3 Newborn with Asymmetric Crying Face

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PRESENTATION

A 35-week-gestation male infant is delivered by cesarean section in the setting of maternal premature rupture of membrane and fetal nonreassuring heart tracing during the process of labor induction. The mother is a 30-year-old, gravida 2, para 1, healthy woman without significant medical history. Maternal antenatal testing results are normal with unremarkable prenatal complications. The neonate is the product of a nonconsanguineous marriage. At birth, the infant's head is delivered without instrumental assistance or birth trauma. He is vigorous and cries immediately after birth. Resuscitation efforts include a brief period of continuous positive airway pressure and oxygen support for increased work of breathing and desaturation; however, the infant recovers quickly and is transferred to the sick newborn unit in room air. His Apgar scores are 8 and 9 at 1 and 5 minutes, respectively. Physical examination reveals a nondistressed premature infant with normal vital parameters. His birthweight, length, and occipitofrontal circumference are 1,840 g, 39 cm, and 31.5 cm, respectively. He is found to have an asymmetric crying face. When he is quiet or sleeping, his face appears symmetric. However, when he cries, the left corner of the mouth is drawn to the left and downward while the right corner does not move. The forehead wrinkling, nasolabial fold depth, and eye closure remain intact and equal on both sides (Fig 1). All extraocular muscle movements are intact. He has an overfolded helix with squared superior portion of helix of bilateral ears and micro-retrognathia (Fig 2). Other physical examination findings are unremarkable. There is no history of a similar condition on both sides of the family.

DISCUSSION

Diagnosis

On day 3 after birth, systolic ejection heart murmur grade 3/6 is heard mostly at the left upper parastenal border on routine daily physical examination. Chest radiography demonstrates normal lung parenchyma and globular-shaped heart with absence of a thymic shadow. Echocardiography reveals a 2.2-mm ostium secundum atrial septal defect with left-to-right shunt, 4-mm subpulmonic ventricular septal defect with left-to-right shunt, and double outlet of right ventricle. Cranial and renal ultrasonography findings are normal. Chromosome study reveals 46,XY. Fluorescent in situ hybridization for 22q11 shows deletion. The neonate is diagnosed with congenital hypoplasia of depressor angularis oris muscle (CHDAOM) on the right side and 22q11.2 deletion syndrome. Further laboratory investigation

Figure 1. When the infant is at rest, the face appears symmetrical (left). When the infant is crying, the left corner of the mouth is drawn to the left and downward (right).

during hospitalization reveals a normal complete blood cell count, electrolytes, calcium, phosphate, and parathyroid hormone levels.

Condition

Asymmetric crying facies (ACF) is a condition in which an infant's face appears symmetric at rest and asymmetric when crying, that is, 1 corner of the mouth is pulled downward while another corner is not moving. (1) ACF can result from facial nerve compression to mandibular branch of facial nerve during the process of labor, leading to nerve injury. Another cause of ACF is hypoplasia or agenesis of the muscles that involve downward movement of lips, namely, depressor angularis oris muscle and depressor labii inferioris muscle, the latter being less common. ACF is a minor anomaly occurring in 3 to 8 per 1,000 live births and predominantly affects the left side more than the right side. (2) This condition needs to be differentiated from facial nerve paralysis, which can be congenital (due to genetic defects) or, more commonly, acquired (from obstetric-related nerve trauma). In ACF, facial asymmetry presents as 1-sided downward movement of the mouth and symmetric eye closure, whereas in true facial nerve paralysis, all the facial muscles on 1 side are affected.

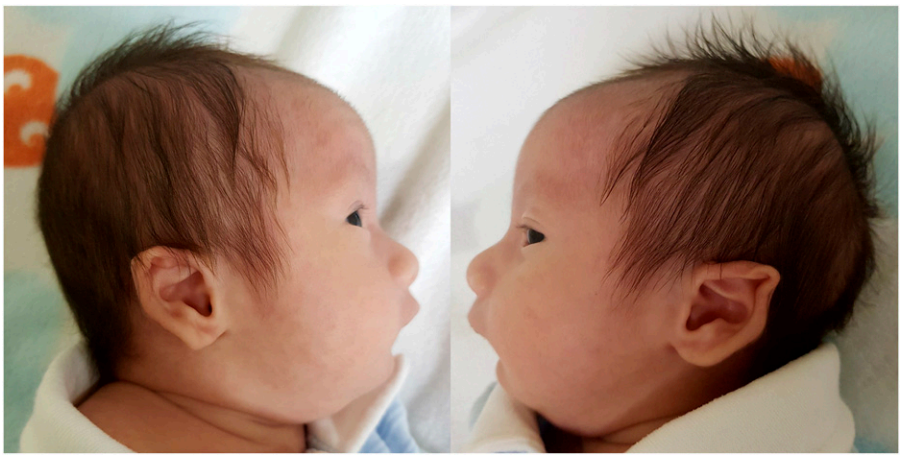

Figure 2. Infant facial features from right and left lateral views, showing overfolded helix with squared superior portion of helix of bilateral ears and microretrognathia.

The ACF that results from CHDAOM can be differentiated from the ACF caused by trauma using several approaches. First, based on the patient history, if obstetric risk factors for fetal compression are present (eg, large fetus, difficult labor, instrumental delivery), nerve compression can be suspected. Second, mandibular asymmetry and maxillary-mandibular asynclitism (nonparallelism of the gums) found during physical examination are important clues to the diagnosis of ACF resulting from nerve branch injury. Finally, further investigation using ultrasonography of facial muscles to ensure the absence or hypoplasia of depressor angularis oris muscle and electrodiagnostic testing to ensure paucity of motor unit potentials, lack of muscle fibrillation, and normal conduction velocity of the facial nerves may be useful to confirm a diagnosis of CHDAOM. (3)

The cause of the CHDAOM is not known; however, the pattern of inheritance does not suggest a single gene disorder, but favors complex multiple causative factors. (4)(5) Many other congenital anomalies may be present in infants with ACF that results from developmental error in muscle or nerve development. (6) The risk of associated anomalies in infants with ACF is 3.5-fold higher than in normal infants and it is reported that 5% to 70% of infants with ACF have associated anomalies, depending on the population and study criteria. (2) The common associated anomalies occur in the head and neck (48%), cardiovascular system (44%), musculoskeletal (22%), genitourinary tract (24%), central nervous system (10%), gastrointestinal tract (6%), and miscellaneous minor anomalies (8%). (4) Common head and neck anomalies are auricular malformation (dysplastic or hypoplastic ears, low-set ears, preauricular tags), mandibular or maxillary hypoplasia and palate anomaly. Common cardiac anomalies are atrial septal defect, ventricular septal defect, and patent ductus arteriosus. Other types of congenital heart diseases have also been reported. In other organ systems, various associated anomalies include those of the musculoskeletal system (syndactyly, clinodactyly, cortical thumb, hemivertebra), genitourinary system (inguinal hernia, renal hypoplasia, vesicoureteral reflux, cryptorchidism), central nervous system (auditory dysfunction, agenesis of the corpus callosum, hydrocephalus, brain cyst), and gastrointestinal system (esophageal atresia, imperforate anus, megacolon). (2)(4)(7)

The association of ACF and 22q11 deletion syndrome is well-described. Pasick et al report a 14% incidence of ACF in patients with a 22q11 deletion syndrome, which is significantly higher than in the general population. (6) In addition, various genetic syndromes, including trisomy 21, 4p deletion, Klinefelter syndrome, and VATER association (vertebral defects, imperforate anus, tracheoesophageal fistula, radial and renal dysplasia), are diagnosed in patients with ACF. (2)(5)(7)

Treatment

In the case of nerve compression, the prognosis is good and spontaneous resolution is expected. In contrast, in the case of CHDAOM, spontaneous resolution is less likely. (2) However, treatment of CHDAOM is usually not required because the asymmetry will become less obvious as other facial muscles compensate for the child's facial expression when the child grows up. In those who need treatment for cosmetic purposes or to correct functional deficit and improve quality of life, various therapeutic strategies are available. These include plastic-reconstructive procedures on the lower lip on the affected side or additional blocking/weakening of the contralateral nonaffected side. (8) Associated congenital malformation and genetic syndromes are managed accordingly.

Progression

The infant described here developed clinical congestive heart failure at 1 week of age. Treatment consisted of fluid restriction, diuretics (furosemide and spironolactone), and digitalis glycosides (digoxin), and resulted in significant clinical improvement. The patient passed the otoacoustic emissions hearing screening and was discharged from the hospital in stable condition at the age of 2 weeks. The parents were counseled about the prognosis of CHDAOM and 22q11.2 deletion syndrome. Parental genetic tests to detect chromosome 22q11 deletion were recommended to provide guidance for future pregnancies.

Lessons for the Clinician

- Asymmetric crying facies (ACF) is not uncommon and can be found in approximately 3 to 8 per 1,000 live births.
- Physicians should be able to differentiate between ACF and true facial nerve paralysis.
- Congenital hypoplasia of depressor angularis oris muscle can be associated with other congenital malformations and genetic syndrome. Therefore, a thorough physical examination and diagnostic evaluation are required.

American Board of Pediatrics Neonatal-Perinatal Content Specifications

- Know the significance of persistent neuromotor abnormalities in infancy (including asymmetries).
- Know the clinical and diagnostic features of the DiGeorge sequence (velocardiofacial syndrome, 22q11 deletion).

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Parent Resources from the AAP at HealthyChildren.org

- Micrognathia & Pierre Robin Sequence: https://www.healthychildren.org/English/health-issues/conditions/Cleft-Craniofacial/Pages/Micrognathia-Pierre-Robin-Sequence.aspx
- Infantile Spasms: What Parents Need to Know: https://www.healthychildren.org/English/health-issues/conditions/head-neck-nervous-system/Pages/Infantile-Spasms-What-Parents-Need-to-Know.aspx
- Children with Facial Asymmetry: https://www.healthychildren.org/English/health-issues/conditions/Cleft-Craniofacial/Pages/Children-with-Facial-Asymmetry.aspx
- Umbilical Cord Care: https://www.healthychildren.org/English/ages-stages/baby/bathing-skin-care/Pages/Umbilical-Cord-Care.aspx

For a comprehensive library of AAP parent handouts, please go to the *Pediatric Patient Education* site at http://patiented.aap.org.

Case 3: Newborn with Asymmetric Crying Face

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