Göztepe Tıp Dergisi 29(1):60-61, 2014
doi:10.5222/J.GOZTEPETRH.2014.060
ISSN 1300-526X

OLGU SUNUMU *Genetik*

Fraser syndrome: A new case

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SUMMARY

Fraser syndrome is characterized by cryptophthalmos, cutaneous syndactyly, malformations of the larynx and genitourinary tract, craniofacial dysmorphism, orofacial clefting, mental retardation, and musculoskeletal anomalies. Fraser syndrome is a rare, autosomal recessive condition. Herein, we report a case of a two day old infant with Fraser syndrome.

Key words: Fraser syndrome, cryptophthalmia, infant

ÖZET

Fraser sendromu: Yeni bir olgu

Fraser sendromu kriptoftalmus, kutanöz sindaktili, larinks ve genitouriner bölge malformasyonları, kraniyofasiyal dismorfizm, orofasiyal yarık, mental gerilik ve kas-iskelet sistemi anomalileri ile karakterizedir. Fraser sendromu nadir, otozomal resesif bir durumdur. Bu belgede, Fraser sendromlu iki günlük bir yeni doğanı rapor ettik.

Anahtar kelimeleri: *Fraser sendromu, kriptoftalmi, yenidoğan*

Fraser syndrome is a rare, autosomal recessive syndrome characterized by cryptophthalmos (hidden eye), syndactyly, ambiguous genitalia, hypertelorism, a broad depressed nasal bridge, ear anomalies, umbilical hernia, anal stenosis and diastasis of the symphysis pubis [1,2]. Here, we report a male infant with Fraser syndrome.

CASE

A two-day-old male term neonate born to third degree consanguineous parents by normal delivery. On examination at birth, he weighed 2.8 kg and measured 50 cm in length. He had bilateral cryptophtalmos, hypertelorism, a broad depressed nasal bridge, long philtrum and hirsutism. Both ears were low set, and dysplastic (Fig. 1). His echocardiography had demonstrated tetralogy of Fallot, and patent ductus arteriosus. The genitalia and anal opening were normal. However, ultrasound examination of the abdomen revealed right ureteropelvic junction obstruction. His chest radiogram was unremarkable. Karyotype analysis was 46,XY.

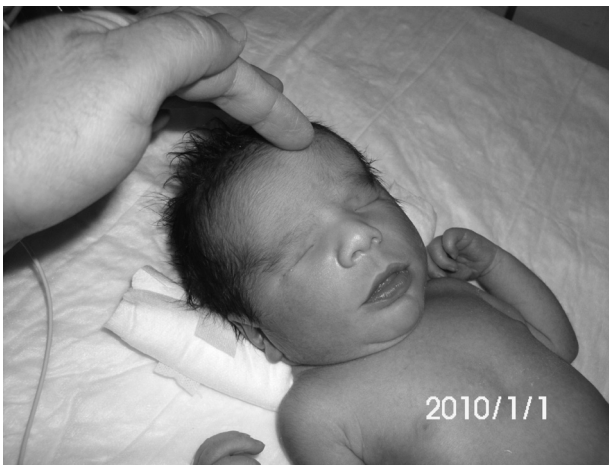

Figure 1. Fraser syndrome.

DISCUSSION

The present report describes a case of Fraser syndrome [3]. Fraser syndrome comprises of cryptophthalmos with defects of the eyes, especially in the anterior segment, combined with anomalies of ears, nose, limbs, urogenital system and other anatomical regions. It is inherited in autosomal reces-

Geliş tarihi: 23.05.2013
Kabul tarihi: 17.12.2013
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sive fashion and is caused by mutations in FRAS1 gene located on the long arm of the 4. chromosome (4q21) [1].

Diagnostic criteria of Fraser syndrome proposed by Thomas et al. [4] which require 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 congenital malformations of the nose, ear, larynx, cleft lip and/or palate, skeletal defects, umbilical hernia, renal agenesis and mental retardation. Furthermore, occasional central nervous system abnormalities in Fraser syndrome include microcephaly, hydrocephalus, encephalocele, abnormal gyral pattern, and meningomyelocele [1,5-8].

Our case had cryptophtalmos, dysplastic nose, long philtrum, hirsutism, tetralogy of Fallot , patent ductus arteriosus, and ureteropelvic junction obstruction. Death of the patient with Fraser syndrome is related primarily to the renal or laryngeal defects [1].

The present report describes the Fraser syndrome. The prognosis is dependent on the severity of the associated defects. Prenatal diagnosis by means of ultrasonography and fetoscopy is possible.

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