

Micrognathia and associated anomalies, aneuploidies and genetic syndromes

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Objective

To follow up the cases of antenatally detected micrognathia with respect to associated anomalies, aneuploidies and genetic syndromes and assess the completeness of investigations.

Methods

This is a retrospective audit conducted at our centre from January 2007 to December 2018 of all fetuses diagnosed to have micrognathia. The diagnosis was made in the first trimester by examination of the facial profile in the Nuchal Translucency (NT) measurement image where the tip of the mandible was noted to be behind the tip of the maxilla or the naso-palatine line. In the second and third trimesters, suspicion of micrognathia was confirmed by measuring Inferior Facial Angle (IFA) and Jaw Index (JI). Detailed ultrasound included an in-depth cardiac evaluation for all fetuses. All couples were offered invasive testing and genetic consultation. Sources of antenatal outcome data included examination of maternal medical records and prenatal genetic results, where available. Postnatal outcomes were obtained by telephone on 2 occasions, firstly to confirm birth details and secondly to get development history using a standard ASQ-3 questionnaire by telephone.

Results

72 cases of micrognathia were detected, 15 and 57 in the first trimester and the second/ third trimesters, respectively. 12/ 15 (80%) and 43/ 57 (75.4%) had associated structural and/ or growth abnormalities in the first trimester and the second/ third trimesters, respectively. 4 were lost to follow up. 51/ 68 (75%) had associated abnormalities, involving mainly the growth, skeletal, central nervous and cardiac systems. 37/ 51 (72.5%) had termination of the pregnancy, 6/51 (11.7%) had an intrauterine fetal demise and 7/51 (13.7%) had live births. 1 is an ongoing pregnancy. Follow up beyond 6 months was available in 2 babies, both of which have global developmental delay at 27 and 18 months. 17/ 68 (25%) fetuses had isolated micrognathia. 5/ 17 (29.4%) had termination of the pregnancy, 1 had an intrauterine fetal demise and 11/ 17 (64.7%) had live births. Follow up beyond 6 months was available in 7 babies. 1 baby has significant intellectual developmental delay at 4 years and 2 babies have minor speech delays at 18 months and 3 years. 3 babies are developing normally at 6, 9 and 24 months currently. 1 baby which had a cleft of the soft palate detected postnatally is developing normally at 7 years. 18 and 5 couples in the associated and isolated group respectively opted in favour of invasive testing. 9/18 (50%) had confirmed chromosomal or genetic anomaly of which 2 had trisomy 21, 2 had Trisomy 18, 1 had a Mosaic Turner Syndrome, 2 were recurrence of previous genetic syndromes and 1 was an array anomaly. In the isolated group, there were no chromosomal or genetic anomalies detected antenatally. However, the babies found to have developmental issues postnatally did not opt for antenatal testing.

Conclusion

Micrognathia can be reliably confirmed on antenatal ultrasound by careful examination of the facial profile. There is a high association with fetal structural and growth abnormalities and hence a detailed evaluation of the fetus must be performed. Genetic tests on the fetal sample must be guided by the presence or absence of associated anomalies and family history of any genetic abnormalities as aneuploidies, duplications/ deletions and single gene disorders may all be associated with micrognathia. Hence, micrognathia remains an ominous sign with significant postnatal morbidity.