INTRODUCTION

Ethmocephaly is the rarest subtype of holoprosencephaly, which occurs due to failure of diverticularization and rotation of the prosencephalon into two normal cerebral hemispheres within the fifth gestational week. The incidence of ethmocephaly is 1 in 15,000 live births and 1 in 250 in abortuses, thus mostly eliminated prenatally. Clinically, ethmocephaly presents with a proboscis situated above hypoteloric orbits in the midline, microphthalmos, absent nasal structures and lowset malformed ears. The condition is closely related to cyclopia, a severe form of holoprosencephaly where the two eyes are fused together in a single median orbit. Amniotic band syndrome encompasses asymmetrical congenital malformations due to ring-like constriction bands in the limbs, head, face or trunk. The case presented here involves ethmocephaly with amniotic band syndrome, which is likely the first of its kind, published in the literature.

CASE REPORT

A 1-hour-old male baby was admitted in a special care neonatal unit with respiratory distress, absent nose and closely set eyes. The baby was born normally at 28 weeks gestation to a 22-year-old primigravida mother. The mother had history of antepartum hemorrhage (APH) once in the first trimester of pregnancy. She was taking homeopathic medicines for infertility for two years prior to conception. The family history was negative for a consanguineous marriage or congenital malformation. The baby weighed 945 gm at birth, with a crown to heel length of 38 cm and head circumference of 27 cm. On clinical examination, there was ocular hypotelorism with microphthalmos in left eye, a proboscis located above the eyes in the midline, absent nasal structures and lowset malformed ears. The condition is closely related to cyclopia, a severe form of holoprosencephaly where the two eyes are fused together in a single median orbit. Amniotic band syndrome encompasses asymmetrical congenital malformations due to ring-like constriction bands in the limbs, head, face and occasionally the trunk. Here, we present a case of ethmocephaly and amniotic band syndrome involving the right thumb. To the best of our knowledge, (MEDLINE search), the association of ethmocephaly and amniotic band syndrome has not been previously described in the literature.

DISCUSSION

Embryologically, by the end of the fourth week of gestation, the forebrain vesicle prosencephalon differentiates into telencephalon and diencephalon. During the fifth week, two telencephalic vesicles form the cerebral hemispheres and lateral ventricles. The diencephalon differentiates into the thalamus, hypothalamus...
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and third ventricle. Holoprosencephaly results due to failure of diverticularization and rotation of the prosencephalon into two cerebral hemispheres within 35 days of gestation. Ethmocephaly is the rarest subtype of holoprosencephaly with undivided cerebrum and fused single ventricle, a proboscis separating hypoteloric orbits, microphthalmos, lowset malformed ears and absent nasal structures.

The occurrence of ethmocephaly is mostly sporadic but in some cases chromosomal abnormalities such as trisomy 13, 13q-, trisomy 18 and triploidy have been detected. Other causes include diabetes mellitus, ethanol, retinoids, cytomegalovirus and accidental ingestion of the Veratrum californicum plant. The prenatal ultrasonographic diagnosis of ethmocephaly is based on an absent midline echo, single dilated ventricle and fused thalami. Other congenital malformations such as hydrocephalus, arachnoid cyst, Dandy-walker malformations and hydranencephaly have to be ruled out.

Amniotic band syndrome is a spectrum of asymmetrical congenital malformations due to ring-like constriction bands in the limbs, head, face and occasionally the trunk. According to the widely accepted “extrinsic theory” by Torpin and Faulkner (1965), there is separation of amnion from chorion in early pregnancy producing free-floating tissue bands. These amniotic bands either wrap around parts of the embryo or are swallowed by the fetus causing growth restriction and structural abnormalities of the fetus.

Our case presented with ocular hypotelorism with microphthalmos in the left eye, a proboscis located above the eyes at the midline, absent nasal structures and bilateral low set ears with lobulated pinnna on the right side. The condition is differentiated from cyclopia, a severe form of holoprosencephaly where the two eyes are fused together in a single median orbit. Another differential diagnosis was cebocephaly, where a flattened nose with single nostril is located below close-set eyes. The infant also had a constrictive circumferential groove in the right thumb. The infant was diagnosed as a case of ethmocephaly with amniotic band syndrome. Based on a thorough MEDLINE search, we believe, the occurrence of amniotic band syndrome with ethmocephaly had not been previously reported in the literature.

In conclusion, ethmocephaly carries a poor prognosis, as in most cases it is incompatible with life. Hence, the early antenatal ultrasonographic diagnosis, chromosomal karyotyping, parental counseling and termination of pregnancy can reduce the incidence of stillbirth and perinatal mortality.

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