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Case Report: Giant Bilateral Open Lip Schizencephaly in Newborn Girl

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Abstract

Introduction: Schizencephaly is a rare congenital disorder characterized by the presence of a cleft that extends from the cerebral cortex to the lateral ventricles. The prevalence of schizencephaly is estimated to be 1.48 per 100,000 births. Currently, it is divided into three types: trans-mantle, closed-lip and open-lip.

Case Description: A newborn female was admitted to the neonatology intensive care unit immediately after birth due to multiple birth defects including frontal bossing, low-set ears and digits malformation. This was accompanied by neurological manifestations; mainly generalized hypotonicity. Thorough investigations revealed a large bilateral open lip schizencephaly.

Discussion: Congenital malformations are present in 2-3% of all births globally. It ranges from mild cosmetically repaired defects to life threatening conditions. Open lip schizencephaly is an extremely rare neurologic malformation that has multiple presentations and prognosis depending on the extent of brain tissue affected.

Conclusion: Congenital malformations are related to genetic and environmental factors. This is why a multidisciplinary approach is required in most cases.

Keywords: Open lip Schizencephaly; Malformation; Neonate

Introduction

Congenital malformation is any structural defect present at birth. These anomalies may have medical surgical or cosmetic consequences. Congenital anomalies can be related to genetic and chromosomal causes or to environmental causes such as intra-uterine exposure to certain viruses or teratogens. These defects can be isolated findings or a part of a more complex syndromic combination.

Epidemiologically, congenital malformations have a global prevalence of 2-3% of all births [1]. About 65% of these malformations are related to genetic basis [2]. Studies in different regions of the world showed different results regarding the frequency of each type of congenital malformation. According to one study, central nervous system malformations have the highest incidence, followed by cardiac and musculoskeletal anomalies [3].

Schizencephaly is a rare congenital anomaly due to the neuronal migration failure. This leads to the formation of a cleft extending from cerebral hemispheres to neuroepithelial cells lining the lateral ventricles. The term is derived from the Greek word "schizen" that mean "to divide". It was first introduced by Yakovlev and Wadsworth in 1946 during their studies on cadavers.

The prevalence of schizencephaly is 1.48 per 100,000 births (combined live birth/stillbirth) in one study [4]. Currently, schizencephaly is classified into three different types: type I, which is the trans-mantle form where column of abnormal grey matter extends from ependyma to the pia matter without CSF containing cleft. Type II is closed-lip schizencephaly where CSF containing cleft abutting lining lips of abnormal grey matter opposed. Type III is the open-lip form where CSF-containing cleft present, non-abutting lining lips of abnormal grey matter [5].

The case below presents an extremely rare case of giant bilateral open-lip schizencephaly with poor prognosis.

Case Presentation

A case of a female neonate born by C-section at full term. She was found at birth to have multiple congenital malformations including: frontal bone bossing, hypertelorism, microphthalmia, broad nasal bridge, cleft palate, low set ears, wide spaced nipples, syndactyly in hands, polydactyly in feet (Figure 1). On physical examination, she was found to have hypoactivity, poor suck and generalized hypotonicity. The baby was admitted to the neonatology intensive care unit (NICU) for investigations and management.

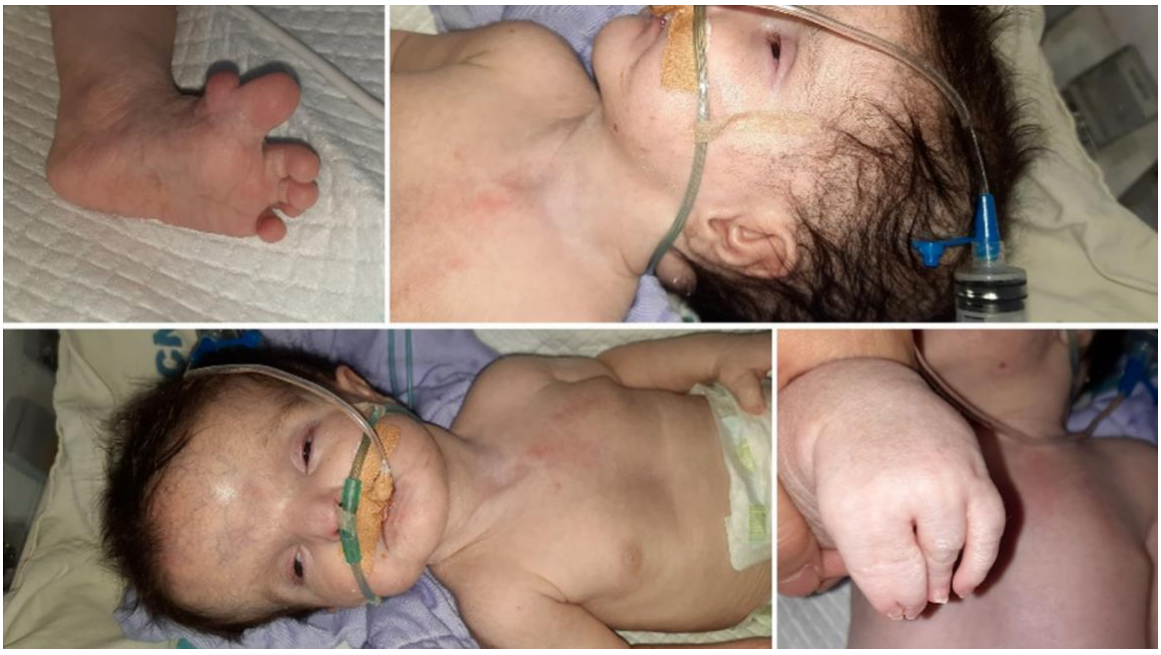

Figure 1: External malformations present at birth.

Maternal history shows 28 years old G1P1 mother with no past medical problems. She is a current smoker and receiving no medications. No substance abuse is present. The pregnancy was a product of in-vitro fertilization (IVF) following 8 years of primary infertility. This was the result of a non-consanguineous marriage. She regularly followed up with a gynecologist who suspected hydrocephalus by prenatal ultrasound. Family history is negative for medical issues.

Computed tomography (CT) scanner cerebral done upon admission to NICU due to pre-natal history of possible hydrocephalus and post-natal hypotonicity. CT scan revealed giant bilateral open lip schizencephaly, complete agenesis of the corpus callosum and the falx with dysplasia of the thalamus, cerebellum and both frontal lobes. Brain MRI was not done due to financial issues of the family. The respiratory status deteriorated progressively with need for oxygen by nasal canula to maintain normal oxygen saturation.

On the second day, the newborn started having frequent seizures. Several anti-epileptic medications failed to control them. The patient was well tolerating the oral intake by oro-gastric tube due to absence of suck reflex. The whole situation was discussed with the family who decided not to do any neurosurgical intervention and not to resuscitate in case of cardiac arrest due to the expected bad prognosis of such malformation. About 20 days later the baby girl passed away. The family was asked to perform genetic analysis of both parents before planning for a future pregnancy.

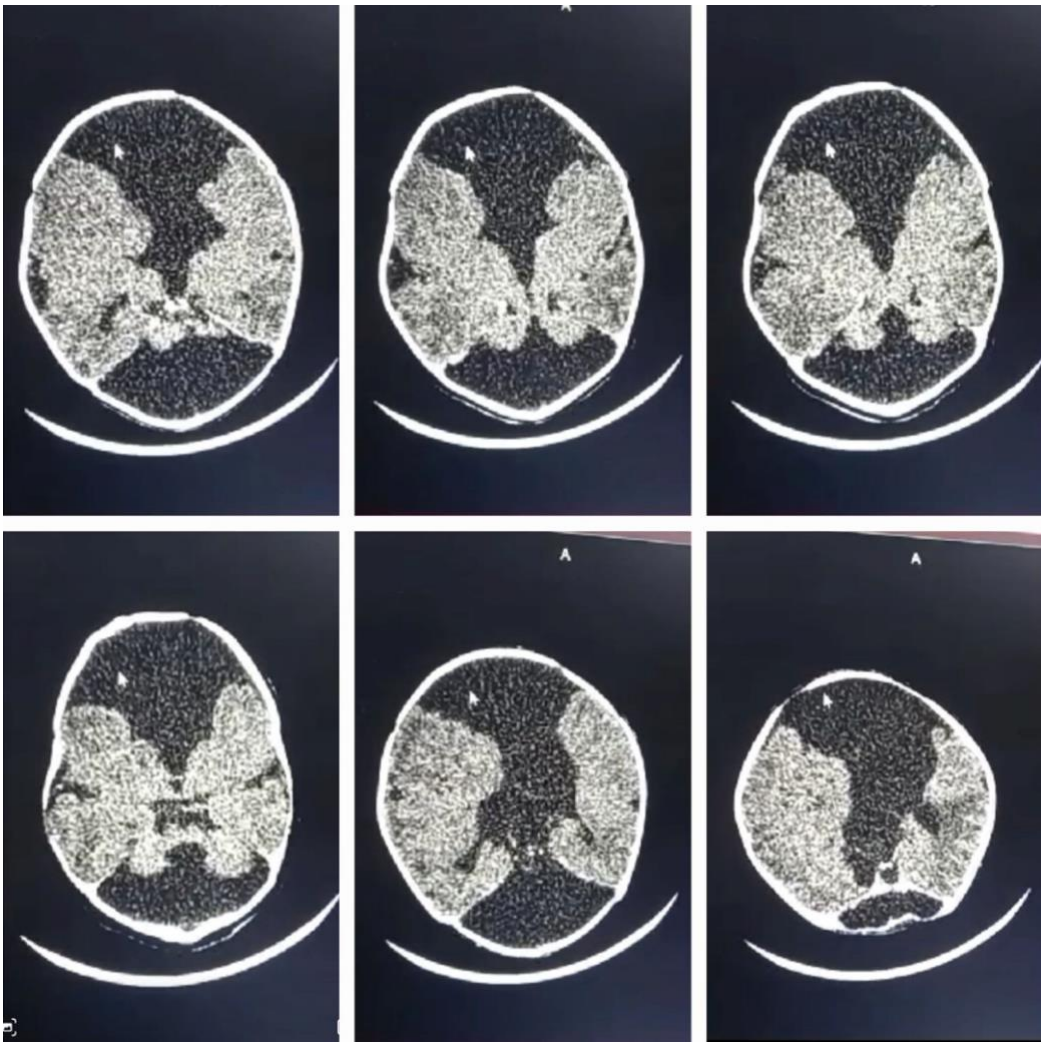

Figure 2: CT scan cerebral showing bilateral large open lip schizencephaly.

Discussion

Schizencephaly is a very rare congenital malformation of the brain. It is a defect in neuronal cell migration during the second to fifth month of gestation. The cause underlying this disorder is unknown but several theories suggested relation to teratogenic agents, ischemic insult of fetal brain tissue, viral infections and young maternal age. The case presented above shows a smooth prenatal coarse with no viral infections or teratogenic exposure. Moreover, the mother is not below 20 years of age as described in literature regarding the maternal age.

Schizencephaly can be isolated or associated with other malformations of the central nervous system. The case described above was associated with anomalies of the face and extremities.

It can be closed-lip with a mild coarse and diagnosis in adulthood due to epileptic seizures or mild motor deficit. The other type is open-lip form which has a severe coarse and is diagnosed earlier due to refractory seizures, intellectual disabilities and significant motor weakness. The newborn girl in the case above had giant bilateral open lip schizencephaly with refractory seizures and motor weakness that passed away after a sudden cardiorespiratory arrest.

Conclusion

Proper prenatal follow up is highly encouraged to avoid such unexpected birth malformations that has huge psychological impact on the family. A possible link to IVF pregnancies may be present since the literature showed other cases with schizencephaly following an IVF pregnancy.

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