Case Report

A Boy with Bilateral Complete Cryptoph-thalmos in Pakistan with Subsequent Blaming and Shaming for his Mother

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Cryptophthalmos is a very rare congenital anomaly, in which there is developmental defect in the surface ectoderm part of the eye, with partial or complete failure of eyelid formation and is characterized by skin passing continually from the forehead to the cheek over a malformed eye, or failure of separation of lids. As in Fraser syndrome[1] it is mostly accompanied by urogenital anomalies, syndactyly, and cognitive disorders.[1] We report a unique case of bilateral complete cryptophthalmos in Pakistan where the mother was blamed for being responsible for the birth of malformed child.

CASE REPORT

A seven-day old full-term male infant presented to the eye outpatient department of Aga Khan University Hospital, Karachi with bilateral complete unformed lids, with continuous skin covering both the eyes. He is the first child of his parents who come from the low socio-economic class and was delivered by lower segment cesarean section in a local hospital in Karachi. According to his mother he had spontaneous breathing after delivery. She had no complication during delivery. His birth weight was 2.2 kg. His parents were non-consanguineous with was no family history of such condition. His appetite was good, bowel habits were normal and sleep was also normal.

On ocular examination, there was complete closure of both eyelids by skin from the forehead (Figure 1). No eyelashes or eyebrows were seen. On putting very bright light on the closed eyes, there were sudden head movements. There were also movements behind the closed lids. On digital ocular examination, soft globes were palpable. On B-scan ultrasonography, both eyes showed a malformed anterior segment, but formed vitreous cavity.

On general examination, the body weight was 2.9 kg. Head circumference was normal for age. Both ears were small and malformed. His nasal bridge was broad and depressed. There was a swelling in the right inguinal region, suggestive of inguinal hernia (Fig.1).

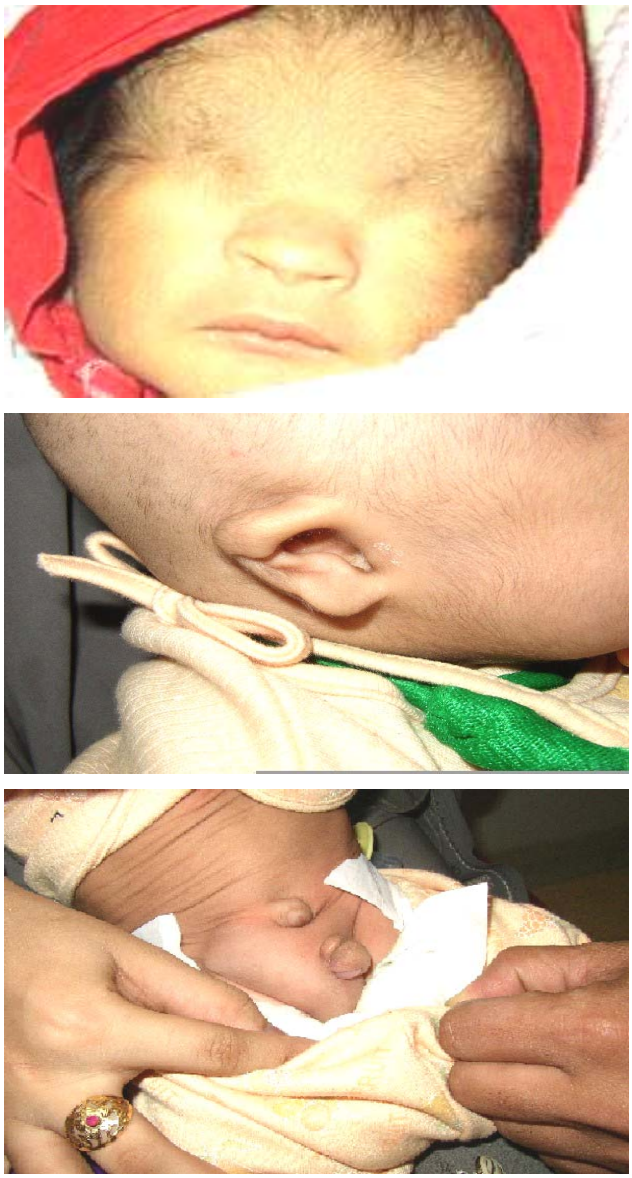

Fig. 1: Showing broad & depressed Nasal bridge, small malformed ears and swelling at right Inguinal area.

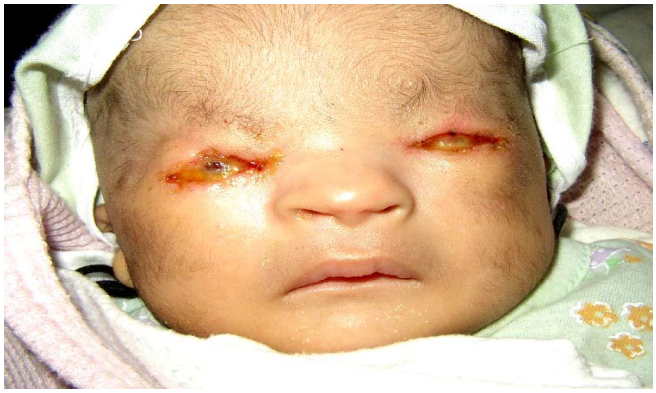

Fig. 2: The children 24 hours after the surgery

The child underwent reconstructive surgery with the aim to form the lids (Fig.2). The surgery was performed under general anesthesia and was done jointly by an ophthalmologist (TAC) and a plastic surgeon. During surgery, it was noted that the cornea were adherent to the skin and the eyes were small with completely malformed anterior segments. Lids were created and conjunctiva was stitched to its inner surface in both eyes. The child was sent for conformers to be fitted in both sockets with the hope to prevent future adhesions. It was very difficult for them to find conformer of that small size and the smallest size found was fitted in with difficulty. Few days later, the conformer fell off the sockets and lids started to adhere again. Parents were advised further surgery in future but they got lost to follow up citing financial and social reasons. The mother was only 18 years old and she reported that her in laws were holding her responsible for her malformed child. They went on to seek advice from alternate therapists.

DISCUSSION

This is one of only few reported cases of bilateral complete cryptophthalmos. In 2005, Egier and colleagues[2] reported the 6th case of bilateral complete cryptophthalmos[3].

To the best of our knowledge this is the 7th reported case of this condition in the world and the first where bilateral complete cryptophthalmos is associated with inguinal hernia beside other reported malformations such as anomaly of ears and genitalia and depressed nasal bridge.

The parents of the infant were young and belonged to low socioeconomic class with little education. They had a non-consanguious marriage and this was their first child. There was no family history of similar condition from both sides of the family. Sadly, the mother was blamed for the malformation. Such social implications have not been highlighted in other reported cases. The infant had not been screened for other possible systemic anomalies such as cardiac, pulmonary and renal.

An attempted lid construction revealed eyes with malformed anterior segment. There was micro-cornea with opacities and disorganized and poorly visible anterior chamber. The aim of the surgery was to visualize the eyes and assess their anatomical and functional status because ultrasonography revealed normal looking vitreous cavity with no retinal detachment.

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