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ASYMMETRIC CRYING FACIES: AN INDEX OF OTHER MALFORMATIONS*

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Congenital hypoplasia or absence of the depressor anguli oris muscle is a minor congenital anomaly causing asymmetrical crying facies (ACF). The interesting aspect of this abnormality lies in the frequently associated abnormalities. Cardiac, urogenital, musculoskeletal, respiratory and cervicofacial defects have been described in cases with ACF. Therefore it is suggested that ACF can be used as an index of other congenital malformations. We report three cases with ACF who had varied congenital anormalies. *Key words: asymmetric crying facies.*

An infant whose face appears symmetrical at rest yet whose face is pulled downward to one side when crying is said to have "asymmetrical crying facies (ACF)"[1]. The cause of the facial asymmetry in this disorder is congenital absence or hypoplasia of the depressor anguli oris muscle (DAOM) at the corner of the mouth[2-5].

A variety of anomalies have been observed in patients with ACF[1]. Therefore it is suggested that ACF can be used as an index of other congenital anomalies[1]. We report here three infants who were referred for evaluation of ACF and were investigated for other congenital anomalies.

Case Reports

Case 1

A 12-month-old female infant was referred to our clinic for evaluation of a striking asymmetry on the left corner of the mouth when crying, as well as intermittent cyanosis. She was the seventh baby of this mother following a full-term uncomplicated gestation, labor and delivery. She weighed 3400 g at birth, and no neonatal complications had been recognized. Her motor and mental development had been normal, and her parents and siblings were healthy.

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On physical examination, the pulse rate was 118/min. The patient had a normal appearance at rest and asymmetric facies when she was crying; the left corner of the mouth drew left and downward while the right moved very little (Fig. 1). There was no sign related to facial paralysis. She had mild perioral cyanosis and clubbing. On auscultation of the heart, a systolic ejection murmur and a third heart sound were heard, and the second sound was single and loud. The rest of the physical and neurological examination findings were normal.

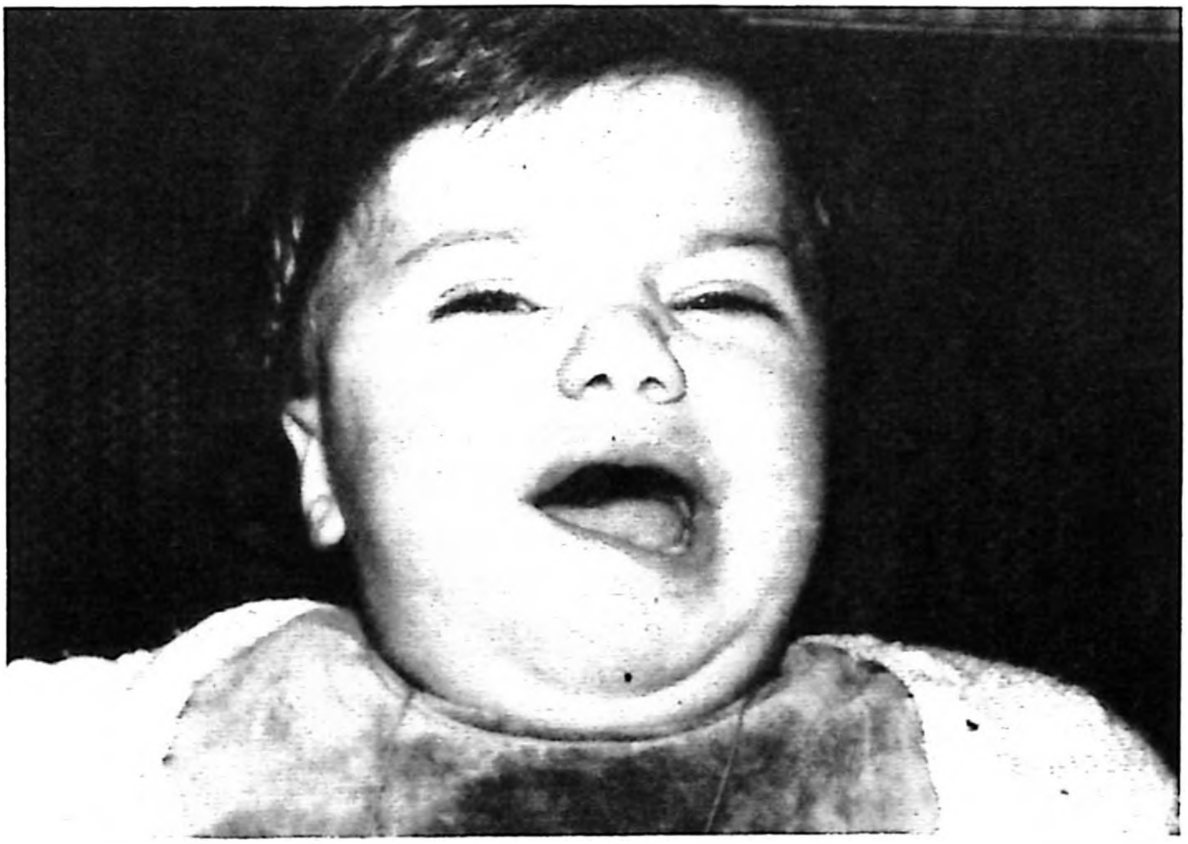

Fig. 1: Facial asymmetry of Case 1 on crying.

Arterial blood gases indicated mild hypoxemia, and the chest x-ray showed cardiomegaly and increased pulmonary vasculature. The electrocardiogram suggested biventricular hypertrophy, and echocardiography (ECHO) showed absence of the ventricular septum and a single ventricle (Fig. 2). On electroneuromyographic (ENMG) examination, the facial nerve excitability was intact and equal on each side; the right DAOM showed diminished motor unit activity (MUA) even while the patient was crying, while the left DAOM showed normal MUA. At rest there were no denervation potentials. The other laboratory investigations were normal. The diagnosis of hypoplastic right DAOM and single ventricle was made.

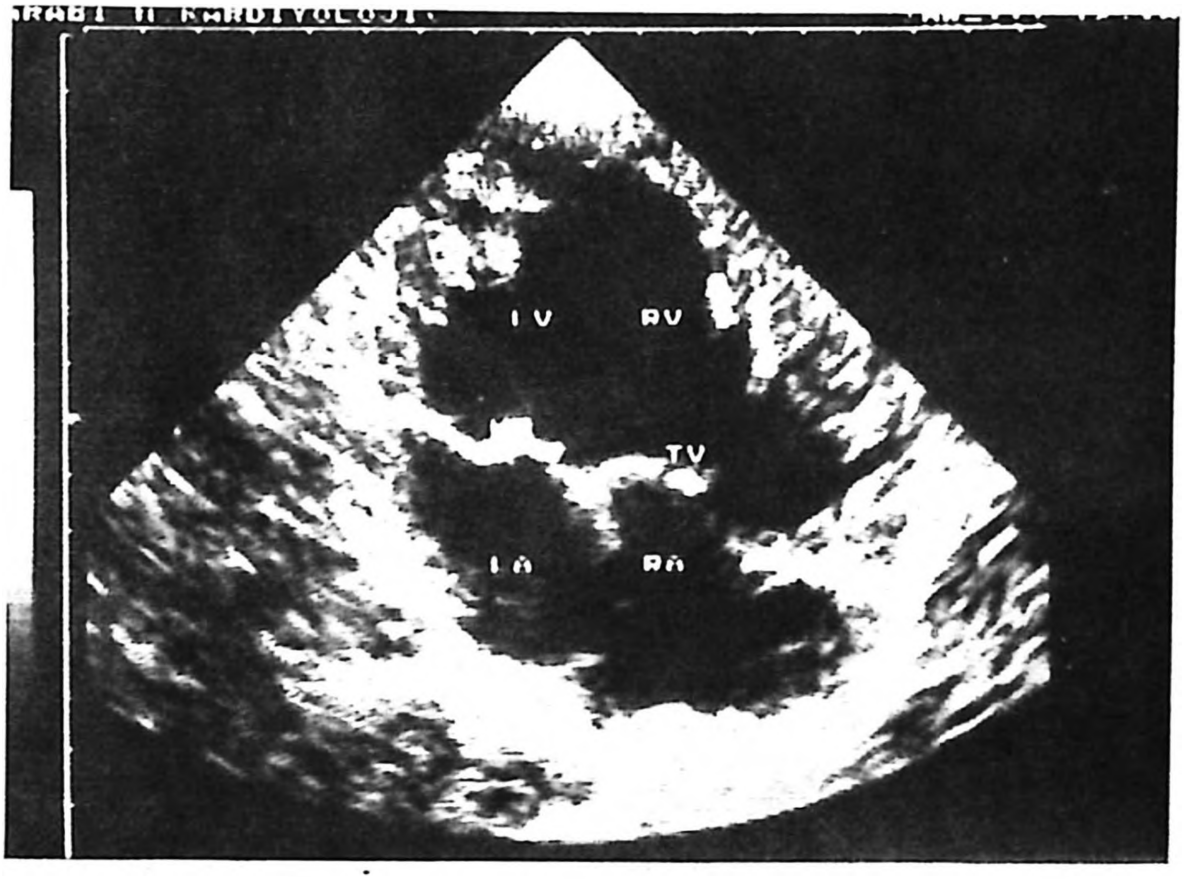

Fig. 2: Echocardiogram of Case 1 showing absence of the ventricular septum.

Case 2

A nine-month-old girl was referred to our clinic for evaluation of striking asymmetric facies when crying, intermittent cough and tachypnea. The patient was the product of her mother's third, full-term, uncomplicated pregnancy. She had two healthy male siblings and her motor mental development was normal.

The physical examination yielded the lack of downward contraction of the right angle of the mouth when crying, suggesting weakness of the right DAOM. Her pulse was 110/min, and respirations were 44/min. On the left lung, the percussion note was increased, breath sounds were reduced and a mild wheeze was heard. The heart sounds were best heard at the right sternal border. The other system examinations were normal.

Anteroposterior chest x-ray showed an over distended hyperlucent left-upper lobe with mediastinal and cardiac shift, and herniation of the left lung into the right hemithorax with compression of the right lung (Fig. 3). suggesting congenital lobar emphysema (CLE). The chest tomography findings were similar to those of the chest x-ray. Bronchography demonstrated incomplete distal filling of the left-upper bronchi. ENMG suggested the diagnosis of hypoplastic right DAOM.

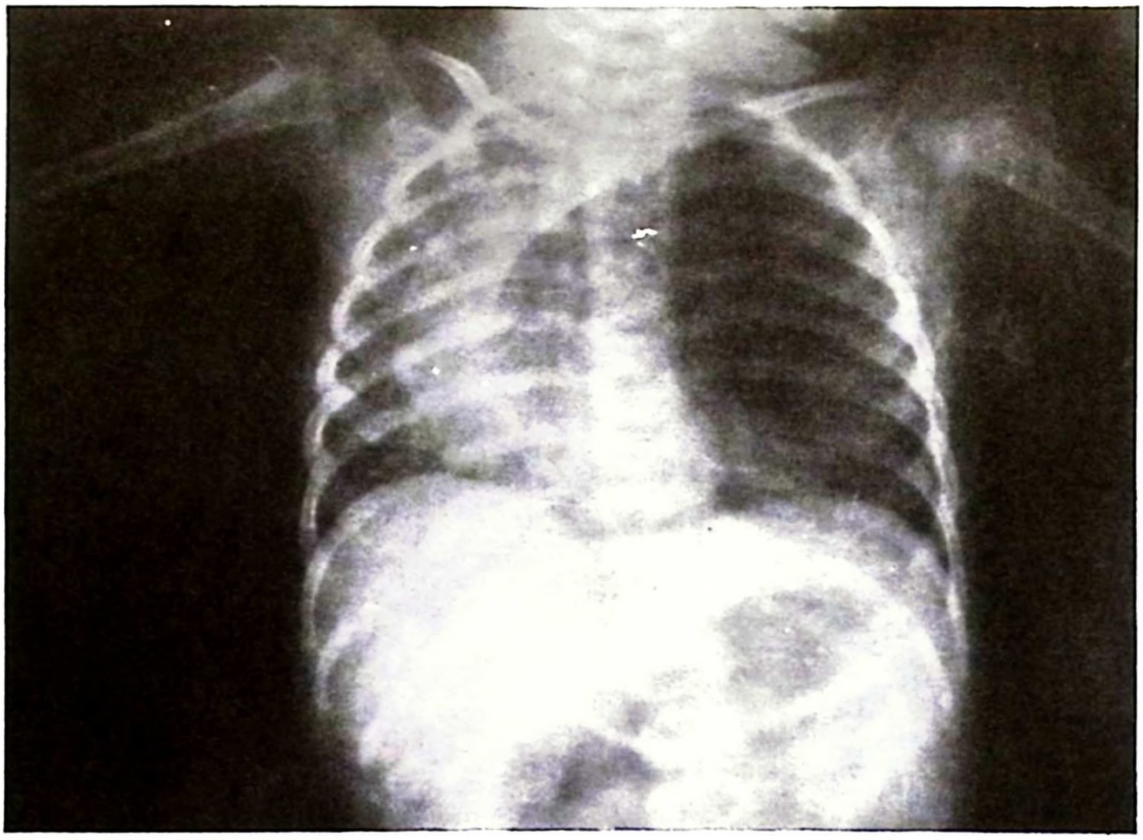

Fig. 3: Chest x-ray of Case 2 showing an overdistended, hyperlucent left-upper lobe with mediastinal and cardiac shift, and herniation of the left lung into the right hemithorax.

The patient's pulmonary disease was treated with medical and supportive therapies for three months without improvement; then she was operated on and a left-upper lobectomy was performed. Macroscopic and microscopic examination of the excised material suggested the diagnosis of CLE. After surgery demonstrated near-normal total lung functions.

Case 3

A two-day-old male infant was admitted to our clinic with the complaint of multiple anomalies. He was his mother's first child and the product of a full-term uncomplicated gestation, labor and delivery. His parents were healthy and first-degree relatives.

Physical examination revealed left-sided ACF. His weight was 3400 g and length was 50 cm. He had micrognathia, cleft plate, short neck, bilateral high-set scapula and barrel thorax (Fig. 4). The rest of the physical examination findings were normal. ENMG suggested the diagnosis of hypoplastic left DAOM. The other investigations were normal.

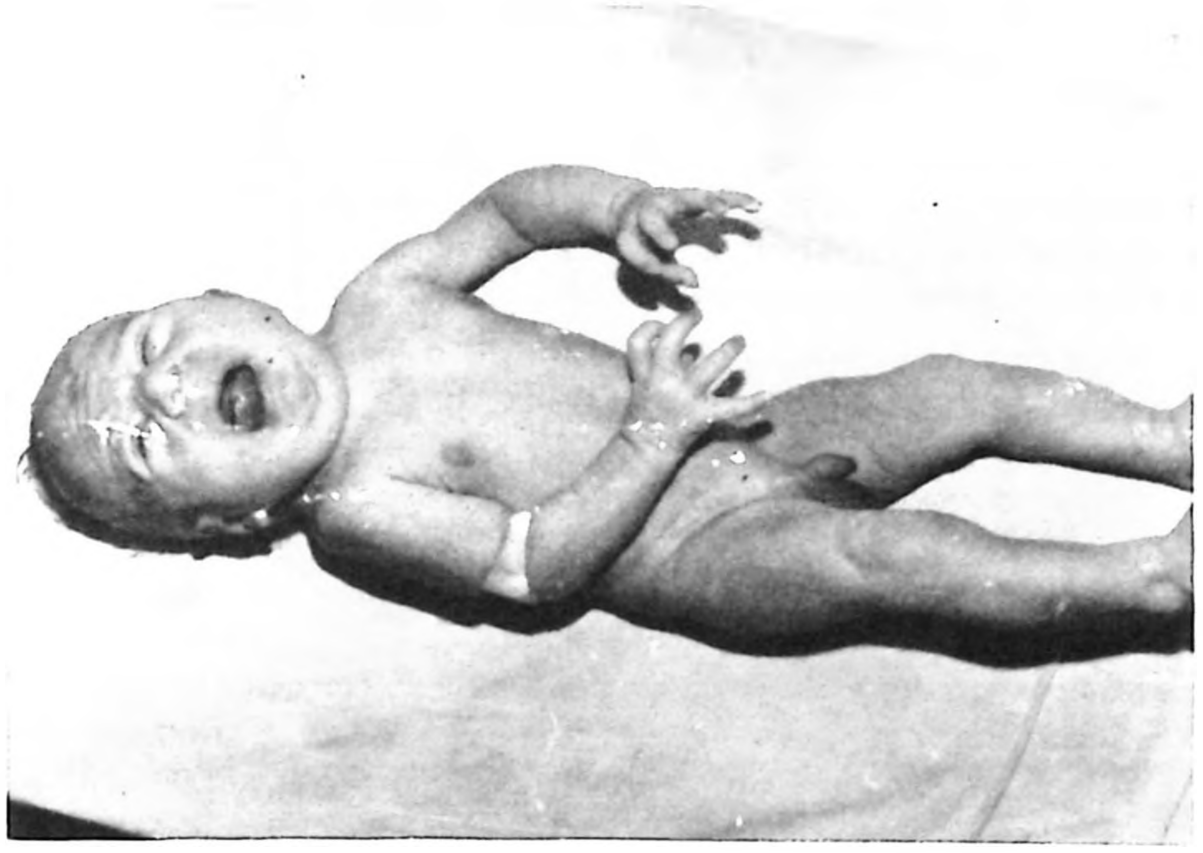

Fig. 4: Photograph of Case 3 showing ACF, micrognathia, short neck, high-set scapula and barrel thorax.

Discussion

Congenital hypoplasia of the DAOM is a rare congenital anomaly which is sometimes confused with congenital facial palsy[4]. The etiology of this anomaly, which causes ACF in an infant, is still debated[5]. It is proposed that it has a polygenic inheritance[5]. Its frequency, as calculated in two large newborn surveys, varies from 1/122 to 1/155[4]. The diagnosis of ACF is basically clinical. It is necessary to make a differential diagnosis of the paralysis of the seventh cranial nerve using electrophysiological techniques[2].

The interesting aspect of this abnormality lies in the frequently associated malformations. First Cayler[6] described the association of ACF with congenital heart defects, and proposed the name "Cardiofacial Syndrome" for this new condition. A number of associated anomalies other than cardiac have been subsequently observed in patients with ACF. Urogenital, musculoskeletal, respiratory and cervicofacial defects are most frequently associated[1,2,5,6]. In 1987, de Echaniz et al.[2] stressed the high incidence of an association with congenital malformations (eight times that in the general populations). Therefore it is suggested that ACF can be used as an index of congenital anomalies[1,7,8], and there is evidence that the condition is genetically determined[5].

The presence of a single ventricle is a rare congenital heart disease. As far as we know, two cases with ACF associated with a single ventricle have been reported to date[6]. We also report here an infant with ACF and a single ventricle.

Congenital lobar emphysema in infants is characterized by severe overinflation of the pulmonary lobe, usually causing severe respiratory distress[9]. A familial occurrence has been reported[9]. Our Case 2 may be the first reported case in the literature with the combination of ACF and CLE.

Cleft palate has been previously reported in a few cases with ACF[5], but micrognathia, short neck, high-set scapula and barrel thorax are reported here for the first time (Case 3).

Finally, in light of the literature, we suggest that all children with ACF be carefully investigated for other congenital anomalies.

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