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Case Report

Surgery Section

# Hemiscrotal Agenesis: A Rare Congenital Anomaly

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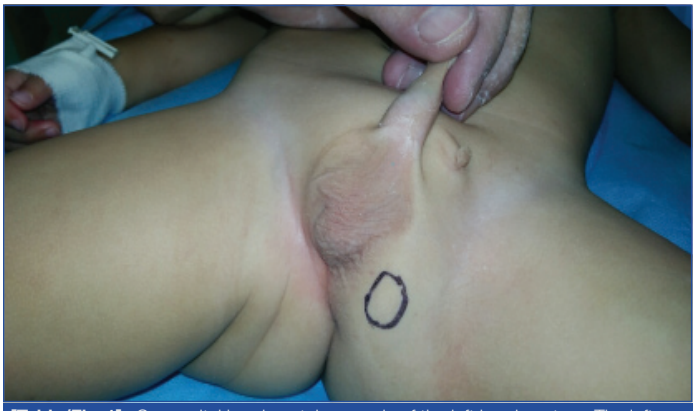
## ABSTRACT

Hemiscrotal Agenesis (HSA) is the rarest developmental malformation of the scrotum. It is characterized by the absence of either half of the scrotal rugae with an intact midline raphe. We report the case of a 16-month-old boy with HSA, with an island of scrotal tissue in the pubic tubercle region and ipsilateral cryptorchidism. To our knowledge, this is the first case of HSA with heterotopic development of scrotal tissue.

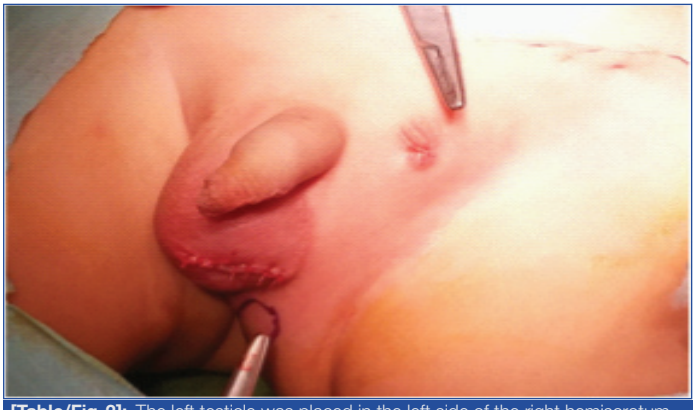
**Keywords:** Cryptorchidism, Perineum, Scrotum

## CASE REPORT

A 16-month-old boy was referred to the Department of Paediatric Surgery of the Aghia Sophia Children's Hospital, in Athens, Greece, because of unilateral cryptorchidism. The physical examination revealed a flat left perineal skin with absence of the ipsilateral rugae tissue, while the median raphe was normal [Table/Fig-1]. At the same time, an island of macroscopically looking scrotal tissue, which was inserted into the skin after the traction of the left gubernaculum and testis, measuring approximately 1 cm x 1 cm was observed at the left pubic tubercle region [Table/Fig-1]. The left testicle was palpable under the subcutaneous tissue in the midline of the perineum, between the scrotum and the anus [Table/Fig-1]. The right testicle was located in its normal anatomic position, in the right hemiscrotum and penile anatomy was normal. Also, obtaining our patient's medical history revealed unilateral polydactyly (six toes on his left foot), mild tricuspid insufficiency and performance of pyloromyotomy for repair of hypertrophic pyloric stenosis. The child subjected to surgical procedure through a combined inguinal and scrotal incision and the left testicle was placed in the left side of the right hemiscrotum [Table/Fig-2]. The scrotal tissue island was resected both for cosmetic reasons and the risk of malignant transformation and it was sent for pathologic examination, which revealed that it consisted of fibrous connective tissue and muscular fibers and it was surrounded by skin.



**[Table/Fig-1]:** Congenital hemiscrotal agenesis of the left hemiscrotum. The left testicle is palpable in the perineum. A distinct island of scrotal tissue is located in the pubic tubercle region.



**[Table/Fig-2]:** The left testicle was placed in the left side of the right hemiscrotum. The scrotal tissue island was resected.

## DISCUSSION

Congenital anomalies of the scrotum are uncommon. Scrotal disorders including bifid scrotum, scrotal ectopia and penoscrotal transposition are far less common [1], while complete Congenital Scrotal Agenesis (CSA) has only been described in six cases [1-6] and HSA in three [7-9]. The rarity of this congenital disorder does not allow for statistical analysis of the cases that have been reported so far. Furthermore, the fact that scrotum agenesis may either present as an isolated anomaly or be associated with a variety of other congenital defects such us face anomalies, growth retardation, cardiac anomalies, anterior ectopic anus, digit anomalies, undescended testis and intersex anomalies [3,4,7,8], makes it difficult to determine its pathophysiology.

During the foetal development the labioscrotal folds are formed during the seventh week of gestation on either side of the urogenital folds. Embryologically, the scrotum development occurs between the 10$^{th}$ and 16$^{th}$ week of gestation, when the labioscrotal folds move caudally, merge in the midline and enlarge, under the influence of androgens. Hence, most theories regarding CSA and HSA involve either mechanical or endocrine disorders that prevent the labioscrotal folds from merging together in the midline and differentiating to form the scrotum. Janoff DM et al., suggested that CSA may be caused by failure of the development of the labioscrotal folds [1], while Mohan PP et al., suggested it may be caused by localised deficiency of 5a-reductase type 2 [6].

Our case is the first one that has been reported with a distinct island of scrotal tissue located at an ectopic position in the pubic tubercle region. Furthermore, to our knowledge, it is the first case of HSA with a macroscopically normal testicle located under the subcutaneous tissue of the perineum, between the scrotum and the anus. It is our belief that HSA is a congenital anomaly of multifactorial etiology. The distinct scrotal tissue island might indicate partial regression of the left labioscrotal fold. The remaining tissue possibly differentiated under the influence of androgens to scrotal rugae tissue. Alternatively, local lack of sensitivity to androgens may have resulted in partial differentiation to scrotal tissue.

## CONCLUSION

HSA is an extremely rare congenital developmental anomaly of the scrotum. We have reported a case of HSA with a distinct island of

scrotal tissue situated at the inguinal area with the ipsilateral testicle located at the perineum.

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