PENOSCROTAL TRANSPOSITION: A CADAVERIC REVIEW

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ABSTRACT

Introduction: With only 20 documented cases, the congenital malformation known as complete extreme penoscrotal transposition with an intact scrotum, midline raphe, and absence of hypospadias is an extremely rare abnormality. Penoscrotal transposition occurs when the scrotum fuses above the penis for a complete abnormality or does not fuse at all for an incomplete abnormality. This failure of the scrotum to descend below the penis occurs during development, when the genital swellings fail to descend below the penis and fuse. The irregularity has been known to follow an X-linked recessive pattern of inheritance in some cases. In others it results from a defect of chromosome 13. This disease would have significant clinical implications for a patient in day to day activities, as well as sexually. However, little is known as to whether this transposition impacts spermatogenesis. Method: Dissection and observations were made from a 72-year-old male as part of a gross anatomy course at Philadelphia College of Osteopathic Medicine, utilizing the methods of the dissection manual. Biopsy of the testes were taken and without further fixation processed for light microscopy, using standard techniques. Results: Upon dissection, the individual exhibited a complete extreme penoscrotal transposition, with the scrotum fused above the penis in a shawl fashion, with no hypospadias. The tunica albuginea was thickened, fused in the midline, and adhered to the testis. Histology of the testis supports that spermatogenesis was not altered. The organization of the seminiferous tubules and developing germ cells appeared to be normal. Discussion: Along with the penoscrotal transposition, many abnormalities are typically also present. It was observed that the patient had several hernias, mega colon, and many abdominal adhesions. One interesting anomaly observed during dissection was bilateral fused adductor magnus/adductor longus muscles. As a senior computer analyst, this individual likely had no cognitive or mental impairment, however, his cause of death was due to end stage Parkinson's disease. Living with this disease would have been very difficult because the patient has virtually no protection of the testis as they are positioned superior to the penis. It would have had significant psychosocial implications for the patient, especially with regard to reproduction and sexual intercourse.

INTRODUCTION

With only 20 documented cases, the congenital malformation known as complete extreme penoscrotal transposition with an intact scrotum, midline raphe, and absence of hypospadias is an extremely rare abnormality. Penoscrotal transposition occurs when the scrotum fuses above the penis for a complete abnormality or does not fuse at all for an incomplete abnormality. This failure of the scrotum to descend below the penis occurs during development when the genital swellings fail to descend below the penis and fuse. Normally, the genital tubercle and the labioscrotal swellings give rise to the penis and the scrotum. The labioscrotal swelling migrates dorsally (caudally) and fuses inferior to the developing penis. This process is regulated by the presence of androgens. However, in individuals with penoscrotal transposition, the labioscrotal swelling and genital tubercle did not respond appropriately to androgens. The labioscrotal swelling failed to fuse and migrate to the appropriate target area. Previous studies of individuals with penoscrotal transposition indicate there are genetic causes. DNA sequence of several individuals found mutations of the sequence for androgen receptors. These genes are located on the X-chromosome. Also, several men were found to have mutations on chromosome 13. However, it remains unclear how these mutated genes result in penoscrotal transposition. In addition to the altered position of the genitalia, these men often have other anomalies including: musculoskeletal (e.g., scholiosis, hypoplastic thumb or radius); gastrointestinal (e.g., imperforate anus); cardiac conditions (e.g., patent foramen ovale, mitral valve prolapse) and disorders of the central nervous system (e.g., copus callosum agenesis, lateral ventricle dilation).[1] The presence of extreme penile scrotal transposition provided a unique opportunity to examine the anatomy of numerous systems and educate healthcare workers on this condition.

METHOD

The body of a 72 year-old male was donated to the Humanity Gifts Registry of Pennsylvania. This is the anatomical board for the state of Pennsylvania that was established in 1893 by the Anatomical Gift Act of Pennsylvania. Age and cause of death and occupation were obtained from the death certificate. The body was used in fall gross anatomy courses at Philadelphia College of Osteopathic Medicine. Dissection of the abdomen and perineum was conducted with modifications following the procedures in Grant's Dissector (Tank, 15th ed.). A biopsy of the testis was taken and processed for light microscopy using standard techniques.

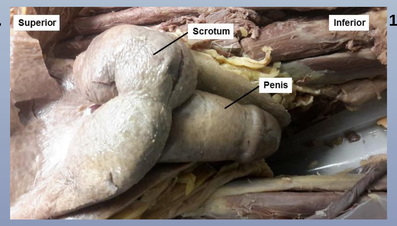
RESULTS

General observations:
- **72 year old male; approximately 5'8" tall weighing proximately 170 lbs.**
- **Occupation: computer analyst.**
- **Cause of death listed as Parkinson's Disease**
- **Examination of the heart indicated the likelihood of an inferior wall myocardial infarction.**
- **The patient had several abdominal scares along the linea alba.**
- **His abdominal surgeries resulted in incisional hernias.**
- **He had a prior cholecystectomy.**
- **Megacolon beginning at the distal end of the sigmoid colon.**
- **Urinary bladder located superior to the pelvic bone.**

Perineum (Figures 1 and 2):
- **Scrotum located above the penis.**
- **Scrotum had no direct connection to penis.**
- **The testes were fused to the scrotal wall by a thickened tunica albuginea.**
- **The thickened tunica in the midline was fused and joined both testes.**
- **The spermatic cord was located in the inguinal canal.**
- **The pampiniform plexus of veins was replaced by a singular testicular vein.**
- **Normal erectile tissue present in the penis.**
- **Histological examination of the testes revealed normal appearance of seminiferous tubules containing Sertoli cells, spermatogonia, spermatocytes and spermatids (Figure 3).**

Thigh:
- **Adductor magnus and adductor longus were fused bilaterally.**
- **Muscle fibers for this fused muscle were in a uniform direction**

1A.



1B.

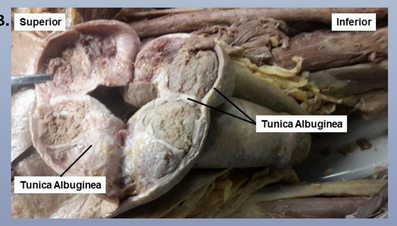


Figure 1. The scrotum is present on the anterior abdominal wall superior to the penis (A). Testes are present within the scrotum. The tunica albuginea is thicker than normal, fused in the midline and adheres to the scrotal wall (B).

2.

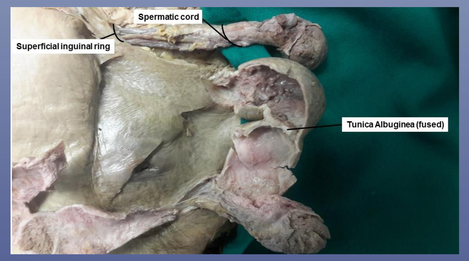


Figure 2. The testes were removed from the scrotum and spermatic cord was defined. The spermatic cord runs through the inguinal canal.

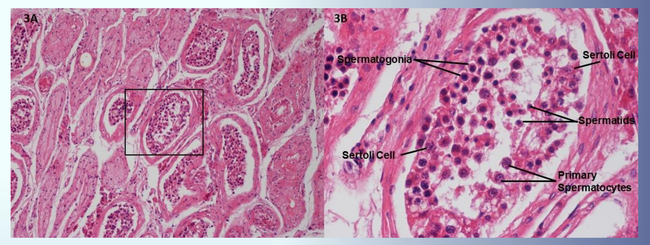


Figure 3. Light micrograph of testis containing seminiferous tubules (A). Sertoli cells, spermatogonia, spermatocytes and spermatids were present in the seminiferous tubules (B).

DISCUSSION AND CONCLUSION

The presence of penoscrotal transposition provided a unique opportunity to examine and describe the anatomy of the perineum as well as other regions. Family history for this individual was not available. The normal subpubic location of the scrotum protects the testes from physical trauma. In addition, the cremaster muscles can raise or lower the temperature to optimize spermatogenesis. The position of the scrotum on the abdominal wall may have altered temperature regulation of the testis in this person. Based on the presence of spermatogenic cells in the testes and erectile tissue in the penis, it is likely that he was fertile. Musculoskeletal anomalies such as scoliosis and clinodactyly have been found in the upper limbs of some men.1 This is the first reported case of a bilateral fusion of the adductor magnus and adductor longus. Fusion of these muscles may have altered his gait. Gastrointestinal anomalies are also associated with penoscrotal transposition. It is unclear whether this individuals gastrointestinal problems were related to the penoscrotal transposition. Furthermore, cardiovascular conditions have been correlated in men with this condition. Although there was no mention of cardiac disease in the death certificate, observations of the heart revealed the possibility that he had an infarction of the inferior wall of the heart. Unfortunately, examination of the brain was not conducted in this study. The presence of anomalies in the ventricles which have been found in some men with penoscrotal transposition, may have contributed to his developing Parkinson's disease.2

There are surgical procedures to reposition the penis superior to the scrotum, such as the Glenn Anderson technique where incisions were made to lower the scrotum and raise the penis, as well as graft tissue from the thigh to the pubic area. This technique and several others, are fraught with complications in nearly 50% of patients, resulting in severe lymphedema.3 Anatomical observations were that the length and elasticity in the spermatic cord may have been insufficient to lower the testes and scrotum any farther. Also, the patient may not have been a candidate due availability of procedures during his infancy when this is normally addressed.

The psychosocial implications of this condition have not been discussed in the literature. The location of the scrotum would have led to greater potential for trauma to the scrotum. Leaning against a counter or lying on his stomach may have been very uncomfortable. His intimate interactions would have required finding a partner who was accepting of his unique anatomy. This may have added an additional layer of strain to his life including depression and anxiety. However, this is difficult to assess without prior medical records. Because penoscortal transposition is rare, there is still a lot to learn about the causes, treatment, and psychosocial needs of these individuals.

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