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Case series of first trimester fetal micrognathia

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Objective

Fetal micrognathia is a rare occurrence in which there is abnormal or arrested development of the fetal mandible. Although fetal micrognathia can be isolated, it is usually a prominent feature in several chromosomal and genetic syndromes. Prenatal diagnosis of fetal micrognathia is usually subjective.

Methods

A retrospective study of cases of 1st trimester fetal micrognathia in the Fetal Medicine Unit of Chelsea & Westminster Hospital was performed. Cases between 2003 and 2022 were retrieved (ultrasound database ViewPoint 5 and 6) and reviewed with medical records. Micrognathia was confirmed by assessing the frontal naso-mental angle (FNM) and inferior facial angle (IFA). As previously validated, the cut-off of 142° for FNM angle and 50° for IFA were used.

Results

During the 19-year study period we diagnosed 18 cases of fetal micrognathia at 11-14 weeks of gestation. The mean FNM angle and IFA were 120.3° and 39.1°, the average gestational age was 13 weeks (range 11^{+5} to 13^{+6}). In 5 cases (28%) the fetal micrognathia was isolated and in 13 cases (72%) other anomalies were detected. In 15 cases, an invasive test was performed and in 7 cases there was a normal result (although 1 case of Cornelia de Lange syndrome and 1 with multiple abnormalities) and in the other 8 cases (53.3%) a genetic abnormality was detected (4 cases with Trisomy 18, 1 with Trisomy 13, 1 with Trisomy 22, 1 case with unbalanced deletion chromosome 7 and 1 case with unbalanced duplication chromosome 8p+/14q+). Three cases did not have invasive testing but 1 case had Stickler syndrome, 1 case had a termination of pregnancy due to multiple abnormalities (no genetic testing) and 1 case had a miscarriage (Trisomy 13), suggesting that over 70% of cases of fetal micrognathia are associated with a chromosomal or genetic condition. Eight couples continued the pregnancy, 4 of these had a chromosomal or genetic abnormality. In cases of isolated micrognathia only 1 case had a genetic abnormality.

Conclusion

Most of the case of 1st trimester fetal micrognathia are pathological and are associated with chromosomal and genetic abnormalities.