Micrognathia

Society for Maternal-Fetal Medicine; Beryl R. Benacerraf, MD; Bryann Bromley, MD; Angie C. Jelin, MD

Introduction

Micrognathia is a condition in which the mandible is undersized for the fetal face, giving the fetus the appearance of a small jaw and overbite on profile facial views.

Definition

Micrognathia and retrognathia both refer to an abnormal mandible. Micrognathia is an abnormally small mandible; retrognathia is a mandible that is displaced posteriorly (although not necessarily small) with respect to the maxilla. Most fetuses who are diagnosed with micrognathia by prenatal ultrasound imaging have a combination of these two disorders.[1] Agnathia (otocephaly) is complete or almost complete agenesis of the mandible, with the temporal bones rotating medially; this results in the ears being horizontal and adjacent to each other or even fused in the expected location of the mandible. This extreme form of micrognathia is very rare.[1]

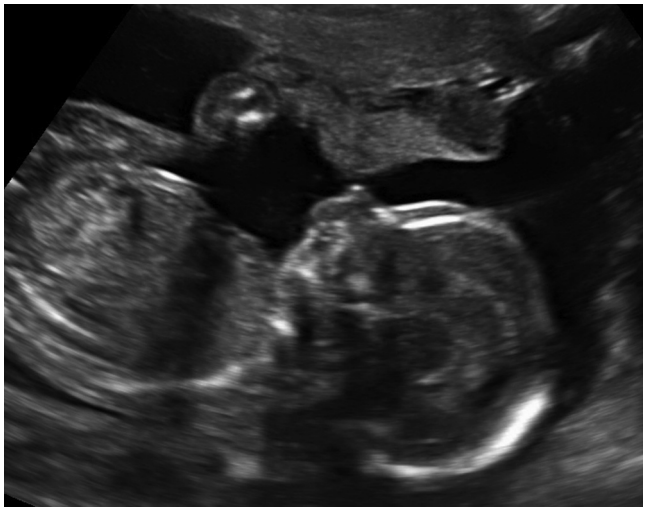
Ultrasound Findings

The midsagittal view of the face is necessary to evaluate the size of the mandible; care must be taken to avoid a flexed neck or chin-tuck fetal position that makes it difficult to see the size and position of the mandible. The chin and mandible are normally easily assessed subjectively and should be aligned with the upper lip and nose in the midsagittal view of the fetal face. The fetus with micrognathia or retrognathia will appear to have an overbite caused by the small and/or posteriorly displaced mandible. Measurement tables are available but are often unnecessary when the finding is clear on inspection of the fetal profile view.[2-4] The diagnosis of micrognathia can be suspected in the first trimester (12–14 weeks of gestation) on the midsagittal view of the fetal face. The sagittal profile must be observed carefully to ensure that the plane of section is truly midline. It is easy to create the impression of micrognathia if the view is not truly midline, although a normal profile cannot be visualized when micrognathia is present. The retronasal triangle view is also useful in the first-trimester evaluation of suspected micrognathia. There is normally a gap where the two mandibular rami come together on the coronal plane of the fetal face that is displayed in the retronasal triangle view. The absence of this mandibular gap is a useful early sign of micrognathia in the first and early second trimesters.[5] Overcalling this anomaly may lead to unnecessary testing, and micrognathia should not be diagnosed subjectively unless it is clearly seen (Figures 1 and 2).

Associated Abnormalities

Although mild micrognathia can be familial, it can also be associated with genetic disorders such as skeletal or neuromuscular diseases. Associated abnormalities include cleft palate and central nervous system (CNS), spine, limb, and hand anomalies. Micrognathia may appear to be isolated sonographically when associated with Pierre Robin

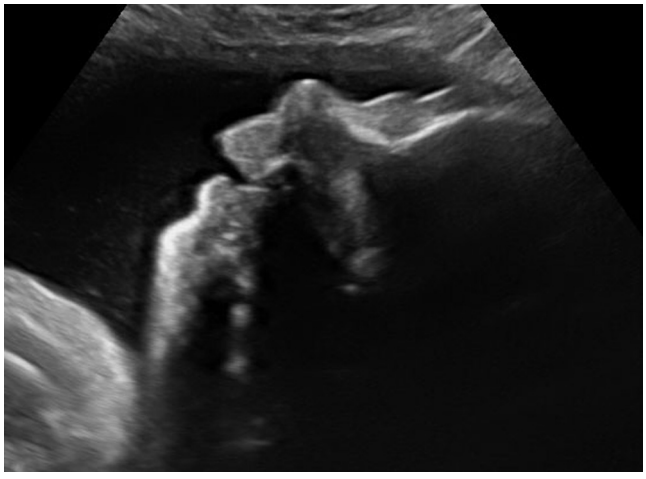
FIGURE 1
A fetus with Pierre Robin syndrome at 18 weeks of gestation



Note the small recessed chin and appearance of an overbite.

SMFM Fetal Anomalies Consult Series #1. Am J Obstet Gynecol 2019.

FIGURE 2
A third-trimester fetus with micrognathia



Note the recessed chin caused by the small size of the mandible and the posterior location of the tongue inside the mouth.

SMFM Fetal Anomalies Consult Series #1. Am J Obstet Gynecol 2019.

sequence, although there is a cleft palate in most cases. Cleft palate is difficult to detect sonographically in the absence of a cleft lip but can be seen in the third trimester with a three-dimensional and flipped-face view, where the palate is seen en-face looking cephalad from a plane inside the mouth.[6,7] It is also important to search for outer ear anomalies when micrognathia is suspected because this finding suggests Treacher Collins or Goldenhar syndrome. When micrognathia is part of a fetal syndrome, anomalies of additional organ systems (cardiac or lymphatic) can aide in the identification of a recognizable pattern of malformations.

Differential Diagnosis

It is important to distinguish between isolated and syndromic micrognathia. Primary mandibular syndromes include Pierre Robin sequence (micrognathia leading to glossoptosis, which affects palate formation and results in a cleft palate; this is sometimes diagnosable in the third trimester), Nager syndrome (acrofacial dysostosis with associated abnormal radii), Treacher Collins syndrome (ear anomalies), and orofacial digital syndromes (CNS and hand anomalies). Skeletal and neuromuscular diseases that result in micrognathia include multiple skeletal dysplasias (achondrogenesis, campomelic dysplasia, diastrophic dysplasia, and others), craniosynostosis syndromes (Shprintzen-Goldberg syndrome), and Roberts pseudothalidomide syndrome. Micrognathia is a feature of many chromosomal abnormalities, including mosaic trisomies 9, 13, and 18; triploidy, and others. These aneuploidies typically have many associated anomalies, such as cardiac defects, anterior abdominal wall defects, CNS anomalies, and limb malformations; detection of these can lead to the correct diagnosis. DiGeorge (22q11.2 microdeletion) syndrome is also associated with micrognathia. Other syndromes that can include micrognathia are too numerous to list but include Smith-Lemli-Opitz syndrome (genital and limb anomalies), Goldenhar syndrome (hemifacial microsomia with microphthalmia, ear tags, facial cleft, asymmetry, and hemivertebrae), Noonan syndrome (cystic hygroma, hydrops, heart defect), Meckel-Gruber syndrome (cystic kidneys, occipital encephalocele, polydactyly), Fryns syndrome (diaphragmatic hernia, heart defect), Pena-Shokeir syndrome (limb contractures, growth restriction), and Joubert syndrome (Dandy-Walker malformation).[1]

Genetic Evaluation

Diagnostic testing with chromosomal microarray analysis (CMA) should be offered when significant micrognathia is detected, particularly if there are additional features that are suggestive of a syndromic diagnosis. If a common aneuploidy syndrome is suspected, karyotype analysis or fluorescence in situ hybridization, with reflex to CMA, is a reasonable approach. Micrognathia can be inherited; however, a de novo variant is common with severe micrognathia. If there are additional anomalies, consanguinity, or a family history of a specific condition, gene panel testing or exome sequencing is sometimes useful because CMA does not detect single-gene (Mendelian) disorders. If exome sequencing is pursued, appropriate pretest and posttest genetic counseling by a provider who is experienced in the complexities of genomic sequencing is recommended.[8] After appropriate counseling, cell-free DNA screening is a reasonable option for patients who decline diagnostic evaluation if a common aneuploidy syndrome is suspected. If micrognathia is isolated, evaluation of both parents is indicated because mild micrognathia can be a constitutionally inherited variant.

Pregnancy and Delivery Management

In addition to a detailed ultrasound examination, careful evaluation of the fetal cardiac anatomy is important, and a fetal echocardiogram should be considered. Fetal magnetic resonance imaging may be useful if the palate is not clearly visualized or if there is concern for cerebral anomalies. The patient should be referred for pediatric consultation to neonatology, a craniofacial clinic, and other specialty services as appropriate based on sonographic findings. Consultation with an ear, nose, and throat specialist may be helpful if airway obstruction is suspected. The patient should be counseled about potential neonatal difficulties with breathing and feeding. Pregnancy termination is an option that should be discussed with all patients in whom a fetal anomaly is detected, although with isolated, mild micrognathia, the prognosis can be excellent. A third-trimester growth ultrasound examination with reevaluation of the mandible, fetal growth, and amniotic fluid index is recommended. No change in route of delivery is needed, although delivery at a tertiary care center is recommended with pediatrics and potentially ear, nose, and throat specialists present and ready to intubate if needed. A lactation consultation should be requested, and a breast pump should be prescribed if desired.

Prognosis

The prognosis depends on the final syndromic diagnosis. Isolated micrognathia is often associated with Pierre Robin sequence with glossoptosis, and airway obstruction should be anticipated at delivery. In some cases, the growth of the mandible can accelerate and normalize into adulthood. Prognostic counseling should be guided by the suspected diagnosis based on sonographic findings and diagnostic testing.

Summary

Prenatally detected micrognathia is often one finding of an associated syndrome. A detailed ultrasound examination with additional imaging should be performed to characterize the prenatal phenotype appropriately. Diagnostic testing is recommended and should be directed by the composite sonographic findings. Delivery at a tertiary care center with the capability for rapid neonatal intubation is preferable in the majority of cases. ■

REFERENCES

1. Paladini D. Fetal micrognathia: almost always an ominous finding. Ultrasound Obstet Gynecol 2010;35:377–84.

2. Paladini D, Morra T, Teodoro A, Lamberti A, Tremolaterra F, Martinelli P. Objective diagnosis of micrognathia in the fetus: the jaw index. Obstet Gynecol 1999;93:382–6.

3. Otto C, Platt LD. The fetal mandible measurement: an objective determination of fetal jaw size. Ultrasound Obstet Gynecol 1991;1:12–7.

4. Rotten D, Levaillant JM, Martinez H, Ducou le Pointe H, Vicaut E. The fetal mandible: a 2D and 3D sonographic approach to the diagnosis of retrognathia and micrognathia. Ultrasound Obstet Gynecol 2002;19:122–30.

5. Sepulveda W, Wong AE, Vinals F, Andreeva E, Adzehova N, Martinez-Ten P. Absent mandibular gap in the retronasal triangle view: a clue to the diagnosis of micrognathia in the first trimester. Ultrasound Obstet Gynecol 2012;39:152–6.

6. Platt LD, Devore GR, Pretorius DH. Improving cleft palate/cleft lip antenatal diagnosis by 3-dimensional sonography: the "flipped face" view. J Ultrasound Med 2006;25:1423–30.

7. Benacerraf BR, Sadow PM, Barnewolt CE, Estroff JA, Benson C. Cleft of the secondary palate without cleft lip diagnosed with three-dimensional ultrasound and magnetic resonance imaging in a fetus with Fryns' syndrome. Ultrasound Obstet Gynecol 2006;27:566–70.

8. International Society for Prenatal Diagnosis, Society for Maternal Fetal Medicine, Perinatal Quality Foundation. Joint Position Statement from the International Society for Prenatal Diagnosis (ISPD), the Society for Maternal Fetal Medicine (SMFM), and the Perinatal Quality Foundation (PQF) on the use of genome-wide sequencing for fetal diagnosis. Prenat Diagn 2018;38:6–9.