Case Report

Isolated Bilateral Complete Cryptophthalmos

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Abstract

Cryptophthalmos is a rare congenital anomaly, characterized by extension of the skin continuously from forehead onto the cheeks and covering eyeballs. Although cryptophthalmos, as a part of Fraser syndrome, has been reported many times, isolated cryptophthalmos without systemic associations is very rare. We present a 6-month-old child with isolated bilateral complete cryptophthalmos, which to the best of our knowledge, is the first case of cryptophthalmos being reported from Vietnam.

Keywords: Cryptophalmos, congenital, eyeball

Introduction

Cryptophthalmos (CO) is a rare congenital condition in which eyelids are unable to divide in the embryo and the skin extends continuously from forehead onto the cheeks covering eyeballs. (Coulon et al 1994, Seal et al 992, Kanhere et al 1999) It is described only one and a half century ago, and appears with a frequency of 20 for every 100,000 newborns (González-Treviño et al 2008). CO is classified into three types: complete, incomplete and abortive (Kanhere et al 1999). Both autosomal recessive and autosomal dominant inheritance have been described, but most cases are autosomal recessive, with consanguinity being reported in 15% to 24.8% cases (Stevens et al 1994, Ramsing et al 1990, Schauer et al 1990, Berg et al 2001, Slavotinek et al 2002). CO is usually accompanied by urogenital anomalies, syndactyly, and cognitive disorders, and is termed as Fraser Syndrome(Stevens et al 1994, Ramsing et al 1990, Schauer et al 1990, Berg et al 2001, Slavotinek et al 2002). Very rarely, it's isolated without any syndromic associations (Kanhere et al 1999). We are reporting a case of isolated bilateral complete case of CO, which to the best of our knowledge is the first case of cryptophthalmos from Vietnam and only the 8th case of isolated bilateral complete Cryptophthalmos (without any systemic associations) ever reported (Chaudhary et al 2010).

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Case report

A 6 months old male child from Vietnam presented to Orbis Flying Eye Hospital Program with bilateral complete cryptophthalmos. He was the first child born to 25 years old mother. There is no history of consanguinity. The child was delivered by cesarean section at term.

There were no postnatal complications. Birth weight was 3kgs, with an Apgar score of 9 and no respiratory distress. The child had normal head circumference. On examination, his eyes were completely covered by skin with absence of brows, eye lids, lashes and palpebral aperture in both sides. (Figure 1) Spontaneous globe movements could be detected under the skin, with soft eyeball palpable bilaterally. The left globe appeared to be smaller in size compared to the right. The child had hypertelorism with depressed hypoplastic nasal bridge. There was no umbilical hernia or genito- urinary defects. There were no other congenital defects of fingers, ears (Figure 2), nose or larynx. MRI (Magnetic Rsonance Imaging) showed two eye balls with intact outer wall, with no lens seen in both globes. (Figure 3a) B Scan confirmed absence of the crystalline lens. (Figure 3b)

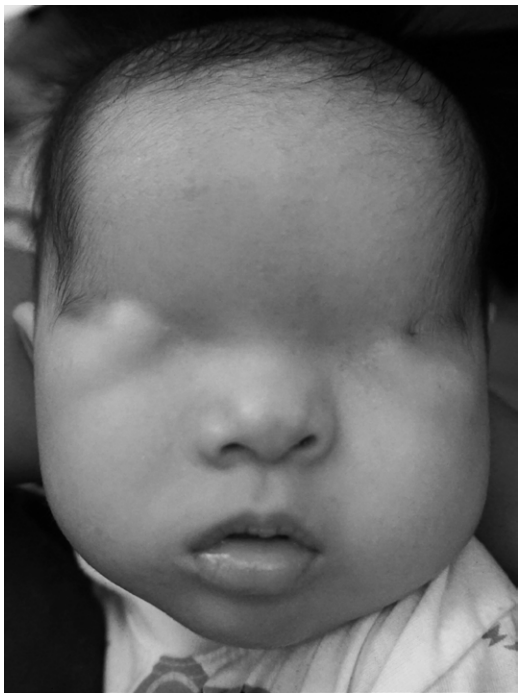

Figure 1: 6 month old child with bilateral complete cryptophthalmos

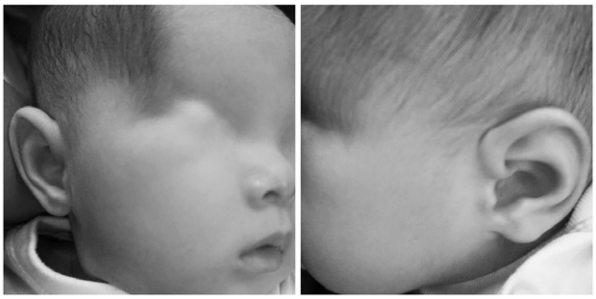

Figure 2: Right and left ear

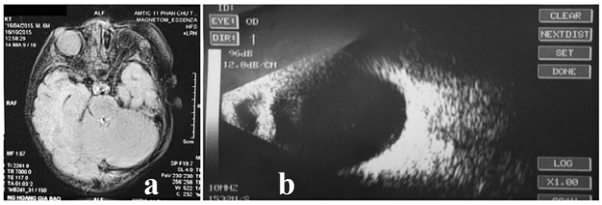

Figure 3: (a) Coronal MRI scan showing absence of crystalline lens (b) USG B-scan showing absence of crystalline lens.

Discussion

The association between cryptophthalmos and multiple congenital anomalies has been well described over the last century, with Thomas describing the diagnostic criteria for Fraser Syndrome (Stevens et al 1994, Ramsing et al 1990, Schauer et al 1990, Berg et al 2001). The pathogenesis of this condition is unknown, with some studies documenting similarities with animal models showing Vitamin A deficiencies and defects in programmed cell death (Thomas et al 1986). Prenatal diagnosis and subsequent management is still a challenge, especially in the developing world Good prenatal screening can help physicians with recognition of some of its characteristics through ultrasonography (USG) examination of the eyes, digits, kidney, and lungs in utero (Berg et al 2001). Bilateral orbital reconstruction with corneal graft and anterior vitrectomy and lid reconstruction has been described for such cases, but the visual deficit usually persists due to clouding of corneal grafts (Kanhere et al 1999, Chaudhary et al 2010). In our case we deferred surgery, due to extensive anterior segment digenesis as

evident on MRI and USG B Scan. Also, the surgery is a staged procedure which requires continued care over a long period of time. We did not plan surgery in this patient because of the lack of infrastructure and expertise in managing this case,

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