Cryptophthalmos: Another challenging problem in our practice

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Cryptophthalmos refers to the congenital absence of the eyelids with the skin passing continuously from the forehead onto the cheek over a rudimentary eye.

The deformity can be classified into three types: complete, incomplete and abortive and can be either isolated or syndromic.[1] The first documented description of a case was by a roman military commander called Pliny the elder at the year of 79 AD. In book VII of his Naturalis Historia he said "We have heard it stated that three children of a couple belonging to the clan of the Lepidi, have been born, although not in uninterrupted succession, with an eye covered with a membrane of skin" providing in one sentence a good description for the condition and its familial occurrence.

Going fast forward to 1872 Zehender and Manz used the term cryptophthalmos for the first time in their description of a six-month old girl. Their Patient also had an umbilical hernia, abnormal external genitalia, impaired anal sphincter and syndactyly so they were the first to suggest that the condition may not be an isolated but rather it is generalized malformation. Moving toward 1962, a genetist called George Robert Fraser concluded again after studying 4 cases of cryptophthalmos and multiple malformations that the disease is not an isolated malformation but part of a syndrome, characterized by four cardinal signs namely: Cryptophthalmos, Anomalies of head, nose and ears, Syndactyly and Genital Abnormalities.[2]

In 1986 Thomas et al. reviewed 124 cases and found that the principal syndromic form is Fraser syndrome. They reported 27 cases of isolated non-syndromic cryptophthalmos. Bilateral form is more common as only four cases have been reported to have unilateral complete cryptopthalmos and they are all in the left eye.[3]

Fraser syndrome is caused by genetic mutations of the FRAS1 gene (4q21) or the FREM2 gene (13q13.3). As a results of this mutations, cryptophthalmos develops secondary to failure in the separation of eyelids occur by a process of controlled necrosis of palperbral tissue between 17-18 weeks of gestation.[4]

A variety of ocular and periocular abnormalities have been described in association with cryptophthalmos is Fraser syndrome; tongue of hair extending across the lateral face (34.2%), absent eyebrows or eyelashes (29.1%) and coloboma of the eyelid (17.9%). Other abnormalities included microphthalmia (21.4%), anophthalmia (6.0%), and corneal opacification (10.3%).[5] However Bilateral congenital orbitopalpebral cysts have been only reported in two previous reports in English literature cyst.[6]

The management of patient with Fraser syndrome and cryptopthlmos requires a multidisplinary team that includes genetist, pediatrician, ophthalmologist and neurosurgeon.[7] Prior to any intervention, visual evoked potential has to be done keeping in mind that light perception is important for the maintenance of circadian rhythm and is better preserved whenever possible.Surgical reconstruction of the eyelid may work for partial form, but the results in complete has not been encouraging. Attempts to recreate the eyelid must take into consideration the four functional layers of the eyelid. Mucous membrane for lubrication and smooth blinking surface, Tarsus for framework, Orbicularis and retractors for blink function and the Skin over the external surface. When performing surgical reconstruction to cryptophthlmos eye surgeon has to take into consideration that the surface epithelium of the eyelid is continuous with the cornea. Surgeon has to be prepared to graft the cornea when there is penetration.[1] However, surgical correction becomes even more challenging in cases like bilateral orbitopalpleral cyst. The possibility of having a communicating nature of the cyst with the globe 'if the globe is not even appeared to be abortive and also the possibility of brain herniation into to orbit warrant a very careful surgical intervention.[6] If the surgery is ever to be done, a neurosurgeon has to be at the operating table just in case` .

References

1. Kanhere S, Phadke V, Mathew A, Irani SF. Cryptophthalmos. Indian J Pediatr. 1999;66(5):805-808.

2. George R. Fraser. Fraser Syndrome: Two millennia of cryptophthalmos from Pliny the Elder to FRAS, FREM and GRIP: A historical perspective. O J Genet. 2013;3(2):1-7.
3. Thomas IT, Frias JL, Felix V, Sanchez de Leon L, Hernandez RA, Jones MC. Isolated and syndromic cryptophthalmos. Am J Med Genet. 1986;25(1):85-98.
4. Harris MJ, McLeod MJ. Eyelid growth and fusion in fetal mice. Anat Embryol1982;164(2):207-20.
5. Slavotinek MA, Tifft CJ. Fraser syndrome and cryptophthalmos: review of the diagnostic vriteria and evidence of phenotypic modulea in complex malformation syndrome. J Med Genet. 2002;39(9):623-633.
6. Mocan MC, Ozgen B, Irkec M. Bilateral orbito-palpebral cyst in a case of cryptophthalmos associated with Fraser syndrome.J AAPOS. 2008;12(2):210-211.
7. Saleh GM, Hussain B, Verity DH, Collin JR. A surgical strategy for the correction of Fraser syndrome cryptophthalmos. Ophthalmology. 2009;116(9):1707-1712.