American Journal of Obstetrics and Gynecology (2005) **193**, 1561–4

ELSEVIER

American Journal of Obstetrics & Gynecology

www.ajog.org

Ultrasonographic diagnosis of glossoptosis in fetuses with Pierre Robin sequence in early and mid pregnancy

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Received for publication February 4, 2005; revised March 10, 2005

KEY WORDS
Glossoptosis
Micrognathia
Pierre Robin sequence
Fetus
Ultrasound

Objective: This study was undertaken to describe the sonographic features of fetal glossoptosis in the Pierre Robin sequence.
Study design: Fetal sonography was prospectively performed in 8000 consecutive pregnancies at 14 to 24 weeks' gestation. In addition we retrospectively reevaluated ultrasound recordings of 4 fetuses from other hospitals, in which the diagnosis of Pierre Robin sequence was overlooked at 22 weeks' gestation. Glossoptosis was defined as a posteriorly displaced tongue that never reached the anterior mandibular alveolar ridge while watching the fetal profile. Micrognathia, which is a component of the sequence, was subjectively defined.
Results: Glossoptosis with micrognathia was detected in 2 fetuses in the prospective group at 14 and 15 weeks' gestation. Both pregnancies were terminated; the diagnosis was confirmed in 1 case where postmortem examination was performed. There were no false-negative diagnoses in the other 7.998 fetuses. Glossoptosis and micrognathia were observed in the 4 retrospective cases.
Conclusion: Sonographic identification of glossoptosis with fetal micrognathia suggests the possibility of Pierre Robin sequence.

The Pierre Robin sequence (PRS) was originally defined as micrognathia, glossoptosis, and cleft palate. PRS is pathogenetically and phenotypically variable. Not all investigators include both glossoptosis and cleft palate in addition to micrognathia in the definition of the disorder.[1-4] The reported prevalence of PRS is 1:2,000 to 1:30,000 and the mortality rate ranges between 2.2% and 30%.[1-4]

There are only few reports on the prenatal diagnosis of PRS.[5-9] In all these reports the diagnosis relied on the detection of micrognathia with or without cleft palate. However, because micrognathia may appear in 274 syndromes and malformation disorders,[10] it cannot be regarded as a single reliable marker of PRS. The detection of fetal glossoptosis might therefore be important for the definite diagnosis of PRS. However, there are currently no reports in the literature on the sonographic detection of fetal glossoptosis.

Antenatal diagnosis of fetal glossoptosis is of great clinical importance because posterior displacement of the tongue may cause acute neonatal respiratory distress

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doi:10.1016/j.ajog.2005.03.052

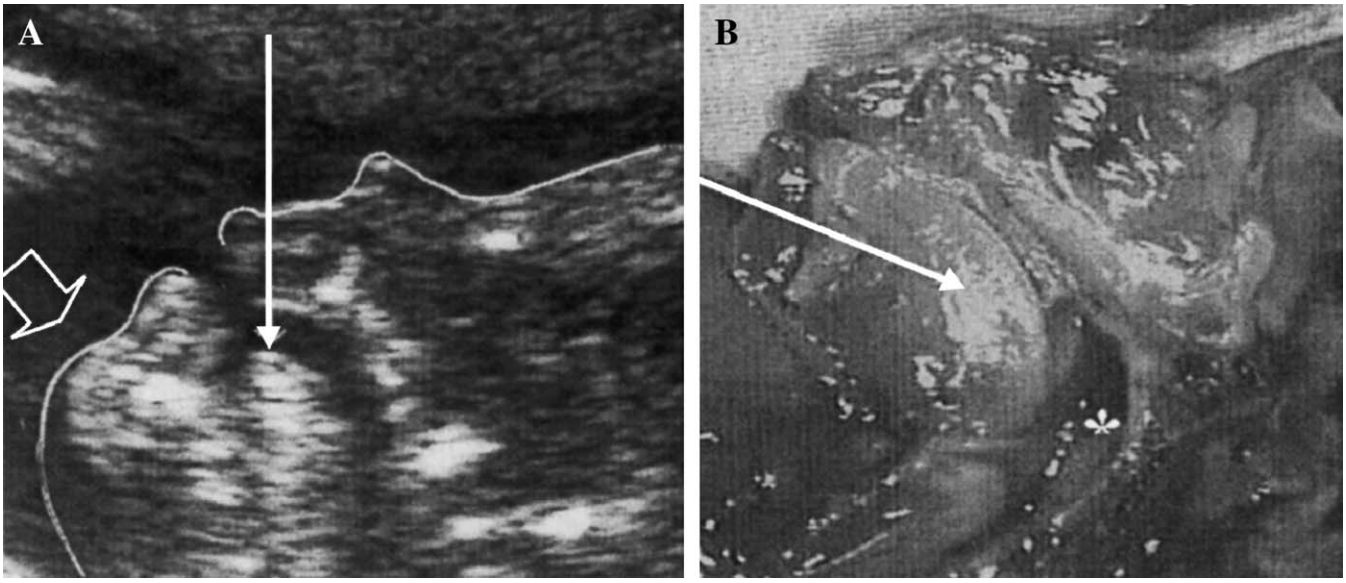

Figure 1 A normal profile of a fetus: **A,** US image of a fetus at 14 weeks' gestation. The tongue (*arrow*) reaches the anterior mandibular alveolar ridge. An *open arrowhead* marks the normal chin. **B,** Postmortem image of a fetus at 16 weeks' gestation. The normally located tongue is marked by an *arrow*. An *asterisk* marks the opened oropharynx.

and is a main reason for later breathing and feeding difficulties in these children.[1-4,11,12]

The current study describes the sonographic features of glossoptosis in fetuses with PRS.

Material and methods

There were 2 study groups: The first consisted of 8000 consecutive pregnancies, which were prospectively scanned by the same observer (M.B.). Of 8000 cases 6723 (84%) had a detailed endovaginal ultrasound (US) examination of all fetal organs at 14 to 16 weeks' gestation; and 1277 (16%) of 8000 cases had a transabdominal US examination at 20 to 24 weeks' gestation. These figures reflect the tendency of women in our practice area to have a detailed US examination in early pregnancy. All US examinations were performed with an ESI 3000 machine (Elscint Ltd, Haifa, Israel) that used a 7.5-MHz annular array vaginal transducer and 3.5 and 5 MHz abdominal probes. A postnatal physical examination was performed in all neonates of the prospective study group.

The second group consisted of 4 cases of PRS from other hospitals, in which the correct diagnosis was overlooked at 22 weeks' gestation. We retrospectively reevaluated the transabdominal US recordings of these fetuses.

Normally, while watching the fetal profile, the tongue can be observed as an echogenic structure, and its anterior tip can be imaged reaching the anterior mandibular alveolar ridge (Figure 1), or even protruding outside the mouth for short periods. Whenever posterior displacement of the tongue was suspected, a careful scanning of the fetal profile and tongue was performed. The diagnosis of glossoptosis was suggested in those cases in which the tongue was posteriorly displaced during most of the scanning session (20-30 minutes) and never reached the anterior mandibular alveolar ridge even during tongue movements (Figure 2).

Micrognathia was subjectively defined relying on the detection of an abnormal fetal profile with a small mandible. Special care was taken to exclude a possible false-positive diagnosis of micrognathia; this may occur during fetal mouthing movements when the maxilla sometimes tends to overlap the mandible, leading to an erroneous impression of a small mandible (Figure 3).

The study was approved by our Institutional Review Board, and all patients gave their consent.

Results

Fetal glossoptosis with micrognathia was observed in 2 of the 8000 pregnancies that were prospectively scanned. The anomalies were detected by endovaginal US in both fetuses. The tongue in these cases was posteriorly displaced during most of the scanning time, and only reached half the way to the anterior mandibular alveolar ridge during its movements. One fetus was detected at 14 weeks' gestation and the other at 15 weeks' gestation. Both fetuses had moderate micrognathia, normal karyotype, and no other structural anomalies.

Both these pregnancies were terminated on parental request. Postmortem examination was performed in 1 case and confirmed the diagnosis of glossoptosis and micrognathia (Figure 2). In the other case, the destruc-