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Case Report

Ethmocephaly: a rare but lethal congenital anomaly

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ABSTRACT

Ethmocephaly is a rare but lethal congenital anomaly. Its incidence rate is 1 in 15,000 live births and 1 in 250 in abortuses. Babies have typical dysmorphic facies in form of orbital hypotelorism, proboscis and low set malformed ears. 32 years old, daily wage labourer delivered a baby with ethmocephaly. Baby had typical facies, and no extracranial abnormality present. Baby was stillborn. She had no antenatal checkup or ultrasonogram done. However, she had history of ingestion of certain plants during early months of pregnancy. Ethmocephaly is a lethal anomaly and mostly eliminated prenatally. Although there is no diagnostic test for ethmocephaly, and confirmation of it is done only by appearance of baby after birth, important clues like recognition of risk factors like maternal diabetes, alcohol intake, ingestion of certain plants, antenatal hydrocephalus, should not be missed. Importance of routine antenatal checkup and ultrasonogram is emphasized. Recognition of risk factors and a possible co-relation with ingestion of certain other plants is also emphasized.

Keywords: Ethmocephaly, Dysmorphic facies, Proboscis, Risk factors, Antenatal ultrasonogram

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.[1]

Facial dysmorphism includes proboscis which separates the narrow set eyes, microphthalmos, low set malformed ears. Orbital hypotelorism is the characteristic clinical finding in making the diagnosis of the aforementioned but it closely resembles cebocephaly which has hypotelorism with proboscis having single nostril or septo-optic dysplasia. Its incidence rate is 1 in 15,000 live births and 1 in 250 in abortuses, thus mostly eliminated prenatally.[2]

CASE REPORT

32 years old woman, presented to our hospital with decreased fetal movement for 3 days and leaking per vaginum for 1 day. Patient had non-consanguineous marriage, was G3P2+0L2, and had gestational age of 35 weeks. Her previous two pregnancies were uneventful and she had no history of abortion.

Patient was a daily wage labourer, had no antenatal checkup or ultrasonogram done. She had no history of diabetes, hypertension, hypothyroidism, any drug intake or infections during pregnancy. However, she had history of intake of certain plants during early pregnancy, details of which could not be elicited.

Stillborn female baby at gestational age of 35 weeks, was delivered through lower segment caesarean section (LSCS). On gross examination; baby was noted to have severe facial dysmorphism; presence of two very closely placed eyes; absence of nasal aperture and presence of ‘snout-like’ tubular appendage above it suggestive of non-functional nose, absent philtrum, low set ears, no other gross congenital anomaly noted and placenta was normal.

Magnetic resonance imaging (MRI) brain, histological, biochemical, chromosomal analysis and postmortem biopsy was not done as consent for these procedures was denied by father.

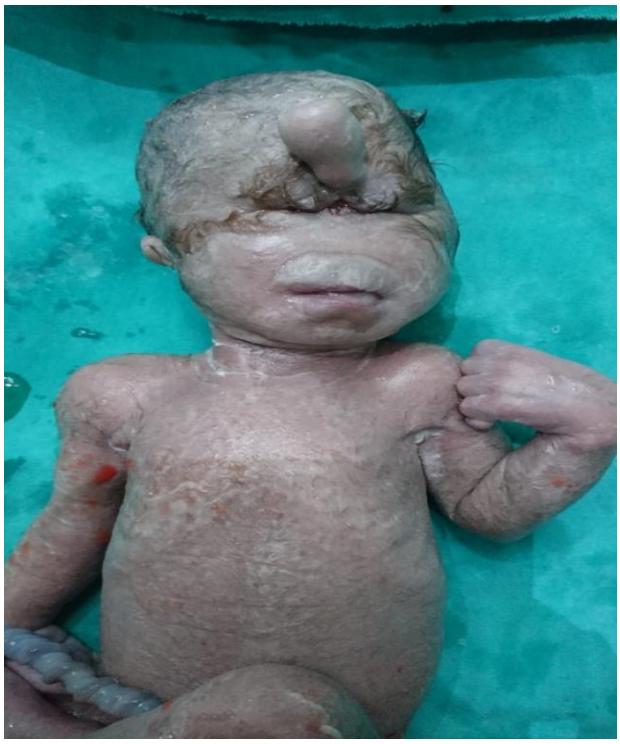

Figure 1: Newborn with ethmocephaly showing facial dysmorphism: orbital hypotelorism, proboscis, and low set malformed ears.

DISCUSSION

Ethmocephaly is a rare but lethal congenital malformation whose etiology is mostly unknown. Possible risk factors include: maternal diabetes (1% risk and a 200-times increase in fetal holoprosencephaly), infections during pregnancy (TORCH), drug intake during pregnancy (alcohol, anticonvulsants, aspirin, lithium, retinoic acid, anticancer agents, and fertility drugs), and genetic causes (twins and in consanguineous marriages).[3-7]

Sonography is the most helpful in the prenatal diagnosis of holoprosencephaly.[8] Holoprosencephaly can be expected to be present in 16% or more of all cases of fetal hydrocephalus.[9]

In our case, no antenatal checkup or ultrasonogram was done. At birth, our case had typical facial features of ethmocephaly (facial dysmorphism- orbital hypotelorism, low set malformed ears, and proboscis). Apart from facial dysmorphism, our patient had no extracranial features. Our case passed undiagnosed during pregnancy as no antenatal checkup or ultrasonogram was done. So, even important diagnostic clues like fetal hydrocephalus were missed.

An important risk factor related to holoprosencephaly is Veratrum californicum, if ingested in first few months of pregnancy.[10] Our patient had an history of ingestion of certain plants during early pregnancy, but we could not elicit the details of it. Although Veratrum californicum is mainly found in the high mountain ranges of the western United States, our case raises an important possibility of certain other plants causing this.

CONCLUSION

Ethmocephaly is a rare and lethal congenital anomaly. Despite the increasing healthcare facility and awareness, a large sum of people is still there who have less awareness and access to healthcare facilities like routine antenatal checkups and sonography. Importance of antenatal checkups, sonography and termination of pregnancy for preventing further psychological trauma to family is emphasized. Risk factors like maternal diabetes and alcohol intake, and a possible co-relation with ingestion of certain plants is also stressed upon.

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