Testicular torsion in a patient with Cohen syndrome

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ABSTRACT

Cohen syndrome is an extremely rare autosomal recessive disorder. A 12-year-old boy with Cohen syndrome applied to a primary health care center because of severe pain in the left groin and was diagnosed with epididymo-orchitis. Despite the administered antibiotic treatment, pain increased. Therefore, the family brought the patient to the emergency department 16 h after the first diagnosis. The patient had mild mental retardation, myopia, and craniofacial dysmorphism, which are components of Cohen syndrome. There was no blood flow on the left testicle at color Doppler ultrasonography. Further, scrotal exploration was performed because of a high risk of torsion. The left testicle was torsioned, and the color was dark blue. Revascularization could not be achieved by detorsion; left orchiectomy and right testicular fixation were then conducted. In conclusion, to the best of our knowledge, this is the first reported case of testicular torsion in Cohen syndrome. If a patient with this syndrome has acute groin pain, testicular torsion should be immediately ruled out with Doppler ultrasonography. These patients may not clearly and correctly express themselves because of mild mental retardation. Moreover, detailed genitourinary, particularly testicular examination may clarify the omitted pathologies and make them well known in future in this syndrome.

Keywords: Cohen syndrome; orchiectomy; testicular torsion

Introduction

Cohen syndrome is an extremely rare autosomal recessive disorder described by Cohen in 1973.^[1] Obesity, mental retardation, craniofacial dysmorphism, high myopia and/or retinal dystrophy, developmental delay, joint laxity, and neutropenia are distinctive characteristics of the syndrome.^[1,2] To the best of our knowledge, any uropathology has not been documented in more than 1500 reported Cohen syndrome cases. Herein, we reported the first testicular torsion case in this special patient group.

Case presentation

A 12-year-old boy with Cohen syndrome applied to a primary health care center because of severe scrotal pain. The patient had received oral antibiotics with an initial diagnosis of epididymo-orchitis. Sixteen hours from the initial diagnosis, despite being administered oral antibiotic treatment, the patient was readmitted to our emergency department with increased scrotal pain. Physical examination revealed

some typical components of Cohen syndrome, such as mild mental retardation, myopia, and craniofacial dysmorphism. Furthermore, scrotal examination revealed left scrotal hyperemia and left testicular tenderness, toughness, and pain. Elevated left testis was also documented. Mild leukocytosis (14.400 per mL) was documented. Microscopic urinalysis also revealed 6–7 leukocytes per field. Color Doppler ultrasonography was finally performed for differential diagnosis, and no arterial blood flow was documented. Based on these signs, scrotal exploration was immediately performed under general anesthesia because of high risk of torsion. We found that the left spermatic cord was torsioned and the left testis was ischemic (Figure 1). Despite detorsion, no color change was observed, and left orchiectomy and right testicular fixation was performed. The postoperative period was uneventful, and the patient was discharged on the first postoperative day.

Discussion

We could not find any urogenital abnormalities, which have been previously reported in

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Submitted:
27.03.2014

Accepted:
03.07.2014

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Figure 1. The color of torsioned testis

Cohen syndrome. This syndrome includes hypotonia and joint laxity in addition to many other findings.^[3] Hypotonia of the abdominal wall and cryptorchidism are commonly observed in diseases such as prune belly and Lowe syndrome.^[4-6] In contrast, no testicular abnormality has been reported in Cohen Syndrome. In our case, the patient had a history of bilateral cryptorchidism up to the second year of his life. Additionally documented testicular torsion makes this case unique in this special patient group.

There are case reports about testicular torsion and some other urogenital disorders in several syndromes^[7,8] In contrast, there is a lack of data regarding urogenital abnormalities, particularly testicular torsion in Cohen syndrome, suggesting a need for special attention and a careful examination of the urogenital system in this syndrome.

Testicular torsion is a surgical emergency that requires immediate intervention to restore the blood flow.^[9,10] Patients who have reduced mental ability may not correctly express themselves, and this may lead to some delay in diagnosis. In this case, torsion was diagnosed 16 h after the onset of symptoms. However, it was too late for a surgical correction. If a patient with Cohen syndrome has acute groin pain, testicular torsion should be immediately ruled out. Doppler ultrasonography may be useful to clarify the situation and rule out testicular torsion in every patient.

In conclusion, physicians should be more suspicious for torsion in a child presenting with acute testicular pain, particularly if he has a syndrome accompanied by decreased mental ability such as Cohen syndrome. Color Doppler ultrasonography should be con-

sidered to avoid misdiagnosis or delayed diagnosis. Moreover, detailed genitourinary, particularly testicular examination may clarify some other previously omitted pathologies that may become typical characteristics of the syndrome in future.

Informed Consent: Written informed consent was obtained from patient and patients' parents who participated in this case.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept - Ö.Y.; Design - Ö.Y., C.Y.; Supervision - H.S.; Funding - E.M., C.Y.; Materials - Ö.Y., C.Y.; Data Collection and / or Processing - E.M.; Analysis and / or Interpretation - Ö.Y., E.M., H.S.; Writer - Ö.Y.; Critical Review - H.S., E.M., C.Y.; Other - Ö.Y.

Conflict of Interest: No conflict of interest was declared by the authors.

Financial Disclosure: The authors declared that this study has received no financial support.

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