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Case Report

Diprosopus tetraophthalmos: a rare congenital anomaly

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ABSTRACT

Diprosopus is an extremely rare form of conjoined twins which is found in newborns where there is partial or complete duplication of face. The etiology and pathophysiology remain unknown and no genetic mutations have definitively associated with the condition so far. This article described a case of an infant born at 33.4 weeks gestation with multiple congenital anomalies including diprosopus tetraophthalmos and discussed the possible hypothesis and implications of prenatal diagnosis.

Keywords: Diprosopus, Tetraophthalmos, Conjoined twins, Craniofacial malformation

INTRODUCTION

The term diprosopus is derived from a Greek word, meaning duplication of face. Diprosopus is an extremely rare form of craniofacial malformation where there is partial or complete duplication of face. It is a rare form of symmetrical conjoined twinning.[1] Less than 40 cases have been reported in the literature till date.[2,3] Diprosopus has a prevalence of 1 in 15 million births. Advanced maternal age, polyhydramnios, and consanguineous marriage are considered high-risk factors for diprosopus. There is a high risk of associated CNS defects, gastrointestinal defects and renal defects with complete duplication. Prenatal diagnosis using ultrasonography, computed tomography (CT) scan, and magnetic resonance imaging (MRI) are possible. Stillbirths occur in 50% of all conjoined twins and the majority of infants who are born alive do not survive long term.[4,5] If diagnosis is made early in pregnancy, termination of pregnancy is sometimes considered as an option.[6]

CASE REPORT

A 26-year female, gravida 3 para 2 living 2 (previous 2 normal vaginal deliveries) at 33.4 weeks of gestation came to the casualty with complains of pain in abdomen and leaking per vagina since 4 hours. Patient had history of two packed cell transfusion in current pregnancy in view of severe anemia. Previous two pregnancies were uneventful. On examination, patient was afebrile and vitally stable. On per abdominal examination, fundal height was corresponding to 34 weeks of gestation. Longitudinal lie and cephalic presentation with one uterine contraction in a span of 10 minutes lasting for a duration of 10 seconds. On Doppler, fetal heart sound was heard. An obstetric ultrasound was done which revealed a single live intrauterine pregnancy of 31.3 weeks with longitudinal lie and cephalic presentation. Placenta was posterior. Liquor was suggestive of polyhydramnios (AFI: 24) with estimated fetal weight of 1541 gram. Ultrasonography also revealed a large posterior fossa with big cerebrospinal fluid cyst measuring 17.6 mm. Fourth ventricle appeared open and was contiguous with posterior fossa cyst with agenesis/hypoplastic vermis, indicating possibility of dandy walker continuum/ arachnoid cyst (Figure 1).

After augmentation of labour, baby delivered in cephalic presentation. It was a male baby with weight of 1.840 kgs. Baby did not cry after birth and had no heartbeat. On examination of the baby, baby had duplication of eyes, nose, maxilla and mandible suggestive of diprosopus associated with a posterior wall cyst, possibly Dandy Walker syndrome (Figure 2-4).

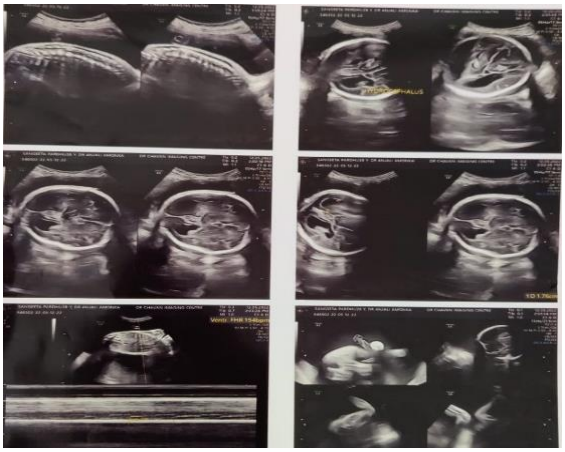

Figure 1: Dandy Walker continuum/arachnoid cyst.

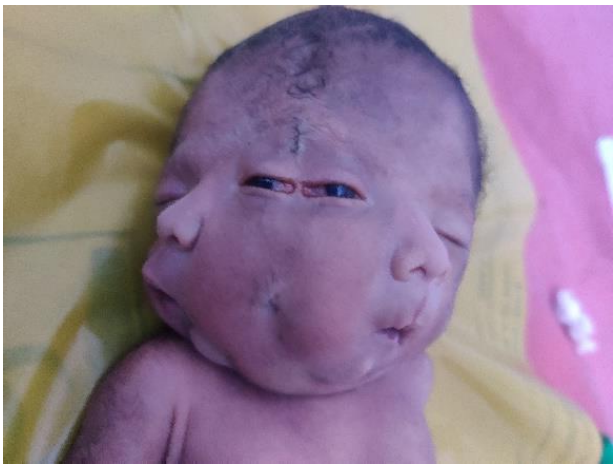

Figure 2: Diprosopus tetraophthalmos.

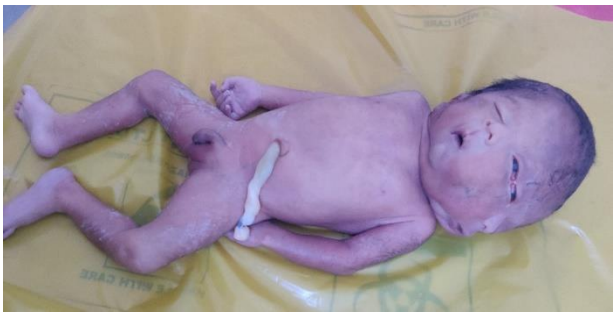

Figure 3: Baby with 2 faces, 4 eyes, 2 nose and 2 mouths and 2 mandibles.

Figure 4: Posterior wall cranial cyst.

DISCUSSION

Diprosopus is an extremely rare form of craniofacial malformation seen in newborns where there is duplication of face which may be partial or complete. Two possible mechanisms have been proposed-cranial bifurcation of the notochord during neurulation and/or an increase in the expression of the protein sonic hedgehog, which is essential for craniofacial patterning during development.[6] Other associated anomalies have been described in diprosopus comprising a neural tube defect like anencephaly or craniorachischisis, cardiac or gastrointestinal defects.[7] Risk factors for diprosopus includes familial tendencies, advance maternal age, previous history and polyhydramnios. Prenatal ultrasonography, CT scan and MRI can detect diprosopus in cases with a high degree of suspicion. Most of the patients are stillbirth and overall survival in live birth is poor due to multiple associated anomalies. Treatment options include surgical repair in selected incomplete variety and management of complications arising out of other anomalies.[6]

CONCLUSION

Diprosopus is a rare form of conjoint twins with very few reported cases in literature. Diagnosis can be made on a prenatal ultrasonography, computed tomography or MRI. If detected in early pregnancy, termination of pregnancy should be advised. Most cases usually result in stillbirth and overall survival rate is associated with very poor prognosis due to multiple associated anomalies.

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