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IMAGES IN CLINICAL PRACTICE

INFANT WITH ASYMMETRIC FACIES WHEN CRYING

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A seven-month-old male infant presented to the emergency department with mild respiratory symptoms. During the observation, asymmetry of the face with a downward deviation of the right labial commissure was noted when the patient was crying (Figure 1). At rest, the face was symmetric except for a slightly thinner and inward-turned lower left lip (Figure 2). According to the parents, these alterations have been present since the first days of life. The further neurologic assessment was normal. No other major or minor malformations were detected, and cardiac auscultation was normal without murmurs. He presented normal growth and neurodevelopment. There was no history of problems with sucking or drooling and no family history of congenital abnormalities, including facial asymmetry. The patient was a second-born child from nonconsanguineous parents. Pregnancy was uneventful and the patient was born at 38-3/7 weeks gestation by cesarean section because of prolonged labor, with a birth weight of 3270 g, birth length of 51 cm, and head circumference of 35 cm. Apgar scores at 1, 5, and 10 minutes were 7, 8, and 10, respectively.

Figure 1. Facial asymmetry observed when crying with the right side of the mouth deviated downward while the left side remained unmoved.

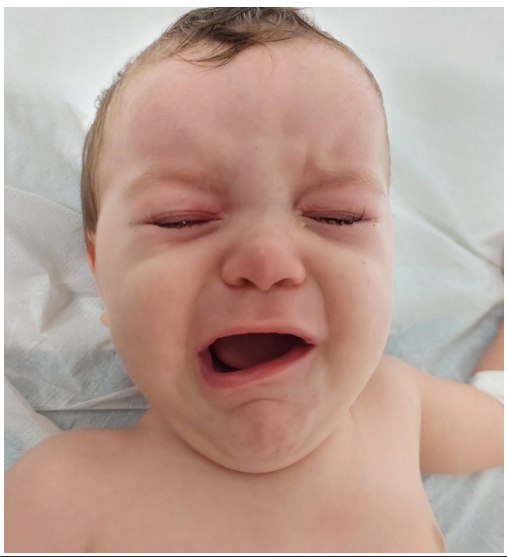

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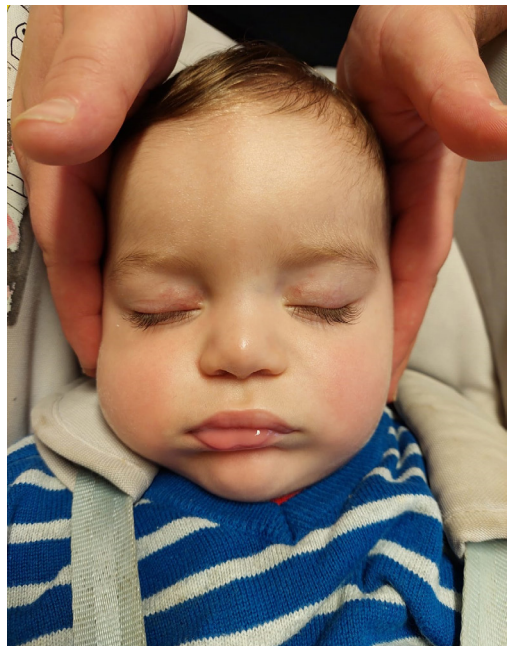
Figure 2. Symmetrical facial expression at rest with only the left lower lip slightly thinner and turned inwards.



What is the diagnosis?

Asymmetric crying facies (ACF) was diagnosed and assumed to derive from left depressor anguli oris muscle (DAOM) hypoplasia. ACF is defined by facial asymmetry observed during crying and is characterized by a downward deviation of the unaffected side of the mouth's angle, while the affected side remains unmoved. The facial expression is symmetric at rest, though the lip from the affected side can be slightly thinner and turned inward.[1] ACF has an estimated incidence of 0,31% to 0,82% of live births and is more prevalent in males (M:F ratio of 2:1).[1] The etiology of the asymmetric crying facies can be explained by two theories: the traumatic theory and the faulty development theory. In the first one, ACF is caused by the compression of one of the facial nerve branches in utero or during labor. In the second theory, the cause of the ACF is the hypoplasia or agenesis of the DAOM or the hypoplasia of the depressor labii inferioris muscle.[2] ACF due to nerve compression is typically an isolated abnormality. Potential risk factors include primiparity, multiple births, high birth weight, difficult labor or delivery, forceps delivery, and

uterine tumors.[1,2] Clues in physical examination for this mechanism are the presence of mandibular asymmetry and maxillary-mandibular nonparallelism.[1] On the other hand, the hypoplasia and agenesis of DAOM can be caused by intrauterine viral infections, genetic factors, or a brainstem-level defect.[3] Familial occurrence has been reported, with autosomal dominant inheritance with variable penetrance being suggested.[3] An association with other major and minor congenital malformations has been reported in up to 15% of patients with DAOM hypoplasia.[1] Such anomalies can involve every system, but the most commonly reported are cardiovascular and cervicofacial malformations.[2,3] A well-known association of ACF with congenital heart disease and 22q11 microdeletion has already been described.[2] ACF is a clinical diagnosis and can be differentiated from other entities as true facial paralysis based on the clinical history and physical examination. Typically, sucking, forehead wrinkling, eye closure, nasolabial fold depth, and tearing are preserved and symmetric in ACF.[1] Ultrasonographic and electromyographic testing of the facial muscles can be helpful diagnostic aids if a physical examination is not revealing.[2] Early determination of ACF cause is important since it affects therapeutic management and, thus, the prognosis.[1] In most cases, spontaneous resolution can be expected if the cause is traumatic and no further evaluation is necessary. However, if there is agenesis or hypoplasia of DAOM or DLIM, a careful physical examination is important to exclude associated malformations. And if these are present, performing an echocardiogram and eventually a FISH analysis for 22q11 microdeletion is crucial. If the physical examination is normal, observation only is appropriate.[1,2] As the child grows, risorius and other facial muscle use become more frequent. Smiling appears and dominates the child's facial expression, leading to a less noticeable facial asymmetry.[4] However, surgical intervention may be required to improve cosmetic outcomes.[1] In conclusion, in ACF, physical examination can be sufficient for its diagnosis, for the differential diagnosis with facial palsy, management, and parent reassurance. Despite being a benign condition in most of the cases, clinicians should not dismiss the possibility of association with other clinically significant malformations.[2]

Compliance with ethical standards
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