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Case report
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Congenital isolated asplenia accidentally discovered during acute peritonitis

Akut peritonit sırasında tesadüfen fark edilen konjenital izole asplenia

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Abstract

An extremely rare condition, congenital asplenia has 2 distinct types: heterotaxy syndromes and isolated congenital asplenia (ICA). Ivemark syndrome is one of the heterotaxy syndromes characterized by asplenia, malformations of the heart, and malposition of internal organs in the chest and abdomen. ICA cases are also fatal in childhood, but there are reported living adult cases. Those affected are typically at increased risk for fulminant sepsis and carry a higher risk of noninfectious complications, such as thrombocytosis and mesenteric thrombosis. We herein report the unusual case of a patient with congenital asplenia, which was discovered fortuitously during an emergency laparotomy for peritonitis due to ulcer perforation.

Keywords: Congenital isolated asplenia, Adult peritonitis

Öz

Çok nadir görülen bir durum olan konjenital aspleninin 2 farklı tipi vardır: heterotaksi sendromları ve izole konjenital aspleni (ICA). Ivemark sendromu, asplenia, kalbin malformasyonları ve göğüs ve karındaki iç organların malpozisyonu ile karakterize heterotaksi sendromlarından biridir. ICA vakaları çocuklukta da ölümcüldür, ancak bildirilen canlı yetişkin vakaları mevcuttur. Etkilenenler tipik olarak fulminan sepsis için yüksek risk altındadır ve trombositoz ve mezenterik tromboz gibi bulaşıcı olmayan komplikasyon riski daha yüksektir. Burada ülser perforasyonuna bağlı peritonit için acil bir laparotomi sırasında tesadüfen saptanan doğuştan asplenisi olan bir hastanın olağandışı olgusunu sunuyoruz.

Anahtar kelimeler: Konjenital izole asplenia, Yetişkin peritoniti

Introduction

Isolated congenital asplenia is an exceptional form, and rare cases have been reported in the literature (Table 1) [1-4]. It is usually diagnosed after the onset of severe infections in infants. In adults without a history of severe sepsis in infancy, the presenting sign may be thrombocytosis [5]. Our case shows that congenital isolated asplenia patients can remain asymptomatic for a long time, or even lead normal lives.

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Table 1: Classification of asplenia

• Congenital	• Acquired
Isolated	Splenectomy
Familial	Trauma
Non-familial	Hemolytic anaemia
	ITP
	Malignancy
• Syndromatic	• Functional/hyposplenism
Ivemark	HbSS/Sickle-cell anaemia
Storkmorken	Portal hypertension
Kartagener	Storage diseases (Amyloidosis, M. Gaucher)
Meckel	Iatrogenic (radiotherapy)
Pallister-Hall	
Cystic liver, kidney, pancreas	
MLRD (microgastria-limb reduction defects association)	
Smith – Fineman – Meyers	
Schmidt	

ITP: idiopathic thrombopenic purpura

Case presentation

A 65-year-old male patient of Moroccon origin, a chronic smoker without a pathological personal or family history, was consulted urgently due to diffuse abdominal pain. Physical examination revealed a conscious, febrile, dehydrated patient who was hypotensive with a blood pressure of 90/60 mmHg. He had generalized abdominal defense of the left pelvis and iliac fossa and diffuse tenderness in all four abdominal quadrants. Laboratory results showed leukocytosis at 18 g/L, C-reactive protein at 250 mg/L, slight thrombocytosis at 500 g/L, and acute renal failure. Hepato-pancreatic functional tests were normal. Lung radiograph showed pneumoperitoneum right below the diaphragm, consistent with perforation of a hollow organ, after which we decided to operate on the patient. An urgent laparotomy was performed, which revealed the presence of about 1 L of pus intraabdominally, and anteriorly perforated duodenal bulbus. The perforation was sutured, and intraabdominal lavage and drainage were done. During abdominal exploration, no spleen was observed in the left hypochondrium (Figure 1). Postoperative follows-up were uneventful. Subsequently, a thoraco-abdominopelvic CT scan was obtained, which confirmed the absence of the spleen, and excluded any other abdominal or thoracic malformation (Figure 2).The postoperative hematologic assessment revealed the presence of Howell-Jolly corpuscles confirming functional asplenia.

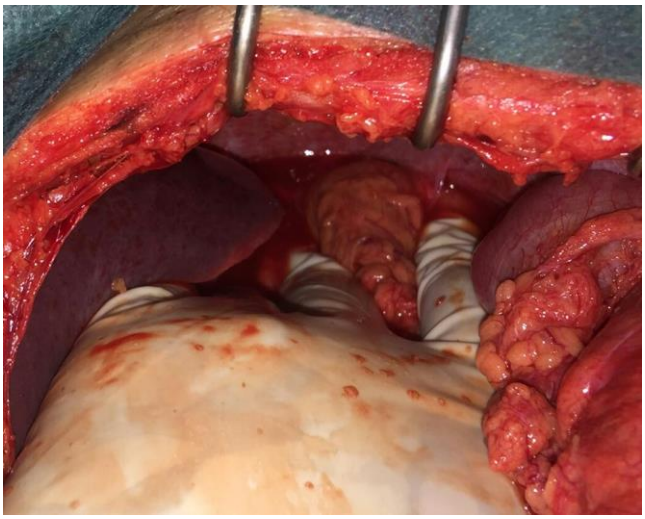

Figure 1: Per operatory image shows an absence of spleen in a left hypochondrium

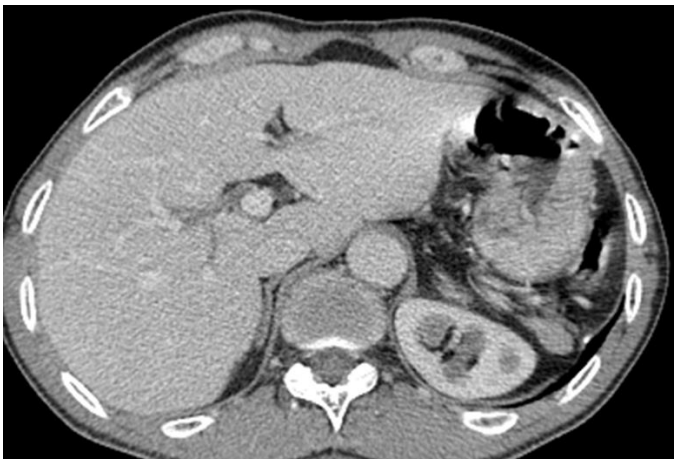

Figure 2: CT image shows an Empty splenic lodge occupied by the tail of the pancreas

Discussion

Congenital asplenia, a poorly understood and rare form of primary immunodeficiency, is either associated with malformation syndromes such as heterotaxia, or presents as an isolated finding, as was the case in our patient [2]. Heterotaxia syndrome with situs abnormalities (Ivemark syndrome) is a sporadic, autosomal recessive syndrome seen in cases of parental consanguinity. Patients with Ivemark syndrome generally die by the age of 6 months. On the other hand, ICA, with the plausible autosomal dominant mode of inheritance, is often fatal in early childhood or complicated with life-threatening infections such as meningitis and purpura fulminans or noninfectious complications such as thrombocytosis and mesenteric thrombosis [6,7].

Bolze et al. [7] studied 33 patients with isolated congenital asplenia from 23 families, including 5 families previously reported by Mahlaoui et al. [6] and those described Ferlicot et al. [8], and suggested that heterozygous coding mutations in RPSA on chromosome 3p21 underlie most cases of isolated congenital asplenia, with apparently complete penetrance. RPSA is not likely to have been identified through a candidate-gene approach, as RPSA is ubiquitously expressed and is not known to be involved in spleen development [7].

The clinical record that we report is interesting since this patient had congenital isolated non-syndromic asplenia, discovered incidentally at the age of 65 years. The anamnesis was negative for any septic episode or other anomalies. Prolonged survival in the absence of cardiopulmonary and infectious complications is therefore possible in patients with congenital asplenia. Rare cases, to our knowledge, have been reported [9]. But in most cases, in the absence of cardiac malformations, asplenia is discovered at autopsy, as mentioned in the case of a child who died from pneumococcal sepsis [10].

This prompted our comprehensive literature search of adult ICA cases that presented with complications other than those related to infection [2]. Eighteen adult cases of ICA were identified and analyzed since the first report of Myerson and Koelle in 1956. Eleven of the 18 reported cases were sporadic and the remaining were familial. Familial cases are generally asymptomatic and usually diagnosed after a close family member/child suffers a life-threatening or fatal infection secondary to congenital asplenia. Associated findings include thrombocytosis, mesenteric vein thrombosis, chronic thromboembolic pulmonary hypertension, and pneumococcal

sepsis (Table 2). Seven adult patients with ICA had invasive bacterial infections [6-12]. In addition, there were 5 adult cases of ICA with thrombocytosis but no infectious events were found [13,14].

Table 2: Adult isolated congenital cases reported from 1956 till 2015

Case No.	Familial or sporadic	Age at diagnosis/gender	Clinical presentation	Outcome
1	Sporadic	36 years/Male	Pneumococcal sepsis/Waterhouse-Friderichsen syndrome	Deceased
2	Sporadic	37 years/Male	Thrombocytosis	Alive
3	Sporadic	56 years/Female	Thrombocytosis/myocardial infarction	Alive
4	Sporadic	56 years/Male	Thrombocytosis	Alive
5	Sporadic	60 years/Female	Pneumococcal sepsis	Alive
6	Sporadic	77 years/Male	Mesenteric vein thrombosis	Alive
7	Sporadic	52 years/Female	Pneumococcal sepsis	Deceased
8	Sporadic	72 years/Male	Thrombocytosis	Alive
9	Familial	20 years/Female	Pneumococcal sepsis/2 children affected	Alive
10	Familial	35 years/Male	Asymptomatic/5 children affected	Alive
11	Familial	45 years/Male	Meningitis-pneumococcal/2 children affected	Alive
12	Familial	Unknown/Male	Asymptomatic/1 child affected	Alive
13	Familial	25 years/Male	Thrombocytosis/1 child affected	Alive
14	Familial	Unknown/Female	Asymptomatic/2 children affected	Alive
15	Familial	27 years/Male	Asymptomatic/sister affected with AVM	Alive
16	Adopted	22 years/Male	Small bowel AVM bleed, Mycoplasma pneumonia	Alive
17	Sporadic	28 years/Male	Streptococcal pneumonia/ulcerative colitis	Alive
18	Sporadic	44 years/Female	Thrombocytosis/Chronic thromboembolic pulmonary hypertension	Alive
19	Sporadic	67 years/Female	Waterhouse-Friderichsen syndrome/lung fibrosis	Deceased

Reflecting another risk associated with thrombocytosis in adults, Takahashi et al. [15] also described the case of a 44-year-old female with ICA who had chronic thromboembolic pulmonary hypertension. The diagnosis of functional asplenia was confirmed by the analysis of the peripheral blood smear, which revealed the presence of Howell-Jolly corpuscles [10]. The diagnosis of isolated congenital asplenia is confirmed apart from any infectious complication or cardiovascular malformation which probably explains the prolonged survival in our patient. Finally, the interest of vaccination against pneumococcal infections and even long-term antibiotic prophylaxis was discussed, but vaccination did not seem justified in this very particular situation [9].

Conclusion

This clinical presentation suggests that isolated congenital asplenia is compatible with a normal and prolonged existence in the absence of any infectious complications, and cardiovascular and digestive tract malformations.

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