Coc Cer Derg/Turkish J Ped Surg 2022;36(1): 55-57
doi: 10.29228/JTAPS.53680

Case Report

Çocuk Cerrahisi Dergisi
Turkish Journal of Pediatric Surgery
Volume 36
Number 1
2022

Hyperimmunoglobulinemia D Syndrome Presenting With Acute Scrotum: A case report

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ABSTRACT

Mevalonate kinase deficiency, known as hyperimmunoglobulinemia D syndrome originates from recessive mutations in the gene encoding mevalonate kinase enzyme. Hyperimmunoglobulinemia D syndrome cases are characterised by repeated attacks of fever with an intense inflammatory syndrome accompanied by lymphadenopathy, abdominal pain, diarrhea, arthralgia, hepatomegaly, splenomegaly, and skin rashes. The clinical phenotype in hyperimmunoglobulinemia D syndrome, which is an autoinflammatory disease, has a broad range depending on the severity of mevalonate kinase activity. However, only one case presenting with orchitis has been reported. The case here reported is of a paediatric hyperimmunoglobulinemia D syndrome patient with orchitis presenting with clinical testis torsion.

Keywords: *Mevalonate kinase deficiency, orchitis, acute scrotum*

Geliş/Received: 21.06.2021
Kabul/Accepted: 30.08.2021
Published online: 01.04.2022

Cite as: Karakaya AE, Güler AG, Kaya MS, Güngör Ş. Hyperimmuno- globulinemia D syndrome presenting with acute scrotum: a case report. Coc Cer Derg/Turkish J Ped Surg 2022;36(1):55-57 doi: 10.29228/JTAPS.53680

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Introduction

Mevalonate kinase deficiency, known as hyperimmunoglobulinemia D syndrome (HIDS) originates from recessive mutations in the gene encoding mevalonate kinase enzyme[(1)]. HIDS cases are characterised by repeated attacks of fever with an intense inflammatory syndrome accompanied by lymphadenopathy, abdominal pain, diarrhea, arthralgia, hepatomegaly, splenomegaly, and skin rashes. The clinical phenotype in HIDS, which is an autoinflammatory

disease, has a broad range depending on the severity of mevalonate kinase activity. However, only one case presenting with orchitis has been reported[2]. The case here reported is of a paediatric HIDS patient with orchitis presenting with clinical testis torsion.

Case Presentation

A 2-year-old male presented with the complaints of redness, swelling, and pain in the left testis. The child had previously been diagnosed with HIDS from determination of MEFV(NM-000243.2) gene c.2040G>C (p.Met680lle) heterozygote and MVK(NM_000431.4) gene c.1129G>A (p.Va1377lle) variation homozygote gene mutations which were examined at the age of 1 year when investigating the complaints of recurrent fever, vomiting, and rashes, that had started at the age of 3 months. In the physical examination, the left testis was found to be hyperemic, oedematous, and painful to the touch. The laboratory test results were WBC:13.36 $x10^3$ cells/uL, Hb: 10.5 g/dL, PLT: 246 $x10^3$ cells/uL, and CRP: 21. Biochemical parameters were seen to be normal. In the scrotal colour Doppler USG examination, the dimensions and thickness of the left epididymis were seen to be increased compared to the right side, the left testis dimensions and thickness values were lower than those of the right side, and anechoic free fluid was observed measuring 1 cm at the deepest point of the left scrotum. Upon the report that the blood supply of the left testis was decreased compared to the right, surgery was decided with the suspicion of torsion. There was observed not to be torsion in left testis but there was hyperemic, peritesticular fluid, which was accepted as orchitis and the operation was terminated. Empirical antibiotherapy was started. No discharge or infection developed in the wound site during follow up, and the clinical condition returned to normal.

Discussion

HIDS was first described by Van der Meer et al in 1984[3], and since that time various clinical and laboratory characteristics of HIDS have been reported[4,5]. It is possible that HIDS has become more common with increasing awareness of reported cases. With the reporting of the uncommon presentation of the current case, it was hoped to contribute to the expansion of the clinical spectrum of HIDS.

Consistent with the literature, the current case had symptoms of recurrent fever and rash before the age of one year[1,2]. In addition, while under treatment at 1 year after the diagnosis, the patient experienced the first attack of orchitis with clinical presentation of testis torsion. This is the second case to be reported in literature of the combination of HIDS and orchitis. In the first reported case in literature[2], the patient had two orchitis attacks and there was a family history of orchitis, which was not present in the current case. Orchitis is a characteristic of other periodic fever syndromes, and after the reporting of these cases, it should be kept in mind that HIDS cases may present with the clinical condition of orchitis.

Houten et al[6] and Drenth et al[7] showed that HIDS originated from mutations in the MVK gene, which are compound heterozygote in most HIDS patients. In 80% of patients with one mutation, it is V3771 mutation on exon 10[8,9].

In contrast to the literature, the current case was determined with both heterozygote mutation in MEFV gene exon 10 and homozygote mutation in MVK gene exon 11.

Testis torsion is a clinical diagnosis, and patients present with severe pain, nausea and vomiting. Patients with suspected torsion in the history or physical examination must be admitted for emergency surgical evaluation before the development of permanent ischaemic damage. Time should not be lost with imaging[10]. Despite the clinical consideration of left testis torsion in the current patient and the reduced blood flow in the epididymis seen on Doppler USG, findings consistent with orchitis were observed during the operation. This could be explained by the intense inflammation expected in HIDS.

In conclusion, this report constitutes the second case in literature of a patient diagnosed with HIDS presenting with clinical orchitis. This case report also shows that orchitis and testis torsion may be confused clinically in HIDS cases.

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