PEDIATRIC UROLOGY
Case Report

Testicular torsion in a patient with Cohen syndrome

Cohen Sendromlu bir çocukta testis torsiyonu

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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 the 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.

Key words: Cohen syndrome; orchiectomy; testicular torsion

ÖZET

Cohen Sendromu oldukça nadir görülen otozomal resesif bir hastalıktır. Bu sendromun komponentlerinden hafif mental retardasyon, miyopi, özel yüz görünümü olan 12 yaşında bir erkek çocuk sol kasık ağrısı nedeni ile 1. basamak sağlık merkezine götürülmüş ve epididimorşit tanısı konularak antibiyotik tedavisi başlanmış. Ağrının daha da artması nedeni yaklaşık 16 saat sonra acil servise getirilen hastaya yapılan skrotal doppler ultrasonografi tetkikinde sol testis kanlanmasının olmadığı görülerek testis torsiyonu tanısı ile skrotal eksplorasyon yapıldı. Detorsiyone edilmesine rağmen rengi düzelmeyen testise orşiektomi uygulandı aynı seansta karşı testise de fiksasyon yapıldı. Bu vaka literatürde Cohen Sendromu'nda sunulan ilk testis torsiyonu vakası. Kendilerini ifade etmekte güçlük çeken mental kabiliyetleri kısıtlı hastalarda testis ağrısı değerlendirilirken mutlaka skrotal doppler ultrasonografi uygulanmalıdır. Cohen Sendromu'nda genitoüriner sistemin teferruatlı irdelenmesi yeni bilgiler sağlayabilir.

Anahtar kelimeler: Cohen Sendromu; orşiektomi; testis torsiyonu.

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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 mcL) 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

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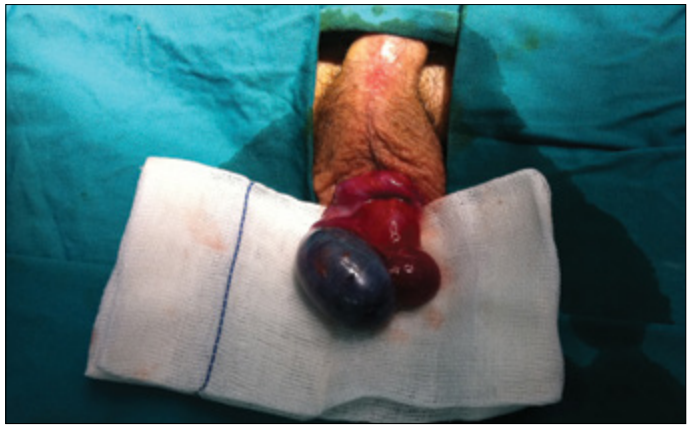

Figure 1. The color of torsioned testis

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 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 considered 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.

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References

1. Cohen Jr MM, Hall BD, Smith DW, Graham CB, Lampert KJ. A new syndrome with hypotonia, obesity, mental deficiency and facial, oral, ocular and limb anomalies. J Pediatr 1973;83:280-4. [CrossRef]
2. Norio R, Raitta C, Lindahl E. Further delineation of the Cohen syndrome; report on chorioretinal dystrophy, leukopenia and consanguinity. Clin Genet 1984;25:1-14. [CrossRef]
3. Kivitie-Kallio S, Norio R, Cohen syndrome: essential features, natural history and heterogeneity. Am J Med Genet 2001;102:125-35. [CrossRef]
4. Sutherland RS, Mevorach RA, Kogan BA. The prune-belly syndrome: current insight. Pediatr Nephrol 1995;9:770-8. [CrossRef]
5. Haset S, Smith H, Holland AJA. Prune belly syndrome. Pediatr Surg Int 2012;28:219-28. [CrossRef]
6. Loi M. Lowe syndrome. Orphanet J Rare Dis 2006;1:16. [CrossRef]
7. Papatsoris AG, Mpadra F, Karamouzis M, Likaki-Karatza E, Karatzas T. Torsion of undescended testis in a man with Down's syndrome. Int J Urol 2003;10:233-5. [CrossRef]
8. Flynn BJ, Myers SM, Cera PJ, Mowad JJ. Testicular torsion in an adolescent with Fragile X Syndrome. Pediatrics 2002;109:E16. [CrossRef]
9. Ciftci AO, Senocak ME, Cahit Tanyel F, Buyukpamukcu N. Clinical predictors for differential diagnosis of acute scrotum. Eur J Pediatr Surg 2004;14:333. [CrossRef]
10. Kılıç N, Balkan E. Çocuklarda Akut Skrotum Patolojileri. Güncel Pediatri 2004;2:122-5.