

# Congenital unilateral lower lip palsy and eventration of diaphragm

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## ABSTRACT

**Congenital unilateral lower lip palsy is a rare but well-known limited variation of congenital unilateral facial palsy. We report a three-month-old boy with diaphragmatic eventration and isolated lower lip palsy, a combination that to our knowledge, has not been described before. Probable causes of this combination of multiple congenital malformations, in this case, could be due to nonrandom and heterogeneous mutations. The diaphragmatic eventration was treated successfully.**

**Keywords: congenital facial nerve palsy, congenital lower lip palsy, diaphragmatic eventration, facial nerve palsy**

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## INTRODUCTION

Congenital unilateral lower lip palsy (CULLP) is a rare but well-known limited variation of congenital unilateral facial palsy (CUFP).<sup>(1)</sup> We report a rare combination of diaphragmatic eventration and CULLP that has not been described before.

## CASE REPORT

A three-month-old boy, a product of first degree consanguineous marriage, was born by spontaneous vaginal delivery after 38 weeks gestation. He weighed 2,580 g and Apgar scores were nine and ten at one and five minutes, respectively. He was discharged on the second day after delivery. He required hospitalisation for pneumonitis once. The baby was brought back to the hospital for vomiting. He was healthy apart from tachypnoea, which was aggravated by feeding and was associated with vomiting. There was no cough, cyanosis or history of upper respiratory tract infections. On examination, the boy was found to be active and weighed 3,200 g. The respiratory rate was 50/min, and the air entry was decreased in the left lower lung field. General examination showed asymmetric crying facies resulting from the preserved depressor anguli oris muscle, suggestive of CULLP (Fig. 1). This facial anomaly was noted by the parents at birth. There was no family history of



**Fig. 1** Clinical photograph shows asymmetry of the lower lip while the child is crying.



**Fig. 2** Frontal chest radiograph shows the abdominal contents in the left chest.

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**Fig. 3** Intraoperative photograph shows the defect in the diaphragm with the abdominal contents entering into the chest.



**Fig. 4** Intraoperative photograph shows the defect in the diaphragm after dissection, which was subsequently repaired.



**Fig. 5** Postoperative frontal chest radiograph shows good expansion of the left lung.

congenital facial paralysis. Pupils were normal in size and reactive to light. His palpebral fissures were symmetrical and extraocular movements by Doll's manoeuvre were normal. Blinking was normal. Facial sensations were normal. There was brisk response to loud sounds. The child was crying well with normal sucking and swallowing. Anterior fontanelle was open and lax. Head circumference was 37 cm. Tongue was normal in shape and bulk. There were no features of craniofacial malformations (i.e. epicanthic folds, flattened nasal bridge, hypertelorism, microphthalmia, micrognathia, high arched palate, dental defects, lacrimal duct defects, and external ear defects). There was no evidence of other congenital malformations (i.e. metacarpal hypoplasia, brachydactyly, syndactyly, or camptodactyly, talipes equinovarus with lower leg hypoplasia, scoliosis, kyphosis, lumbar vertebral defects, and aplasia of abdominal muscles). The heart sounds were normally heard and there were no additional sounds. Chest radiograph showed raised left hemidiaphragm and a shift of the mediastinum to the right (Fig. 2). A diagnosis of left eventration diaphragm with right CULLP was made. A left subcostal laparotomy was performed. The basal segments of the lower lobe were collapsed and liver-like in consistency (Fig. 3). The eventrated diaphragm was plicated with nonabsorbable sutures (Fig. 4). An intercostal drain was left for three days (Fig. 5). The postoperative course was uneventful, and the infant was discharged in a good condition on the seventh postoperative day. At six months follow-up, there was no recurrence of the eventration. However, the CULLP persists.

## DISCUSSION

CULLP is a different entity than the severe form of CUFP.<sup>(1,2)</sup> CULLP is also known by a variety of terms. For example, "congenital hypoplasia of the depressor anguli oris muscle"<sup>(3)</sup> or "asymmetrical crying facies" have been used to characterise this mimical dysfunction.<sup>(4)</sup> Since the depressor anguli oris muscle is present in most of these patients, the descriptive term CULLP is preferred.<sup>(5)</sup> While unilateral facial palsy might be caused by an obstetric trauma, facial palsy in the absence of trauma, either uni- or bilateral, appears to be a genetic condition for which two separate loci on 3q4 and 10q have been found.<sup>(6)</sup> In three unrelated patients, CUFP has been described as part of a wider syndrome on chromosome 22q11.<sup>(7)</sup> Bilateral anophthalmia, facial asymmetry, and psychomotor retardation have been associated with deletions of the long arm of chromosome 14 [del (14)(q22.1q23) and (14)(q22.1q22.3)].<sup>(8,9)</sup> In the present case, the causes of this combination of multiple congenital malformations, in association with eventration of the diaphragm, can

be nonrandom and heterogeneous mutations.<sup>(10)</sup> Several other groups have also suggested that the combination of symptoms is due to nonrandom mutations,<sup>(11)</sup> with a neurovascular ischaemia due to amniotic bands.<sup>(12,13)</sup>

The underlying mechanism of some of these malformations is presumed to be a disturbed migration of neural crest cells during early embryogenesis.<sup>(14)</sup> These multiple-associated anomalies reinforce the theory of a malformative process, such as a missing vacuolation and a disturbance of the facial development taking place before the second month of embryonic life.<sup>(13)</sup> Diagnosis of CULLP is made on a clinical basis. No routine electromyographical or neurographical studies are necessary.<sup>(1)</sup> Several surgical techniques are employed for the treatment of CULLP. The ideal time for the intervention, however, is controversial.<sup>(1)</sup> Some clinicians advocate early (pre-school) surgery for the animations of children's faces, while others propose surgery at a later stage, but not before adolescence.<sup>(15)</sup> Muscle transplantation for facial paralysis has been shown to be effective.<sup>(16)</sup> However, the possibilities of reconstructive surgery are limited. The majority of CULFPs are not of traumatic origin and carry a poor functional prognosis.<sup>(1)</sup> In this case, diaphragmatic eventration was treated successfully and the patient was doing well at follow-up.

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