

Case Report

Sudanese infant with Fraser syndrome

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Abstract:

We are reporting here a case of Fraser syndrome which is a very rare autosomal recessive disorder characterized by cryptophthalmos, cutaneous syndactyly, craniofacial dysmorphism, malformation of the upper airways and genitourinary tract. Diagnosis of Fraser syndrome is based on clinical signs.

Key words: Fraser syndrome (FS), cryptophthalmos (CO), syndactyly

Fraser syndrome (FS) is a rare autosomal recessive disorder (Figure 1).

Cryptophthalmos (CO), or "hidden eye" is a term invented by Zehender and Manz in 1872¹. Since their original description till 1995, approximately 149 cases of complete or partial cryptophthalmos have appeared in the world English literature². Fraser³ defined the first report of autosomal recessive cryptophthalmos syndrome, while Francois⁴ observed the other main features of hand, orofacial and genital abnormalities. There are more than 200 published case reports of patients with CO and FS and several comprehensive reviews have previously been published².

Case report:

This was a 35 days female, a second baby of consanguineous healthy parents; father 24 years and mother 25 years.

She was an outcome of a normal vaginal delivery following full term uncomplicated pregnancy. Mother was not on regular antenatal care. She cried immediately after birth, started feeding and passed urine and meconium during the first 24 hours. The midwife noticed the complete fusion of the baby eyelids and abnormally fused fingers (figure 2). She advised the parents to seek medical advice. There was family history of similar conditions; two of her paternal cousins

had similar clinical abnormalities and died immediately or after few days of birth due to severe respiratory distress

She was seen initially in ophthalmology clinic and then was referred to Omdurman Pediatrics Emergency Hospital.

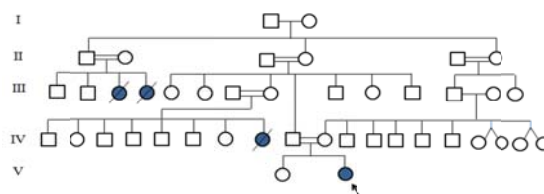


Figure (1) the family pedigree

There was no history of cough, cyanosis or difficulty during feeding. There were no change in bowel habits, no vomiting or abdominal distension. The urine was normal in amount, color and frequency. There patient can move her limbs normally, no weakness or abnormal movements,



Figure 2: bilateral cyptophthalmos

On examination her weight was 4.3kg, crown heel length was 55 cm and head circumference was 37cm. The craniofacial

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examination showed prominent forehead, opened anterior fontanelle 7x4cm, depressed nasal bridge, broad nose with midline nasal groove, bilateral CO with presence of both eyeballs and absence of eye brows and lashes (figure2). Both ears were small and low sited (figure 3). She had cutaneous syndactyly in both hands and feet and bilateral brachydactyly of the big toe (figure4: a & b). Genitalia showed clitoromegaly with hypoplasia of the labia and she had low set umbilicus (figure 5). Ultrasound for abdomen showed bilateral dysplastic kidneys with normal internal female genital organs. Echo cardiography showed normal heart structure and function.

These clinical findings were consistent with the diagnosis of Fraser's syndrome.



Figure 3: low set ear, depressed nasal bridge

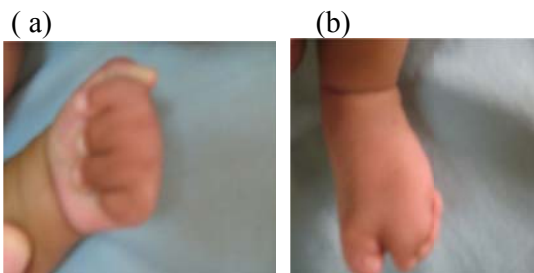


Figure 4: syndactyly

Discussion:

The diagnostic criteria for Fraser syndrome are classified to major and minor criteria. The major criteria are: cryptophthalmos, syndactyly, abnormal genitalia and sibling with FS; while the minor criteria are: congenital malformation of nose, ears, larynx, cleft lip and/or palate, skeletal defects, umbilical hernia, renal agenesis and mental

retardation. The criteria to distinguish isolated CO from CO with other malformations and FS were provided by Thomas et al 1986 following a study of 124 cases of CO⁵.

Two major criteria and one minor criterion or one major and at least four minor criteria were required for the diagnosis of FS⁵. Our patient had two major criteria (cryptophthalmos and syndactyly) and two minor features (clitromegaly and renal agenesis). From the 117 cases reported²; females were 48.7%; consanguinity was present in 24.8% of cases; and the most common consanguineous union was first cousins; 41% of cases had a significant family history of a relative with CO or physical findings suggestive of FS. Consanguinity marriage and family history were evident in our case. The average paternal age was 27 years of age and the average maternal age was 24 years of age which is similar to ages of our patient's parents.



Figure 5: clitromegaly, low set umbilicus

Slavotinek and Tiff² also considered additional phenotypes associated with FS including other ocular abnormalities (absent eyebrows or eyelashes, coloboma of the eyelid, microphthalmia, anophthalmia and corneal opacification), hand abnormalities (brachydactyly, nail hypoplasia and abnormal palm creases). Clitromegaly in females and cryptorchidism in males were the commonest. Our patient had clitromegaly and abnormal hands.

Cardiac malformations include hypertrophy of the left ventricle, coarctation of the aorta, atrial septal defect, truncus arteriosus and ventricular septal defect. Dextrocardia and

transposition of the great vessels were also noted, none of these were seen in this case.

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