Title:

Abnormal fetal movements in a preterm neonate with micrognathia and pulmonary hypoplasia: a case report

Running head:

Abnormal fetal movements
Abstract

Background:

Micrognathia is a facial malformation characterized by mandibular hypoplasia and a small, receding chin that fails to maintain the tongue in a forward position. We reported a system of prenatal screening that we developed to identify fetuses with compromised central nervous system function by observing fetal behavior.

We report the case of a preterm neonate with micrognathia and pulmonary hypoplasia who presented abnormal fetal movements.

Case presentation:

A 27-year-old Japanese primigravida at 33 weeks of gestation was referred to our hospital. Ultrasonographic examination revealed clinical polyhydramnios. Micrognathia was evident on midsagittal and 3D scan. The lung area was less than the mean -2.0 standard deviations for the gestational age. Pulsed Doppler sonography showed normal middle cerebral artery and umbilical artery pulsatility indices. We observed fetal movements for 90 minutes at 34 weeks 3 days of gestation. Movement in all four extremities was observed; however, no breathing or mouthing movements were detected and the fetus had sporadic eye movements. At 34 weeks 5 days of gestation, a cesarean section was performed for non-reassuring fetal status. The female infant had a
birth weight of 1675 g, with an umbilical artery pH of 7.385. Apgar scores were 5 at one
minute and 7 at five minutes. The neonate had mandibular hypoplasia and glossoptosis.
Severe respiratory compromise required immediate tracheostomy and cardiopulmonary
resuscitation with mechanical ventilatory support. However, the neonate’s
cardiopulmonary condition did not improve and she died 21 hours after birth.

Conclusions:
The findings of our ultrasound exam are suggestive of brain dysfunction. In our case, it
is likely that both the micrognathia and pulmonary hypoplasia are caused by
neurological disorder.
Background

Micrognathia is a facial malformation characterized by mandibular hypoplasia and a small, receding chin that fails to maintain the tongue in a forward position. When micrognathia is isolated, it is considered a component of Pierre–Robin syndrome (PRS) [1]. The underlying etiology of PRS has not yet been well established. Mandible growth results from oral motility, which begins during early fetal life [1, 2]. Moreover, the infants with PRS frequently have respiratory disorders related not only to anatomical conditions but to other causes as well. Abadie et al. has suggested a prenatal and neonatal brainstem dysfunctions as a “neuroembryological hypothesis” to explain the onset of some cases of PRS. Previously, we reported a system of prenatal screening that we developed to identify fetuses with compromised central nervous system function by observing fetal behavior [3]. We report the case of a preterm neonate with micrognathia and pulmonary hypoplasia who presented abnormal fetal movements.

Case presentation

A 27-year-old Japanese primigravida at 33 weeks of gestation was referred to our hospital with polyhydramnios and threatened preterm labor. Ultrasonographic examination revealed clinical polyhydramnios (amniotic fluid index: 28cm).
Micrognathia was evident on midsagittal and 3D scan (fig 1-a, b). The lung area of 8.9 cm² in the four-chamber view was less than the mean -2.0 standard deviations for the gestational age (normal: mean±2SD, 20.1±7.6). The biparietal diameter was 82 mm, femur length 51 mm and the estimated fetal weight was 1500 g, suggesting fetal growth restriction. Pulsed Doppler sonography showed normal middle cerebral artery and umbilical artery pulsatility indices. Amniocentesis was performed for a chromosome study, with a result of a 46,XX, karyotype. We observed fetal movements for 90 minutes at 34 weeks 3 days of gestation. Movement in all four extremities was observed; however, no breathing or mouthing movements were detected and the fetus had sporadic eye movements. At 34 weeks 5 days of gestation, a cesarean section was performed for non-reassuring fetal status. The female infant had a birth weight of 1675 g, with an umbilical artery pH of 7.385. Apgar scores were 5 at one minute and 7 at five minutes. The neonate had mandibular hypoplasia and glossoptosis and was diagnosed with PRS. Severe respiratory compromise required immediate tracheostomy and cardiopulmonary resuscitation with mechanical ventilatory support. However, the neonate’s cardiopulmonary condition did not improve and she died 21 hours after birth. At autopsy, the bilateral lungs contained little air, and the lung to body weight ratio was 0.01. Histologically, the epithelium of the pulmonary alveoli was thick and dysplastic as
well as reduced in number. These findings correlated with a lung maturity of 17-24 weeks of gestation.

Conclusions

In this report, we have described a case of micrognathia associated with pulmonary hypoplasia in which abnormal behavioral patterns, including sporadic eye movements were documented on a prenatal ultrasound exam. Normal alternations of eye movement and non-eye movement periods as well as breathing and mouthing movements were not evident. In animals, the neural center that generates the alternation rhythm of the eye movement and non-eye movement periods lies within the pons and/or medulla oblongata[3]. The absence of fetal breathing movements suggests a lesion involving the medulla oblongata, the breathing center. In this case, we have showed brainstem dysfunction prenatally. Abadie et al. proposed that dysfunction of the brainstem region controlling the rhythmic reflex of sucking and swallowing, cardiorespiratory, pharyngeal, and laryngeal functions may contribute to the severe feeding and respiratory disorders seen in infants with PRS. These functional anomalies involve several organs controlled by common neuronal networks located in the brainstem. Micrognathia results from a lack of mandibular movements and respiratory
movements are required for lung development[2, 4]. These observations have led to the idea that these impairments are due to a ‘neuroembryological pathogenesis’. The structural and functional abnormalities, obtained from our ultrasound examination are consistent with this idea. In our case, a postmortem brain examination was not performed. However, the findings of our ultrasound exam are suggestive of brain dysfunction. Thus it is likely that both the micrognathia and pulmonary hypoplasia are neurological, providing circumstantial evidence in support of the hypothesis of Abadie et al.

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References


Figure legends

Figure 1.

B-mode ultrasound scan of the fetal face at 33 weeks of gestation showing micrognathia (arrows: mandible)(a). Three dimensional ultrasound scan of the fetal face at 33 weeks of gestation showing micrognathia (arrows: mandible)(b). Newborn face showing micrognathia(c)