Case Reports

Fraser syndrome with bladder pseudoexstrophy

Jehad A. Daia, MD.

ABSTRACT

We report a one year old girl with Fraser Syndrome (the association of craniofacial abnormalities, syndactaly and cryptophlamos) and multiple urogenital abnormalities including clitorimegaly, left renal agenesis and a unique urinary bladder exstrophy variant (psuedoexstrophy) with intact bladder which herniates through the lower abdominal wall defect.

Keywords: Fraser syndrome, pseudoextrophy, urological abnormality.

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The association of craniofacial abnormalities, syndactaly, cryptophtalmos was first described by Fraser in 1962.[1] Inheritance in Fraser Syndrome is by an autosomal recessive mode. Urogenital abnormalities such as renal agenesis or hypoplasia and enlarged clitoris are a common occurrence.[2,3] Pseudoextrophy; bladder extrophy variant was described as a rare isolated abnormality of the urinary bladder. To the best of our knowledge herein we describe a unique case of Fraser Syndrome with bladder pseudoextrophy.

Case Report. A one year old girl with Fraser syndrome had the typical abnormalities associated with the syndrome i.e. cryptophthalmos and depressed broad nasal bridge, syndactyly of both hands and feet and laryngeal stenosis Figure 1. Also she had multiple urogenital abnormalities including clitorimegaly, left renal agenesis and abdominal wall/ skeletal abnormalities similar to what usually seen in bladder extrophy – but with an intact bladder (psuedoextrophy). These included lower abdominal wall defect through which the bladder was herniated, a low set umbilicus and separated pubic bones (Figure 2). The lower abdominal wall defect was corrected surgically by simple dissection and approximation with minimal tension.

Discussion. According to the diagnostic criteria proposed by Thomas et al,[4] the findings in this patient are compatable with the diagnosis of Fraser Syndrome. The major criteria are cryptophthalmos, syndactyly, abnormal genitalia and sibling with cryptophthalmos syndrome, and the minor criteria are congeital malformation of ears, nose, larynx, cleft lip, skeletal defect, umbilical hernia, renal agenesis and mental retardation in survivors. In this report we have a one year old girl with most of the major and minor criteria of Fraser Syndrome. The findings of a law set umbilicus, separated pubic symphysis and an intact urinary bladder that herniated through the lower abdominal wall defect is considered to be a bladder extrophy variant which is best described as pseudoexstrophy. Bladder extrophy is considered to be secondary to failure of cephaled movement of the body stalk, rendering the midline mesenchymal interposition impossible and causing subsequent rupture of the superficial coloacal membrane. It would appear that in the present case the first embryological event took place (to produce the abdominal wall defect) without rupture of the

From the Department of Urology, King Faisal Specialist Hospital and Research Centre, Riyadh, Kingdom of Saudi Arabia.

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Address correspondence and reprint request to: Dr. Jehad A. Daia, Pediatric Division, Department of Urology, King Faisal Specialist Hospital & Research Centre, MBC 83, PO Box 3354, Riyadh 11211, Kingdom of Saudi Arabia. Tel. +966 (1) 464 7272 Fax. +966 (1) 441 4839.

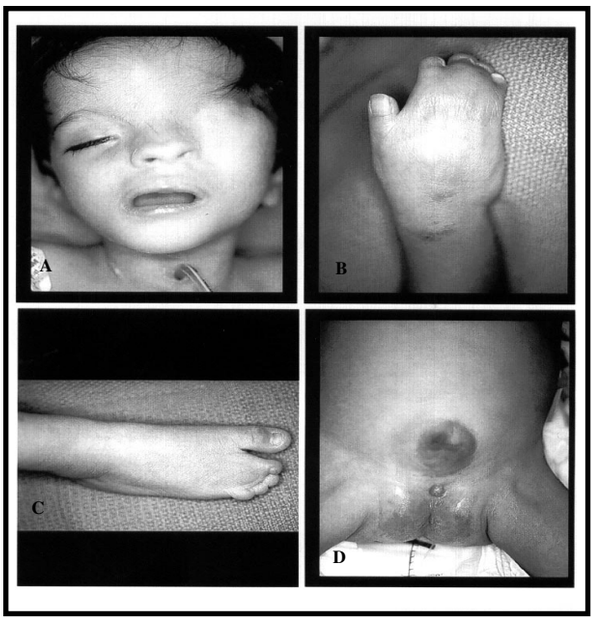

Figure 1 - Clinical photograph showing: (a) Cryptophthalmos and tracheostomy (for laryngeal stenois); (b,c) Syndactyly of hand and foot; (d) Lower abdominal hernia, a low set umbilicus and clitorimegaly.

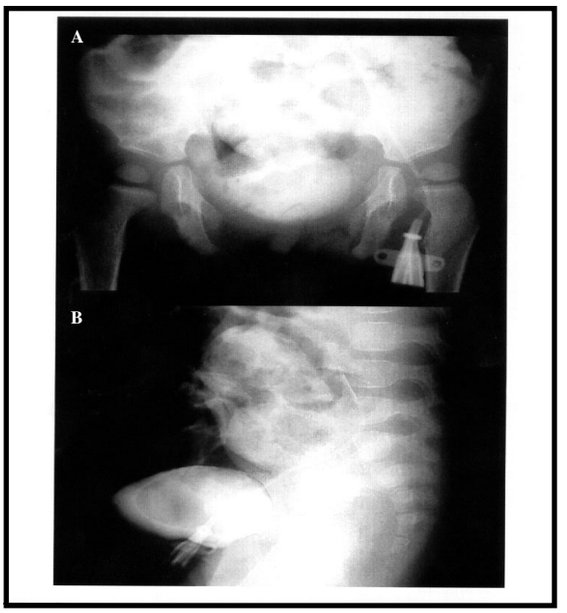

Figure 2 - Radiological photograph showing: (a) Plain x-ray showing wide separation of pubic bones; (b) Cystogram showing urinary bladder herniating through the lower abdominal wall defect.

coloacal membrane (to produce the exstrophy).[5] The variant of an exstrophic abdominal wall with normal bladder with or without association with Fraser Syndrome is unique. To the best of our knowledge this is the first case of bladder pseudoextrophy reported in association with Fraser Syndrome. In Saudi population due to the high consanquinity rate, we believe that Fraser Syndrome does exist but under reported.

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