

## Female Counterpart of Shawl Scrotum in Aarskog-Scott Syndrome

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### ABSTRACT

Aarskog-Scott syndrome (ASS) is an X-linked disorder characterized by facial, skeletal and genital anomalies, including penoscrotal transposition in males. We report on a girl from a family with ASS who exhibits a transposition of the clitoris.

**Key words:** urogenital abnormalities; FGD1 protein, human; X-linked inheritance; Aarskog-Scott syndrome  
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### INTRODUCTION

Aarskog-Scott syndrome (ASS) is an X-linked disorder caused by mutations in the FGD1 gene (Xp11.21) and characterized by facial, skeletal and genital anomalies (OMIM 305400). The main features are short stature, ocular hypertelorism, brachydactyly, and penoscrotal transposition (shawl scrotum) in males (1). Female carriers often show some minor manifestations of the disorder, especially in the face and hands. Penoscrotal transposition is a rare abnormality of the external genitalia in which the scrotum is malpositioned superior to the penis. As far as we are concerned, there are no previous reports on a similar female genital anomaly.

### CASE REPORT

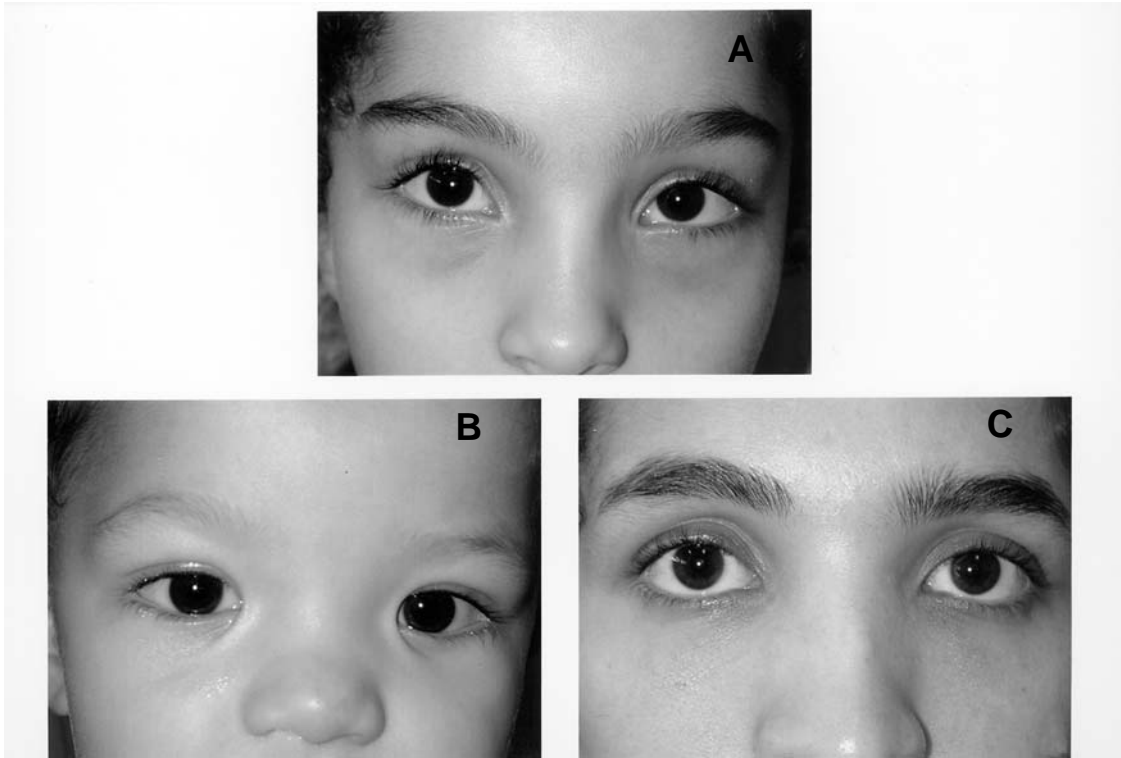
The patient, a 5-year-old girl, was evaluated after the diagnosis of ASS in her 17-day-old brother,

who exhibited micropenis. She was born to an 18-year-old woman and her 21-year-old unrelated husband, and her mother complaint that the girl had no clitoris.

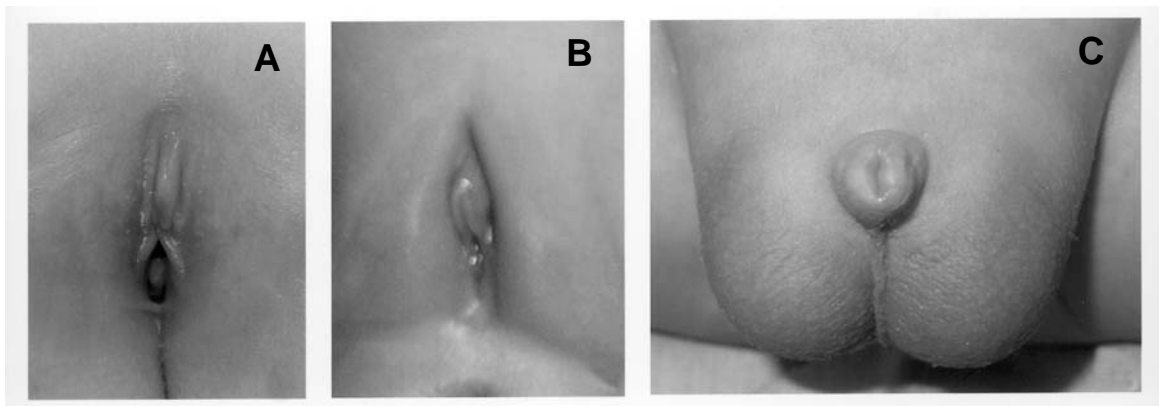
She exhibited ocular hypertelorism (Figure-1), up-slanting palpebral fissures, malar hypoplasia, anteverted nostrils, slight retrognathia, clinodactyly of the fifth fingers and joint hyperextensibility. Genital examination showed labia minora adhesions, and the clitoris was not located in its normal position, i.e., posterior to the anterior labial commissure. Instead, it emerged about 1 cm below (Figure-2).

Her brother had ocular hypertelorism (Figure-1), inner epicanthal folds, prominent ears, malar hypoplasia, small nose, broad nasal bridge, retrognathia, micropenis, bilaterally descended testes, hydrocele and partial penoscrotal transposition (Figure-2), the anus was normally placed.

Physical examination of her mother revealed ocular hypertelorism (Figure-1), malar hypoplasia, broad nasal bridge and normal female external genitalia.



**Figure 1** – Faces showing ocular hypertelorism. A) Index case; B) Brother; C) Mother.



**Figure 2** – A) Abnormal location of the genital tubercle of the index case; B) Normal female external genitalia; C) Partial penoscrotal transposition and micropenis of the patient's brother.

## COMMENTS

The etiology of penoscrotal transposition remains uncertain (2). Embryological origin of the penis and scrotum are respectively the genital tubercle and labioscrotal folds. At the end of the sixth week of

development, males and females have indistinguishable external genitalia. The penis and scrotum achieve their usual arrangement when, under the influence of androgens, the genital tubercle elongates to become the penis, while migration of labioscrotal folds brings

the latter to a caudal and dorsal position to the penis, where they fuse in the midline (3).

Abnormal location of the genital tubercle or abnormal migration of labioscrotal folds may be the origin of penoscrotal transposition, and may also lead to the abnormal location of the clitoris in the present case. The absence of similar reported cases may derive from its rarity, or this may be an under diagnosed feature of ASS. An answer to this question depends on routine evaluation of the external genitalia of female carriers.

### CONFLICT OF INTEREST

None declared.

### REFERENCES

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