Transverse testicular ectopia with disorders of sex development

Katsuya Aoki, Masaomi Kuwada, Kiyohide Fujimoto, Yoshihiko Hirao
Department of Urology, Nara medical University, 840 Shijo-cho, Kashihara, Nara, Japan

ABSTRACT

Transverse testicular ectopia (TTE) is a rare congenital anomaly. Although TTE often coexists with abnormalities such as inguinal hernia and persistent Mullerian duct syndrome, disorders of sex development (DSD) in combination with TTE is extremely rare. We report a case of DSD with sex chromosomal abnormality in combination with TTE. To our knowledge, this case report is a first presentation of such anomaly.

Key words: Disorders of sex development, laparoscopy, transverse testicular ectopia

INTRODUCTION

Transverse testicular ectopia (TTE) is a rare congenital anomaly in which both testes descend through a single inguinal canal. Several co-morbidities of TTE, such as persistent Mullerian duct syndrome and inguinal hernia, have been reported, but TTE with disorders of sex development (DSD) is rare.

We report herein our case of TTE with 45,XO/46,XY DSD with proximal hypospadias.

CASE REPORT

A 8-year-old boy with 45,XO/46,XY DSD with proximal hypospadias was presented to our hospital for further examination for a left inguinal mass. He underwent one-stage hypospadias repair for scrotal hypospadias and exploratory laparotomy in another hospital when he was one year old. At that time the right spermatic vessels and vas deferens, which ran into the left inguinal internal ring along with the left spermatic vessels and vas deferens, was found in the exploratory laparotomy, and the right testis was diagnosed as a vanishing testis via left scrotal approach because a similar structure like the nubbin was located cephalad to the left testis. After that, he was referred to our hospital for a small tumor positioned cephalad to the left testis. Physical examination revealed a normally positioned left testis and a small tumor cephala to the left testis. Ultrasound showed a homogeneous mass like a normal testis cephalad to the left testis, so we suspected right TTE. We performed laparoscopic and left inguinal explorations to confirm the diagnosis. Laparoscopy revealed that the right spermatic vessels and vas deferens ran into the left inguinal internal ring along with the left spermatic vessels and vas deferens, and we didn’t find any persistent Mullerian duct and right intraabdominal testis [Figure 1]. Subsequently, we found two separate spermatic cords and gonads in the left inguinal exploration [Figure 2]. The perioperative histopathological examination revealed normal testicular tissue, and therefore we performed trans-septal orchidopexy for the right testis under laparoscopic guidance.

DISCUSSION

TTE is a rare congenital anomaly in which both testes descend through a single inguinal canal. About 100 cases of TTE have been reported in literature. Although TTE often coexists with abnormalities such as inguinal hernia and persistent Mullerian duct syndrome, DSD in combination with TTE is extremely rare. DSD affect approximately 2 in 10,000 live births, but there are no previous reports about DSD with sex chromosomal abnormality in combination with TTE. This case report is a first presentation.
The precise etiology of TTE is still unclear. Various anatomic factors (defective implantation, rupture, or tearing of the gubernaculum, obstruction of the internal inguinal ring, development of adhesions between the testis and adjacent structures, late closure of the umbilical ring, etc.) are suggested as causative or inducible factors in failure of testicular descent. In this case, considering coexisting DSD with sex chromosomal abnormality, inappropriate androgen secretion or abnormality of androgen receptor might have contributed to the development of TTE. Also, rare cases of anti-Mullerian hormone gene mutations with unilateral TTE with absence of Mullerian duct structures have been reported. Our case was not examined, but molecular studies may find a relevance.

Recently, there have been some reports of laparoscopic surgery for TTE. Laparoscopy clearly reveals the anatomy of internal inguinal ring and intraperitoneal space, and is helpful in performing bilateral orchidopexy in TTE without confusion regarding the laterality of the testes. Especially in case of impalpable testis with DSD, laparoscopy is very useful tool for searching the localization of impalpable testis and internal genitalia at one time.

CONCLUSION

This case is a first report about DSD with sex chromosomal abnormality in combination with TTE. Laparoscopy is very useful for both diagnosis and treatment of TTE especially in patients with DSD to examine the internal genitalia concomitantly.

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