DOI: 10.7860/JCDR/2015/12950.6344

Case Report

Obstetrics and Gynaecology Section

Proboscis Lateralis : A Rare Bilateral Case in Association with Holoprosencephaly

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ABSTRACT

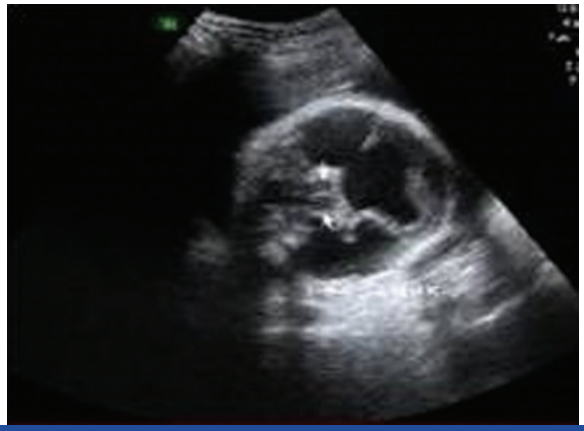
Proboscis lateralis is a very rare congenital craniofacial malformation characterized by a trunk like tubular appendage arising commonly from roof of the orbit near medial canthus. It may be seen as an isolated defect with sporadic occurrence or it may be associated with a spectrum of anomalies. It is usually unilateral and very few bilateral cases of proboscis lateralis have been reported in the literature worldwide. Alobar holoprosencephaly is commonly associated with a single central proboscis and cyclopia. Here we report an unusual case of a bilateral proboscis lateralis seen in association with holoprosencephaly.

Keywords: Arhinencephaly, Craniofacial malformations

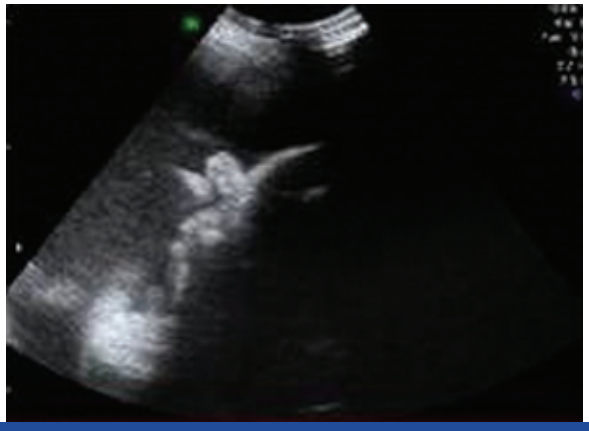
CASE REPORT

A 29-year-old primigravida at 30 weeks of gestation came for antenatal check-up. She had an uneventful pregnancy till then. There was no family history of anomalies. There was no history of infection, drug abuse or radiation exposure. There was no history of diabetes mellitus. Her serological screening for HIV, hepatitis and syphilis was negative. Her family history was unremarkable. She did not undergo anomaly scan in second trimester. She was referred to Department of Obstetrics and Gynaecology, Mahatma Gandhi Medical College and Research Institute, Puducherry, India, for detailed ultrasound scan.

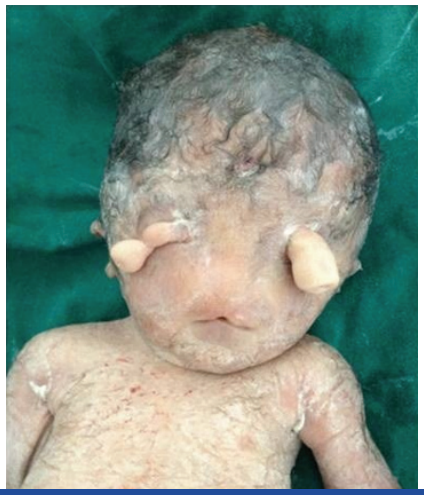
Trans-abdominal ultrasound was done in volusion 730 promachine using 3D probe (3.5Mhz). A single live intrauterine fetus of gestational age 28 weeks 2 days was seen. The supratentorial fossa was CSF filled, with centrally fused thalami devoid of midline structures at that level. A dilated communicating single lateral ventricle with rudimentary occipital horns, depicting features of semilobar holoprosencephaly was seen [Table/Fig-1]. Both the globes were not visualized and a snout like protrusion suggestive of proboscis was seen [Table/Fig-2]. Fetal MRI was done and it showed similar features suggestive of semilobar HPE. The parents were informed and counselled regarding the severity and prognosis of the condition. Genetic screening was advised to the parents; however they did not undergo the screening. She came back in preterm labour and delivered a stillborn female fetus weighing 1.5kg. The fetus showed anophthalmia, bilateral proboscis arising from medial aspect of the orbit on either side, absent nasal bridge and nose, wide open anterior fontanelle and a median cleft lip [Table/Fig-3]. No other obvious anomalies of limbs or other parts of the body were seen. The parents have not consented for an autopsy of the fetus.



[Table/Fig-1]: Ultrasound image of the fetus showing Holoprosencephally



[Table/Fig-2]: Ultrasound image showing Proboscis



[Table/Fig-3]: Showing still born fetus witb Bilateral Proboscis Lateralis

DISCUSSION

Proboscis lateralis is a rare craniofacial malformation. Bilateral proboscis has been reported in only two cases so far. In 1861, Forster first described proboscis lateralis in his monograph on congenital malformations of the human body [1]. Selenkoff reported this anomaly in the autopsy findings of a 34-year-old finnish farmer

[2]. It is one of the rarest facial anomalies with incidence of less than 1 in 1,00,000 live births [3,4]. The nasal cavity on the affected site is replaced by a tubular appendage located off-center from the midline of the face, arising commonly from medial aspect of the roof of the orbit. It is usually associated with heminasal aplasia or hypoplasia, microphthalmia and less commonly with midline clefting [5]. Associated Brain and cranial vault anomalies are seen in 19% of the patients [4]. It is usually unilateral and very few symmetric bilateral cases have been reported [6]. We have encountered only two case reports on bilateral proboscis lateralis [6,7].

Holoprosencephaly, resulting from failed or incomplete division of prosencephalon, is associated with median facial anomalies in 80% of the cases [8,9]. Its most severe form, the alobar holoprosencephaly is associated with cyclopia and proboscis which is single and centrally located [9]. According to DeMyer, proboscis lateralis may also be seen in association with holoprosencephaly [9]. However no case of bilateral proboscis lateralis with holoprosencephaly has been reported so far. Hence we thought it is worth reporting our case, a very rare anomaly of bilateral proboscis lateralis in unusual association with holoprosencephaly.

Proboscis lateralis. When occurs singly, with a normal nose and no other deformities it has better prognosis and amenable to treatment with nasal reconstruction. However, when seen with CNS abnormalities as in our case, prognosis is poor. In 1985, Boo-Chai K classified proboscis lateralis into four groups [1,5,10-14].

Group 1: Lateral proboscis with normal nose (least common),

Group 2: Lateral proboscis with an ipsilateral deformity of the nose,

Group 3: Lateral proboscis with ipsilateral deformity of nose, eye and or ocular adnexa,

Group 4: Lateral proboscis with ipsilateral deformity of nose, eye and or ocular adnexa, along with cleft lip and /or palate.

In this classification there is no separate group assigned for the cases of proboscis lateralis showing bilaterality or association with CNS abnormalities like holoprosencephaly. Hence, probably our case, inspite of showing both these uncommon features, falls into group 4 of Boo-Chai's classification. However, Sakamoto et al., proposed a new classification scheme for proboscis lateralis based on a review of 34 studies involving 50 cases, to differentiate the cases of proboscis lateralis associated with holoprosencephaly, from those associated with other anomalies with good prognosis [11]. This classification system is based on intercanthal distance. In addition to the original four groups of Boo-Chai's classification, they introduced two new groups, Group 5: proboscis lateralis with encephalocele and Group 6: Proboscis lateralis with holoprosencephaly. According to this classification our case seems to fall under group 6 but differs in its bilateral occurrence. Out of the 50 cases reviewed by Sakamoto et al., 17 cases were seen in association with holoprosencephaly (group 6). However, there was no mention of any bilaterality in any of these cases. Further, among the two other case reports of bilateral proboscis that we encountered, neither of them showed association with holoprosencephaly. So probably ours is the first case in which bilateral proboscis, one of the rarest congenital facial anomaly is seen in association with holoprosencephaly.

CONCLUSION

Bilateral proboscis lateralis is an extremely rare congenital anomaly. When such an anomaly is encountered, associated CNS abnormalities like holoprosencephaly must be looked for. Because such a co-existence, though uncommon, is a poor prognostic factor for this structural deformity which is otherwise amenable to surgical reconstruction.

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FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Submission: **Jan 10, 2015**
Date of Peer Review: **May 04, 2015**
Date of Acceptance: **May 13, 2015**
Date of Publishing: **Aug 01, 205**