

Cryptophthalmos Syndrome: A Case Report

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A 20 days female baby presented to us in OPD. She was the 4th child of normal parents with 3 normal siblings. She exhibited few features of cryptophthalmos which fit the criteria of Fraser syndrome.

Key words: Cryptophthalmos, Fraser syndrome, Eye lid defect.

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Cryptophthalmos (CO) is defined as a set of rare congenital eyelid defects in which the lid folds are unable to divide in the embryo and the skin extends continuously from the forehead onto the cheeks covering the eyes.¹⁻³ CO maybe bilateral or unilateral and fluctuates in severity from the presence of rudimentary, distorted eyelids to complete absence of eyelids². Autosomal recessive and autosomal dominant inheritance have been reported, but most cases are autosomal recessive.⁴⁻⁸

CO is of three clinical types:

- Complete
- Incomplete
- Abortive^{2,3}

CO is termed as Cryptophthalmos Syndrome or Fraser Syndrome because in most cases it is usually associated with other malformations or systemic findings. However, it can also be isolated.²⁻⁸ Fraser Syndrome (FS) is a rare congenital autosomal recessive disorder,⁴⁻⁸ the prevalence of which is estimated to be 0.43 per 100,000 live births and 11.06 per 100,000 stillbirths⁹. The first case of FS as described by Zehender and Coworkers (1872).^{2,5,8,12}

We will be presenting a case of unilateral cryptophthalmos with Fraser Syndrome.

CASE REPORT

The patient is a 20 days old female. She is the 4th child of healthy parents. Her three older siblings are normal with no congenital malformation. The infant is full term and delivered by spontaneous vaginal delivery which was eventless. The baby weighed 2900 grams at birth and exhibits normal feeding manner.

Clinical evaluation exhibits complete absence of right side eyelid formation with absent eyelashes. The skin is continuous from the forehead to the cheek, covering the entire globe. The temporal hairline is abnormal growing down towards the cheek on the right side. A small palpable globe is felt beneath the skin and the orbit is shallow with a deficient orbital rim. There is complete Cryptophthalmos of the right side but the left eye is normal, with properly formed lids, lashes and eyeball (Fig. 1). Pupillary reflex, IOP and fundus examination are normal on the left side.

The nose has a wide nasal bridge but is flat. Hypertelorism is also present in the subject (Fig. 1). Distortion is present on the right of the face (Fig. 1).

Other findings that are noted includes:

- Mild brachydactyly of hands (Fig. 2).
- Cutaneous syndactyly of both feet (Fig. 3).
- External genitalia and ears are normal and the patient does not exhibit any other systemic malformations.



Fig. 1: Cryptophthalmos with Hypertelorism



Fig. 2: Mild Brachydactyly of Hands



Fig. 3: Cutaneous Syndactyly of Feet

DISCUSSION

The findings present in this case are consistent with the Fraser Syndrome (FS) according to the diagnostic criteria proposed by Thomas (1986) (Table 1).^{5, 7, 8, 10}

Table 1: Diagnostic Criteria for Cryptophthalmos Syndrome

<p>MAJOR CRITERIA</p> <ul style="list-style-type: none"> • Cryptophthalmos • Syndactyly • Abnormal Genitalia • Sibling with Cryptophthalmos Syndrome <p>MINOR CRITERIA</p> <ul style="list-style-type: none"> • Congenital Malformation of Nose • Congenital Malformation of Ears • Congenital Malformation of Larynx • Cleft lip and/or Palate • Skeletal defects • Umbilical Hernia • Renal Agenesis • Mental Retardation

For the diagnosis, following requirements must be met:

- 2 major and 1 minor criteria.
- 1 major and 4 minor criteria.^{5,7-9}

In this case study, diagnosis is made on the following basis:

- CO and syndactyly as major criteria.
- Malformations of the nose as minor criteria.

CO is the primary feature of FS and has been described in 84% to 93% of the patients. It should also be noted that CO isn't a regular finding in the syndrome.^{5,8} Our case has unilateral complete CO of the right eye.

Syndactyly is taken as a chief feature of FS that occurs in almost 77% of the patients. Syndactyly is always cutaneous and, in most cases, involves fingers and toes.^{5,8}

Genital anomalies in males are:

1. Micropenis.
2. Hypospadias.

3. Cryptorchidism.
4. Phimosis.

Genital anomalies in females are:

1. Clitoromegaly.
2. Bicornuate Uterus.
3. Uterine Hypoplasia.
4. Vaginal Agenesis.
5. Synechiae or hypoplasia of the labia.^{5,8}

In this case study, cutaneous syndactyly of the toes is present, but the genitalia are completely normal.

Kinship is reported in 15 – 24.8% of the cases and an autosomal recessive pattern of inheritance is evident^{5,8}. The parents of the patient, on this case study, are not related. There is 25% recurrence risk of this syndrome, among siblings⁵. FS should be suspected in all cases of stillbirths with renal agenesis. 25% of affected fetuses are stillborn¹⁰.

Currently, prenatal diagnosis of FS by an expert is possible with recognition of some of its characteristics through ultrasonography (USG) examination of the eyes, digits, kidney, and lungs in utero.^{7,11} Therefore; USG is recommended in following patients (babies) with higher chances of Fraser Syndrome:

- Blood related parents
- Families with a previously affected child
- Cases of stillbirths with renal agenesis

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