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CASE REPORT

Asymmetric Crying Facies with Multiple Congenital Malformations: a Case Report

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ASYMMETRIC crying facies (ACF) is characterized by facial asymmetry only when crying. It may present as an exclusive symptom, or be concomitant with other congenital malformations and even some fatal birth defects. Clinically, ACF is featured by drooping of the intact corner of the mouth during crying, while the face appears symmetric at rest. The facial nerve function is symmetric, as determined by frowning, forehead wrinkling, eye closure, and nasolabial fold depth. When ACF alone is present, the cause is often compression of the mandibular branch of the facial nerve that is probably due to birth trauma or abnormal fetal posture *in utero*. ACF with other congenital anomalies is usually caused by hypoplasia of the depressor anguli oris muscle (DAOM) on one side of the mouth. We described a 26-month-old girl with ACF and multiple congenital malformations in this report and reviewed the relevant literature regarding the occurrence, coexisting anomalies, probable etiologies, and surgical intervention. It is suggested that ACF can be an indicator of coexisting anomalies, and a comprehensive

examination should be conducted in these patients.

CASE DESCRIPTION

A 26-month-old girl came to the pediatrician's office with a fever and cough for 3 days. She was the first-born child of non-consanguineous ethnic Han Chinese parents. There was no family history of congenital defects.

The patient was born at 38-5/7 weeks after an uncomplicated pregnancy. Birth weight was 3100 g, and Apgar score was 9. At birth, it was noted that (i) the right corner of her mouth drooped when crying and (ii) she had a congenital anal atresia accompanied with a rectovaginal fistula. Perineal anoplasty and vaginal repair surgery were performed under general anesthesia as a result on the 15th day after birth.

Physical examination revealed normal closure of both eyes and contraction of both occipitofrontalis muscles. There was a marked depression of the right corner of the mouth during crying (Fig. 1) and significant dysplasia of the left auricle. Breath sounds were diminished in the left upper lung. A few moist rales have been audible over inferior lobes of bilateral lungs. Cardiac auscultation revealed a continuous murmur in the left second intercostal space.

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Chest computed tomography (CT) scans revealed left upper lung aplasia (Fig. 2A). Heart color Doppler ultrasound indicated a patent ductus arteriosus and absence of the left pulmonary artery. Congenital stenosis of the left principal bronchus was found *via* fiberoptic bronchoscopy (Fig. 2B).

The patient was diagnosed as pneumonia and ACF syndrome, accompanied by homolateral auricle dysplasia, left upper lung aplasia, absence of the left pulmonary artery, stenosis of the left principal bronchus, patent ductus arteriosus, congenital anal atresia, and rectovaginal fistula. She recovered from pneumonia after 5 days intravenous cefuroxime sodium and andrographolide. There was no more discomfort in the 6 months follow-up. The parents refused genetic testing.

DISCUSSION

According to 5 prospective studies published between 1973 and 2000, ACF occurs in 1 of 160 live births (0.6%).¹ Most ACF cases are secondary to compression of the mandibular branch of facial nerve, or due to a traumatic birth or abnormal fetal posture *in utero*, but not associated

with other malformations. Electromyography testing can show partial denervation of the depressor anguli oris, depressor labii inferioris, and mentalis muscles. These signs may help to confirm the diagnosis of ACF. More than 90% of ACF cases in this description can resolve spontaneously within 4 weeks after birth.

However, there are 9.4% of ACF newborns whose condition is associated with other significant congenital malformations.¹ For this minority of patients, congenital unilateral hypoplasia of DAOM (not the compression of mandibular branch) impairs the corner of the mouth, preventing it from drooping symmetrically to the contralateral side during crying. Coexisting anomalies can involve nearly all systems, but the cardiovascular system is the most commonly involved one. Systems that are less frequently affected are the genitourinary, musculoskeletal, central nervous, cervicofacial, respiratory, and endocrine systems. Some rare anomalies have also been described, such as psychomotor retardation,² hemihypertrophy,³ Glanzmann's thrombasthenia,⁴ and a unique combination of features consisting of bilateral absence of the fifth ray in the feet, solitary median maxillary central incisor, and a narrow apertura piriformis.⁵

The etiology of this syndrome has not been completely elucidated. In the present case, there were no remarkable obstetric perinatal factors or family history. However, some studies have associated it with chromosomal anomalies such as 22q11.2 deletion^{6,7} and 4p deletion.⁸ Besides, retrospective studies reported that a mother of ACF had diabetes mellitus for 6 years,⁸ and two ACF infants were born to mothers with gestational diabetes.⁹ In addition, there was a report of an infant with ACF accompanied with other congenital anomalies that was born to a mother who had ingested isotretinoin during the first month of pregnancy. It was hypothesized that ACF was potentially linked to intrauterine retinoic acid exposure, or embryopathy of the DAOM or the mandibular branch of the facial nerve.¹⁰

Unlike cases of ACF due to partial facial nerve compression, ACF caused by unilateral underdevelopment of the DAOM is less likely to resolve spontaneously, and surgical consultation is advisable. Traditionally, weakening the unaffected side through selective marginal mandibular neurectomy was performed. However, this resulted in lack of emotive movement in some cases. A simple reconstruction can be implemented with a bidirectional (horizontal and vertical) fascia graft to the affected DAOM. The horizontal fascial strip restores the center of the lower lip to its proper position, while the vertical fascial strip achieves symmetry of the lower lip at mouth opening. The ends of the vertical strip are anchored to the lower lip and the

Figure 1. A 26-month-old female with left-sided asymmetric crying facies.

- A. The face appeared symmetric at rest.
- B. The lower lip was pulled toward the intact right side during crying.

Figure 2. Left upper lung aplasia and congenital left principal bronchus stenosis.

- A. High-resolution chest CT scan showed atelectasis of the left upper lung.
- B. Fiberoptic bronchoscopy revealed stenosis of the left principal bronchus.

mandibular bone, respectively, thereby allowing simultaneous movement of the mandible and the lower lip. This method has been considered as a well-balanced surgical option based on its simplicity, minimal invasiveness, and the satisfaction of the patients.¹¹

In summary, herein we described a 26-month-old patient with ACF accompanied with several congenital anomalies in other systems. Unilateral hypoplasia of the DAOM was considered as the cause of her facial anomaly, although the genetic etiology was indeterminate for lack of a genetic test. We conclude that ACF can be an indicator of multiple congenital malformations, especially in cases that do not resolve spontaneously. A comprehensive examination is essential for early diagnosis, proper management, and a better outcome.

Conflict of Interest Statement

The authors have no conflict of interest to disclose.

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