

FRASER SYNDROME- A RARE CASE REPORT AND REVIEW OF LITERATURE

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ABSTRACT

A rare case of Fraser syndrome¹ is being reported here in a 3 month old male child who presented to Ophthalmology department with cryptophthalmos (A rare congenital anomaly in which the skin is continuous over the eyeball with absence of eyelids), anophthalmos, craniofacial dysmorphism, pseudo-hypertelorism orofacial clefting, (cleft lip, cleft palate mental retardation. Based on the clinical profile, clinical diagnosis of Fraser syndrome was made. There are only few cases of Fraser syndrome reported in the literature. The case is being reported here for its rarity.

KEYWORDS: Fraser syndrome, cryptophthalmos, craniofacial dysmorphism, orofacial clefting, pseudo-hypertelorism, mental retardation.

A 3 month old male child born of non-consanguineous parents, presented to Ophthalmology department. Ophthalmological examination revealed cryptophthalmos (A rare congenital anomaly in which the skin is continuous over the eyeball with absence of eyelids), anophthalmos in left eye, craniofacial dysmorphism, pseudo-hypertelorism orofacial clefting, (cleft lip, cleft palate) mental retardation (Fig.1). Right eye on gross examination appears to be normal. Posterior segment in right eye including fundus (C/D ratio .6:1) appears normal. Child underwent complete ENT examination including hearing assessment and there were no signs of hearing impairment except cleft lip and cleft palate. Paediatric consultation was done to assess the mental development of child which reveals delayed all reflexes according to his age and mentally subnormal. Cardiovascular examination (Echocardiography revealed the presence of Partial anomalous pulmonary venous connection). Thyroid profile done on the seventh day of life was consistent with the diagnosis of congenital hypothyroidism [T4: 3.2 µg/L (normal: 4.8-11.6 µg/ L); TSH: 48.4 µIU/L (normal: 0.28-5.6 µIU/L)]. The mother's thyroid profile was normal. Dental examination didn't reveal any abnormality. There was no relevant family history and no history of maternal drug intake during pregnancy. The child was preterm normal vaginal

delivery with no perinatal complications. Radio diagnostic examination of Brain did not reveal any major abnormality as shown in figure 2.



Figure 1.

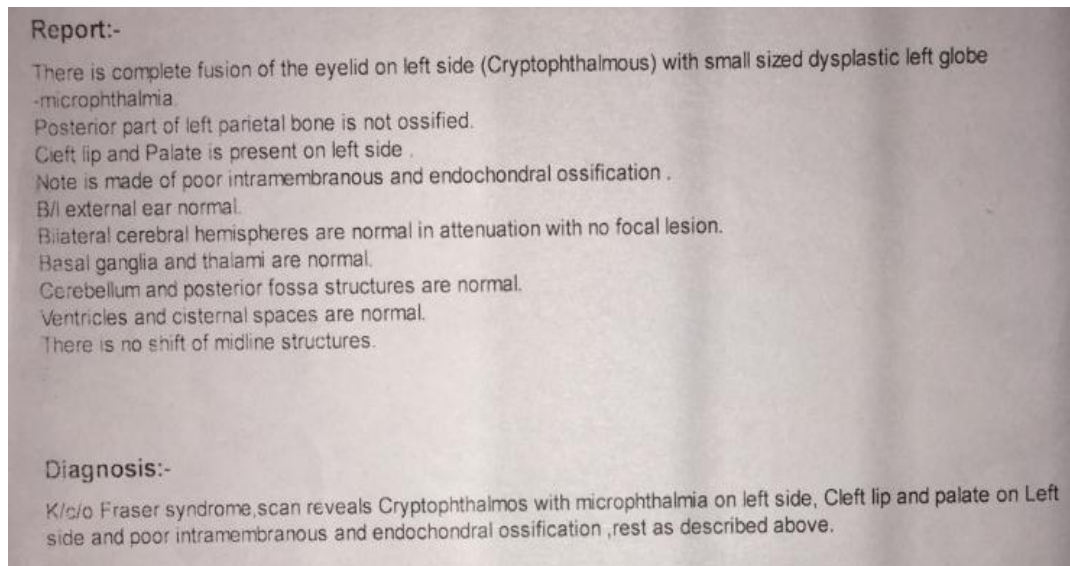


Figure 2.

DISCUSSION

Fraser syndrome comprises of cryptophthalmos with defects of the eyes, especially the anterior segment, combined with anomalies of the ears, nose, limbs, urogenital system and other areas. It is inherited in Autosomal recessive fashion and is caused by mutations in FRAS1 gene located on the long arm of chromosome 4 (4q21). Our patient satisfied the diagnostic criteria proposed by Thomas, et al.^[2] which requires at least two major and one minor, or one major and four minor criteria for the diagnosis. The major criteria include cryptophthalmos, syndactyly, abnormal genitalia, and sibling with Fraser syndrome; the minor ones are (i) congenital malformations of the nose, ear, larynx, (ii) cleft lip and/or palate, (iii) skeletal defects, (iv) umbilical hernia, (v) renal agenesis and (vi) mental retardation. The cardiac anomalies reported thus far in Fraser syndrome include hypertrophy of the left ventricle^[3] a variant of Ebstein anomaly, coarctation of the aorta^[4], an atrial septal defect^[5], an interventricular communication^[6], and a truncus arteriosus and a ventricular septal defect^[4]. In our case Echocardiography revealed the presence of partial anomalous pulmonary venous connection. We could not investigate the cause of congenital hypothyroidism in this child. It is possible that the observed finding may be incidental.

CONCLUSION

The diagnosis of Fraser syndrome can be made on clinical examination. As treatment options are limited, therefore genetic counseling plays an important role. When recommended, surgical treatment may be an appropriate option to correct some of the malformations associated with this disorder.

Conflict of Interest

[No financial interest or any conflict of interest.]

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