained intraoperatively. This is a retrospective study of all patients electing cytogenetic testing at FLS from 7/2014-12/2020. Results were classified as “normal/normal variant,” or “clinically significant,” and were examined with respect to patient demographics and ultrasound findings.

Results

Of 296 patients undergoing FLS, 177 (59.6%) elected cytogenetic analysis (138 karyotype only [78%], 28 CMA only [15.8%] and 11 both [6.2%]). Gestational age ranged from 19-26 weeks, most patients (n=149, 84.2%) were <35 years old, and 9.6% (n=17) had anomalies in at least one twin. Collectively, 8.4% (n=15) received cytogenetic results other than 46, XX or 46, XY. Seven karyotyped cases (4.7%) had pericentric inversions considered normal variants. Clinically relevant results were observed in 8/177 cases (4.5%), including 2/177 karyotypes (1.3%) with balanced translocations, 3/177 karyotypes (2.0%) with unexpected sex chromosome findings, and 3/39 CMAs with abnormalities not detectable by karyotype. Most maternal/fetal characteristics were unrelated to cytogenetic results.

Conclusion

We observed a higher than expected frequency of clinically relevant cytogenetic findings in TTTS cases. Given potential management impacts of cytogenetic results, we recommend offering all patients amniotic fluid karyotyping or CMA at FLS. Consequent follow-up, counseling, and perinatal care are services for which access needs to be anticipated and readily available, either at the intervention center or through the referring provider.