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A RARE CASE OF FRASER SYNDROME

Paediatric Medicine

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ABSTRACT

Fraser syndrome (cryptophthalmos-syndactyly syndrome) is a rare autosomal recessive malformation disorder. Diagnosis is based on criteria established by van Haelst et al. in 2007 with no specific treatment available and carries poor prognosis. Here we present a case of full term newborn born with Fraser syndrome.

KEYWORDS

Fraser syndrome, autosomal recessive disorder, cryptophthalmos-syndactyly syndrome

INTRODUCTION

Fraser syndrome (cryptophthalmos syndactyly syndrome) is a rare autosomal recessive malformation disorder. The incidence of Fraser syndrome is 0.43 in 10,000 or 1 in 200,000 new-borns and 11.06 in 10,000 babies that die before birth (stillbirths). The first description of the syndrome was reported by George Fraser in 1962.

Case Study

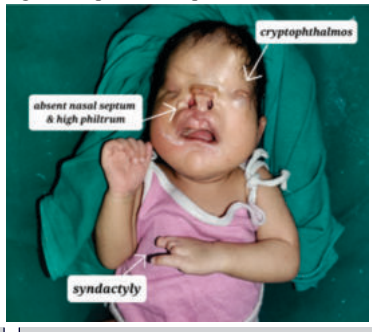
A full-term female neonate born at 37 weeks of gestation by spontaneous normal vaginal delivery, out of 2nd degree consanguineous marriage with birth weight 2.4 kg, appropriate for gestational age, cried immediately after birth to 27 years old 3rd gravida mother. On examination bilateral anophthalmia, absent nasal septum and alar cartilage, wide set eyes, low set ears, micrognathia, retrognathia, high philtrum, left CTEV, left syndactyly was present.

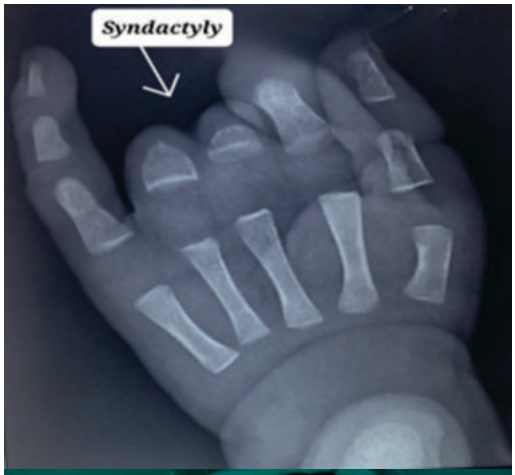
Sibling history: G3P3A0L3. With elder two siblings do not have any similar defects. According to clinical criteria major criteria fulfilled are syndactyly and cryptophthalmos and minor criteria fulfilled are skull ossification defects and nasal anomalies. Patient was diagnosed with Fraser syndrome. Patient expired at 18 days of life before genetic workup.

DISCUSSION

Fraser syndrome affects males and females in equal numbers. Diagnosis is based on the major and minor criteria established by van Haelst et al. in 2007. Major criteria are Syndactyly, Cryptophthalmos spectrum, Urinary tract abnormalities, Ambiguous genitalia, Laryngeal and tracheal anomalies, Positive family history. Minor criteria are Anorectal defects, Dysplastic ears, Skull ossification defects, Umbilical abnormalities, Nasal anomalies. Three Major or Two Major+Two Minor or One Major+Three Minor criteria should be met for the diagnosis of Fraser syndrome.

The syndrome is related to mutations in three different genes (FRAS1, FREM2, and GRIP1) resulting in failure of the apoptosis program and disruption of the epithelial-mesenchymal interactions during embryonic development. There is no specific treatment for the syndrome. Prognosis is poor and depends on the dominant pathology.





CONCLUSIONS

Since Fraser syndrome is an autosomal recessive in inheritance there are 25% chance of having a child with Fraser syndrome again. Genetic counselling of patient's family was done.

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