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Congenital Unilateral Hypoplasia of Depressor Angularis Oris Musclewith Asymmetric Crying Face in Newborn

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ABSTRACT: - BACKGROUND: Asymmetric crying face is a rare congenital condition associated with abnormalities of facial musculature or facial innervation. This is characterized by facial asymmetry, especially when infant cries while other facial expressions remains intact. Asymmetric crying face is caused by congenital unilateral hypoplasia or aplasis of depressor angularis orris muscle and is often confused with facial nerve palsy which causes weakness of facial expressions. This condition is diagnosed by the clinical picture and/or an electromyographic study. We describe clinical features of congenital unilateral (right) hypoplasia of depressor angularisorris muscle in a newborn.
CASE PRESENTATION: A female term neonate was born via Caesarean section to a 30-year-old gravida 2, para 2 mother at 39 weeks gestation. Apgar scores were 8 and 9. The prenatal and perinatal periods were uneventful, and no instrumentation was used for delivery. Deviation of the lower lip to the left side while crying, suggesting right-sided muscle weakness, was noted at the time of delivery. Initial physical examination was normal with no focal neurological deficits. Magnetic resonance imaging (MRI) of the head ruled out intracranial pathology including facial nerve hypoplasia. Clinical diagnosis of Congenital unilateral hypoplasia of depressor angularis orris muscle was made .2 D Echocardiography showed small PFO (Patent foramen ovale) . Rest of screening tests were normal. The baby was later discharged after passing hearing tests.
Conclusion: Congenital hypoplasia of depressor angularis orris is a rare anomaly that manifests as asymmetric crying face. Paediatricians and otolaryngologists need to be aware of this condition. This is a benign condition and functional prognosis is poor. In an isolated anomaly no treatment is required.

Key words: Congenital anomalies, Facial palsy, Asymmetric crying

Introduction: Congenital unilateral hypoplasia of depressor anguli oris was first described by Parmalee as facial weakness apparent during crying in 1931, since then it has been implicated in the pathogenesis of asymmetric crying face [1]. Neonatal asymmetric crying facies (NACF) is often misdiagnosed though it is relatively a common problem. Most common cause of this condition is absence of the depressor anguli oris muscle (DAOM), other less common causes are absence of the depressor labii inferioris muscle (DLIM), or the compression of the mandibular branch of the facial nerve which innervate these muscles.[2] Estimated incidence of asymmetric face is 0.2%–0.6% of infants and in 80% of neonates with ACF (NACF) left-sided predominance was determined.[3,45,6] Drooping of one corner of mouth on the intact side while crying is the characterisic clinical presentation of children with asymmetric crying face. By this characterisic clinical picture and/or an electromyographic study the diagnosis can be established. [7] The B-scan ultrasound which confirms the absence or hypoplasia of the DAOM/or DLIM along with normal appearance of the face apart from the deviation of the angle of the mouth on crying which disappears on rest is diagnostic.[8]
The following are diagnostic criteria for the NACF secondary to DAOM aplasia or hypoplasia 8: (1) Unilateral downward movement of the corner of the mouth while the opposite side does not move during crying, but at rest, the face appears symmetric and normal. (2) On the affected side, usually

there is palpable thinning of the lateral portion of the lower lip . (3) Nostril dilatation with respiration and with tearing, there will be normal and symmetric forehead wrinkling, closure of eyelid, nasolabial fold depth, and frowning. (4) Nerve conduction time and nerve excitability study results are normal. (5) Demonstration of hypoplasia or agenesis of DAOM in B-scan ultrasound is the recommended radiological assessment. [9] It is difficult to obtain accurate views with computed tomography (CT) scan and magnetic resonance imaging (MRI) with the added risk of radiation on CT scan use, so they are not preferred for diagnosis of DAOM.[2, 8] The following are the diagnostic criteria of NACF secondary to compression of the mandibular branch of the facial nerve during delivery : (1) suggestive perinatal history (difficult labour, forceps delivery ,large baby, multiple births), (2) mandibular asymmetry or maxillary mandibular asynclitism, suggested in physical examination (3 Evidence of mandibular branch compression with abnormal excitability and conduction in electromyography.[8] This can be an isolated clinical finding or be coupled with other congenital malformations; in approximately 45% to 70% of cases ACF is associated with other birth defects[10]. Wide variety of congenital anomalies may be associated in children with congenital hypoplasia of depressor anguli oris which include anomalies of cardiovascular, gastrointestinal, genitourinary, skeletal, and central nervous system[11,12,13,14,15,16] . It is known as ACF syndrome, when it is associated with other anomalies. The incidence of associated common anomalies in ACF syndrome are congenital heart disease (44%), head and neck (48%), skeletal (22%) and genitourinary tract anomalies (24%). Accurate diagnosis of this subtle condition and screening for associated anomalies ensures proper management**.**

In neonatal asymmetric crying facies (NACF) family counselling about the problem, workup of different management plans with family members is essential. In NACF Patients fulfilling the above mentioned diagnostic criteria and without any associated physical abnormalities only observation is sufficient with no further work up.[2]Those with mandibular branch compression usually resolve spontaneously within few months, while muscle agenesis or hypoplasia might be less noticeable with age when the functions of other facial muscles and smiling dominate the child's facial expressions[12] . But correction by plastic surgery might be needed, if there is no appreciable improvement .[3]

We report a case of a female newborn who had an asymmetrical face when crying due to right DAOM hypoplasia with patent foramen ovale (PFO) as rare association. The main objective of this report is to shed light on this important benign entity which is usually overlooked and poses a diagnostic dilemma with misdiagnosis with congenital, developmental, and traumatic true facial paralysis with the resultant unnecessary investigations and unjustifiable management .Physicians should have a high index of suspicion for the diagnosis of NACF.

Case report: A female neonate was born via Caesarean section to a 30-year-old gravida 2, para 2 mother at 39 weeks gestation was referred to our Neonatal Intensive Care Unit (NICU). She was born at term gestation through LSCS (Lower segment Caesarean section) in view of severe oligohydramnios. Mother had two previous abortions. The Apgar scores of this baby were 8 and 9 in the first and fifth min, respectively. The prenatal and perinatal periods were uneventful, and no instrumentation was used for delivery that may have resulted in a known birth trauma. Physical examination revealed pulse rate of 150 beats per minutes, respiratory rate of 50 breaths per minute, blood pressure of 60/40 mm of Hg, temperature of 37° C, and oxygen saturation of 99% on room air. She had a birth weight of 2600 g, length of 49 cm, and head circumference of 35 cm. Other systemic examinations were normal. Deviation of the lower lip to the left side while crying, suggesting right-sided muscle weakness, was noted at the time of delivery. This anomaly did not compromise breastfeed. Initial physical examination was normal with no focal neurological deficits. She was admitted to Neonatal intensive care unit (NICU) on first day of her life with diagnosis of Neonatal asymmetric crying face for evaluation. Intracranial pathology including facial nerve hypoplasia ruled out on-contrast magnetic resonance imaging (MRI) of the head. Clinical diagnosis of congenital unilateral hypoplasia of depressor angularis orris muscle was made and screening for other associated

anomalies was done. ECHO showed small patent foramen ovale. Rest of screening tests were normal. The baby was later discharged after passing hearing tests.

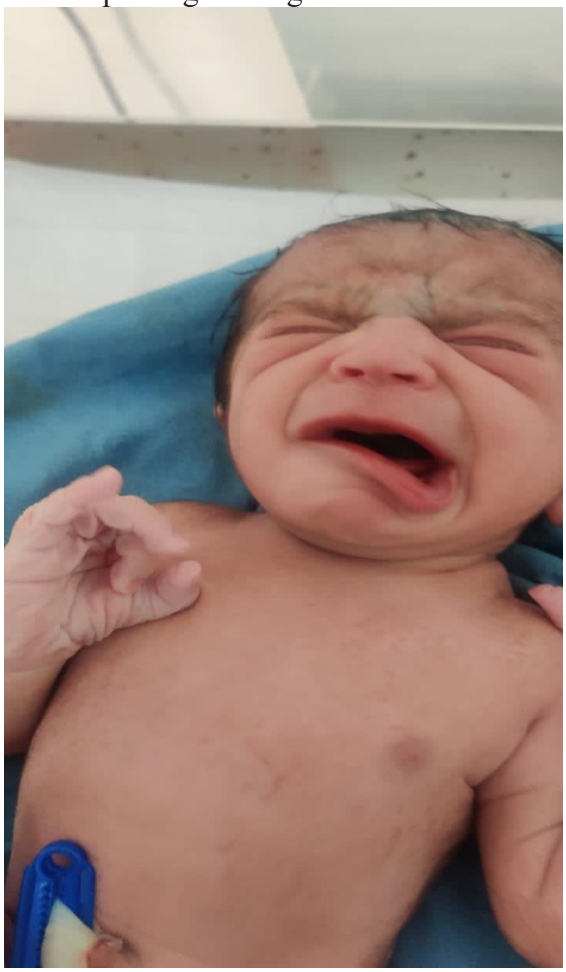

Figure-1: The asymmetrical crying face during crying in the present case.

Discussion: Neonatal asymmetric crying facies **(NACF**) is characterized by facial asymmetry during crying of a newborn, wherein one angle of the mouth deviates to one side (unaffected side), and downwards, while the other side does not move; however, at rest the face is symmetric. The underlying causes are DAOM hypoplasia or agenesis or less commonly the DLIM (the developmental theory) or injury of one of the peripheral branches of the facial nerve especially the mandibular branch which runs superficially over the mandible in neonates (the traumatic theory).[2] The major symptom of NACF is the absence or weakness while crying of the outer and lower movements of the commissure and primarily on the affected side.[17] The downward movement of the lower lip is the result of the action of 4 muscles, the DAOM pulls the corner of the mouth downwards, laterally, and averts it, while lower lip is depressed by the DLIM which extends from the mandible to the lower lip, lower lip is raised and protruded by the mentalis muscle and the platysma muscle which blends in with the DAOM and assists in its function. [8]

The NACF aetiology is multifactorial which includes intrauterine viral infections, chromosomal aberrations, hereditary factors, or a defect located at the brainstem level which can cause a defect in the depressor anguli oris muscle (DAOM) development, one of the muscles that control the movements of the lower lip, on one side of the mouth or nerve development. [18, 19] Autosomal dominant inheritance with variable expressivity and familial occurrence of the disease has also been suggested.[8,20] Association with Chromosomal anomalies, such as 22q11.2 deletion have been revealed by some studies. [21, 22] Therefore, when ACF is suspected in a newborn, genetic deletion testing should be suggested.

NACF has been considered as an indicator of coexisting anomalies, involving nearly all systems, such as the cardiac system, gastrointestinal, genitourinary system, central nervous system, respiratory system, cervicofacial region, musculoskeletal system, skin, and soft tissues, [8] Most commonly involved being the cervico- facial region and cardiovascular system. Major cervicofacial malformations occur frequently on the same side as the DAOM hypoplasia.[21] As per some reports neonates with NACF have 3.5-folds higher risk of associated major congenital anomalies like cardiac,[23,24,25] cervicofacial [26](auricular, mandibular hypoplasia),[27,28] neurological (corpus callosum agenesis, brain cyst, hydrocephalus),[27] gastrointestinal (mega colon, imperforate anus, oesophageal atresia, inguinal hernia), [27,28]genitourinary (cryptorchidism, hypospadias, vesicoureteral reflux, hydronephrosis),[27,28] skeletal (syn/poly/clinodactyly, cortical thumb, and hemi vertebra),[27,28] and genetic syndromes (4P deletion, trisomies 21or 18, Klinefelter, VATER, or Griscelli). [29, 30, 31] Hemi hypertrophy, [32] cystic lymphangioma, [33] collodion baby,[34] and pulmonary agenesis are the Other reported associations with NACF[35] . Some minor anomalies also described to be associated with NACF are strawberry haemangioma, anal/preau- ricular tags, pilonidal sinus, accessory nipple, and single horizontal palmar crease.[27,28] Ear and feet deformations like over folded helix, cup ear, protruding earlobes, and pes valgus/or varus were also described to be associated with NACF. [27] Further genetic evaluation is essential if abnormal cardiovascular findings are detected, to rule out chromosome 22q11 micro deletions which have a known association with NACF.[24, 25] The diagnostic dilemma posed by NACF is of utmost importance as it may misdiagnosed commonly as traumatic, congenital, or developmental true facial paralysis which may be isolated or part of syndromes like Mobius, CHARGE, Goldenhar, hemi facial macrosomia, and hereditary developmental facial paresis.[36]

Physicians should be aware of NACF syndrome and apply its suggested diagnostic criteria to avoid unnecessary workup and to provide the appropriate management plan.

In our case we found an ejection systolic murmur of grade 2 in left 2 nd and 3 rd intercostal spaces. The 2 D Echocardiography revealed patent foramen ovale with left to right shunt.

ACF may present in isolation or as part of some other syndrome, like Digeorge syndrome and VACTERL syndrome (vertebral anomalies, anal atresia, cardiac defects, trachea oesophageal fistula and/or oesophageal atresia, renal and radial anomalies, and limb defects).37In this case; the prenatal ultrasound not detected any obvious anomalies of the foetus. Prenatal ultrasound cannot detect isolated anomalies, and some defects might not be evident in a newborn. In such situations, a thorough physical examination and future screening should be conducted. In isolated anomalies no treatment may be required if the cosmetic problems are minor. Some studies reported that to restore the aesthetic appearance of the asymmetrical lower lip some patients underwent bidirectional facial grafting in horizontal and vertical directions.[38]

Most often NACF is confused with facial nerve palsy secondary to trauma, facial nerve compression of the foetus in the uterus, or developmental aetiologies accompanied with this facial anomaly. in NACF a thinner lower lip on the affected side, retention of normal bilateral wrinkling of the forehead, deep nasolabial folds and closure of the eyelids, can be observed.[8] All these features are present in our case (Figure-1).Congenital facial nerve palsy is manifested with facial asymmetry both at rest and while crying, without other malformations, and to confirm this condition electro diagnostic testing should be done. The use of ultrasound to observe facial muscles could be helpful for differential diagnosis as found by Gupta and Prasad at al[39]. In foetus with NACF ultrasonography could show palpable thinning or absence of the lateral portion of the lower lip, usually on the affected side.[8, 40]

Conclusion: In conclusion, for the early diagnosis of NACF, careful physical examination of newborns and genetic testing are important. For the differential diagnosis of NACF from congenital facial nerve dysplasia, ultrasonography and electro diagnostic testing could be helpful. In addition, an accurate diagnosis is important for parental counselling and to provide them with information regarding the prognosis, recurrence risk, and future diagnostic and treatment options of NACF.

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