Abnormal Fetal Profile at First-trimester Ultrasound Scan Complicated by Severe Polyhydramnios at the Second Half of Pregnancy

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Section 1 – Quiz

Case Description

A 37-year-old pregnant woman, multigravida (G3P1), was referred to our outpatient clinic at 9 weeks of gestation. The first pregnancy was uneventful, and in the second pregnancy, the couple decided for medical termination of pregnancy at 16 weeks of gestation because of prenatal cytogenetic diagnosis of trisomy 21.

In the third pregnancy, the first-trimester ultrasound revealed a fetus with a crown-rump length of 63.4 mm (gestational age of 12 weeks and 6 days) and a nuchal translucency of 1.70 mm (below the 95th centile), normal Doppler of ductus venosus, no tricuspid regurgitation, and present nasal bones. There was, however, a suspicion for fetal micrognathia [Figures 1 and 2]. Remaining morphology of the fetus was considered normal at this stage. The placenta was anterior, the umbilical cord had three vessels, and the amniotic fluid volume was normal. First trimester combined screening had reduced risk for aneuploidies: trisomy 21 (1:6260) and trisomies 18 and 13 (1:100,000) with pregnancy-associated plasma protein-A = 1.26 MoM and free beta-human chorionic gonadotropin = 1.50 MoM. Performance of invasive testing was offered, but the couple refused.

At the mid-trimester ultrasound scan, performed at 20 weeks and 5 days of gestation, the fetus had cephalic presentation, normal amniotic fluid index, and a right anterolateral placenta, with no relation with internal cervical *os*. Biometric parameters were adequate for gestational age. The skull configuration and intracranial structures were normal. Fetal facial profile was abnormal, suggesting mandibular hypoplasia and glossoptosis [Figures 3 and 4]. Nasal bones measured 7.5 mm (above the 5th centile) and morphology of the upper lip was considered normal [Figure 5]. Stomach bubble was visible. There were no spinal, thoracic, cardiac, abdominal, genitourinary, or limb abnormalities, and the fetus had normal masculine genitalia. Taking into consideration the persistent suspicion of microretrognathia, prenatal counseling was made again, and an amniocentesis was proposed to the couple that refused it.

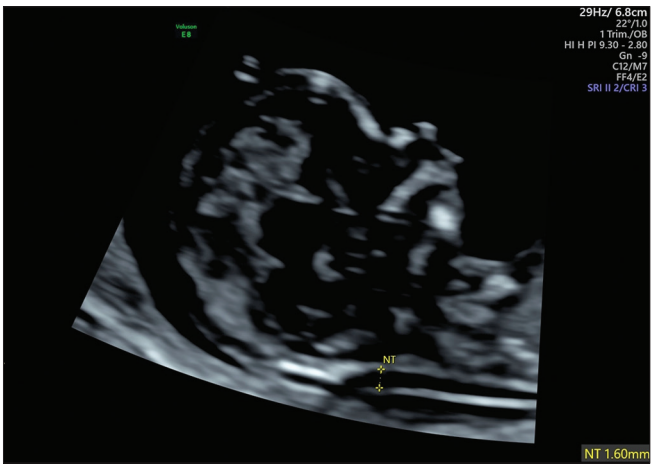

Figure 1: First-trimester ultrasound: Mid-sagittal section of fetal head raising suspicion for subtle retrognathia

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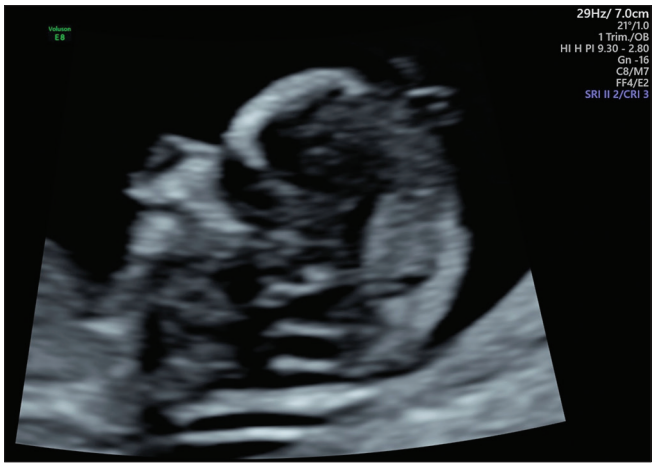

Figure 2: First-trimester ultrasound: Parasagittal section of fetal head with receding chin

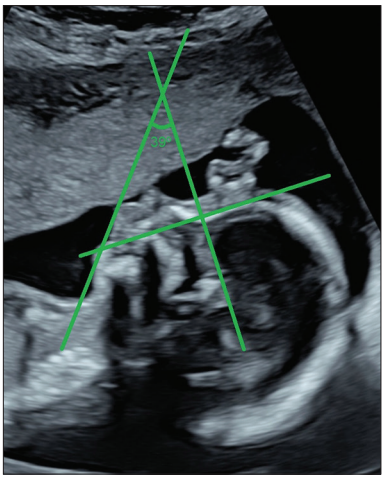

Figure 3: Mid-trimester ultrasound: Inferior facial angle of 39° suggesting micrognathia

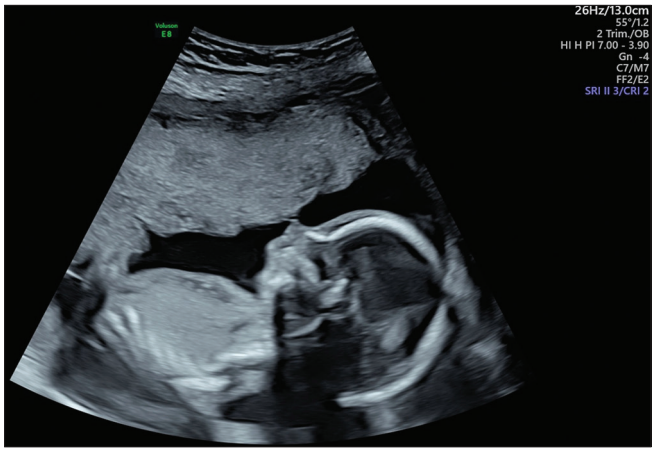

Figure 4: Mid-trimester ultrasound: Glossoptosis as the tongue is displaced posteriorly

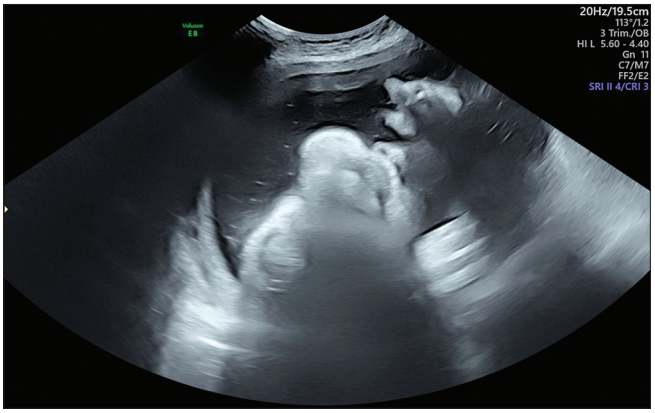

Figure 5: Mid-trimester ultrasound: Integrity of the upper lip excluding cleft lip

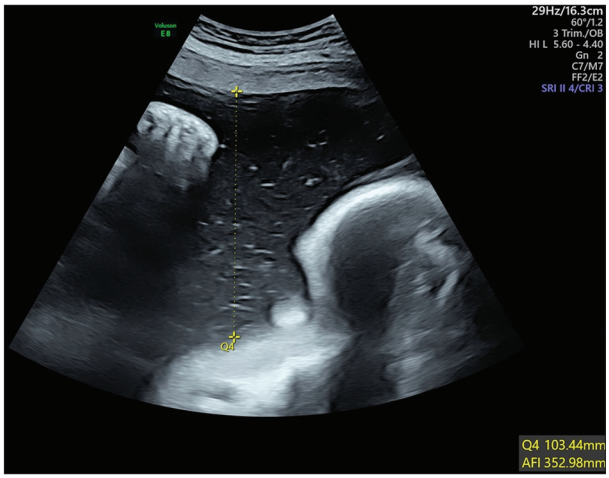

Figure 6: Third-trimester ultrasound: Polyhydramnios

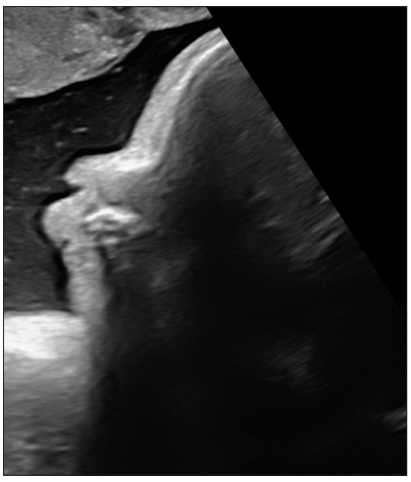

Figure 7: Third-trimester ultrasound: Mid-sagittal section showing abnormal receding position of the chin in relation to the upper lip

No other sonographic anomalies were identified until the third trimester. Screening for gestational diabetes was negative, and there was no evidence for maternal TORCH (Toxoplasmosis, Other [syphilis, varicella-zoster, parvovirus B19], Rubella, Cytomegalovirus, and Herpes infections) seroconversion or isoimmunization. Detailed fetal echocardiography was normal.

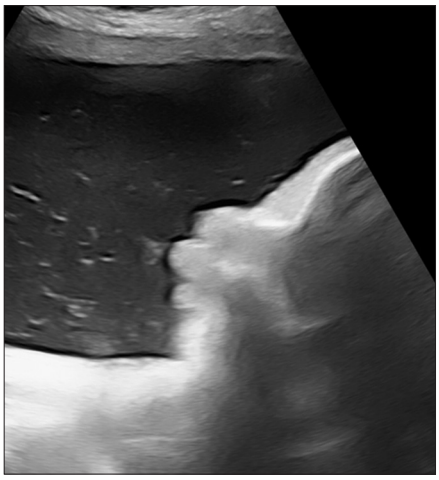

Figure 8: Third-trimester ultrasound: Mid-sagittal section showing prominent upper lip

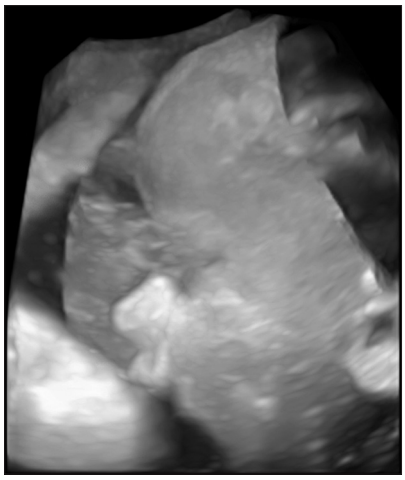

Figure 9: Third-trimester ultrasound: Three-dimensional reconstruction of the abnormal fetal profile highlighting prominent upper lip and receding chin

At 32 weeks and 5 days, ultrasound revealed a mild polyhydramnios with an amniotic fluid index of 24.5 cm and a deepest pocket of 8.9 cm. Estimated fetal weight was 2347 g, corresponding to the 82nd centile. Fetal Doppler and biophysical profile were normal.

Polyhydramnios was progressively increasing in severity. At 36 weeks, the fetus had an amniotic fluid index of 35.2 cm (deepest pool of 10.3 cm [Figure 6]) and an estimated fetal weight of 2942 g (61st centile). Fetal Doppler and biophysical profile were normal. Detailed review of fetal anatomy highlighted a prominent upper lip and receding chin in the mid-sagittal view of the face [Figures 7-9]. The patient was then hospitalized because of discomfort and dyspnea. A controlled induction of labor was initiated at 37 weeks. An emergent C-section was performed because of placental abruption after spontaneous membrane rupture. A male newborn weighing 2760 g and with Apgar index 5/7/8 was delivered and presented signs of airway obstruction. After birth, the newborn was evaluated by Pediatrics Surgery, Plastic Surgery, and Otorhinolaryngology. A complete soft palate cleft and glossoptosis were visualized in addition to the micrognathia prenatally diagnosed and no other morphologic anomalies were detected. The newborn karyotype and array comparative genomic hybridization were normal.

What Is the Diagnosis?

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.