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CASE REPORT

CHOLELITIASIS IN AN ADULT PATIENT WITH MILD HEREDITARY SPHEROCYTOSIS – A CASE REPORT

Bogdan SOCEA[1]✉, Anca A. NICA[1], Alexandru C. SMARANDA[1], Ovidiu G. BRATU[2], Camelia C. DIACONU[3], Vlad D. BĂLEANU[1], Dragoș V. DAVIȚOIU[1], Mihai DIMITRIU[4], Alexandru C. CARÂP[1], Simona BOBIC[1], Vlad D. CONSTANTIN[1]

[1] Emergency Clinical Hospital „Sfântul Pantelimon", General Surgery Clinic, „Carol Davila" University of Medicine and Pharmacy, Bucharest, Romania
[2] Emergency Universitary Central Military Hospital, Department of Urology, „Carol Davila" University of Medicine and Pharmacy, Academy of Romanian Scientists, Bucharest, Romania
[3] Internal Medicine Clinic, Clinical Emergency Hospital of Bucharest, „Carol Davila" University of Medicine and Pharmacy, Bucharest, Romania
[4] Emergency Clinical Hospital „Sfântul Pantelimon", Obstetrics and Gynecology Clinic, „Carol Davila" University of Medicine and Pharmacy, Bucharest, Romania

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Abstract

Hereditary spherocytosis (HS) is an inherited abnormality of the red blood cell, caused by defects in structural membrane proteins. The condition is dominantly inherited in 75% of people. The severity of the disorder is related to the type and amount of membrane disruption, which is genetically determined. A patient who suffers from this disorder is commonly found in a surgical ward when the disease becomes unmanageable by a hematologist. Surgeons encounter complications such as: jaundice, splenomegaly, gallstone sand severe anemia. We present the case of a 66-year-old woman with a history of hereditary spherocytosis who presented at the emergency room for pain in the right upper quadrant, jaundice and anemia and was diagnosed with gallbladder stones and common bile duct obstruction.

Résumé

Cholélithiase chez un patient adulte avec sphérocytose héréditaire légère- rapport de cas

La sphérocytose héréditaire (SH) est une anomalie héréditaire des globules rouges, provoquée par des défauts des protéines de la membrane structurale. La maladie est principalement héréditaire chez 75% des personnes. La gravité de la maladie est liée au type et à l'ampleur de la perturbation de la membrane, qui est génétiquement déterminée. Un patient qui souffre de ce trouble est généralement trouvé dans une salle d'opération lorsque la maladie devient ingérable par un hématologue. Les chirurgiens rencontrent des complications telles que : jaunisse, splénomégalie, anémie sévère du sable biliaire. Nous présentons le cas d'une femme de 66 ans présentant des antécédents de sphérocytose héréditaire, qui s'est présentée à l'urgence

✉ Address for correspondence: Bogdan SOCEA
Emergency Clinical Hospital „Sfântul Pantelimon" General Surgery Clinic
Address: Șoseaua Pantelimon no. 340-342, 1st floor, Bucharest, Romania
Email: bogdansocea@gmail.com; Phone: +40788491091; Fax: +40212550064

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pour des douleurs dans le quadrant supérieur droit, un ictère et une anémie. Des calculs de la vésicule biliaire et une obstruction des voies biliaires ont été diagnostiqués.

Mots-clés: sphérocytose héréditaire, jaunisse, obstruction de voie biliaire principale.

Introduction

Hereditary spherocytosis (HS) is a potential life-threatening hematological disease, that has a variety of mutations that lead to defects in red blood cell membrane proteins. Typically, there are four forms of HS: mild, moderate, moderate to severe and the severe form, depending on the severity of signs and symptoms[1,2]. This hematological disorder has at least five genes mutations that cause the red blood cell membrane to be misshaped and rigid so, instead of a flattened disc, the shape of the cell is spherical[3]. When they reach the bloodstream, because of their inability to change shape, they are removed from circulation and taken to the spleen for destruction. This is the main mechanism for the signs and symptoms of HS, shortage of red blood cells in the bloodstream and the presence of a large number within the spleen, where they undergo hemolysis[4].

When a blood cell breaks down, bilirubin is released. When the red blood cells break down too fast, increased bilirubin is released in the blood stream and jaundice appears. Besides anemia, jaundice is another important sign of HS.

Often, this high level of bilirubin causes gallstones in patients with HS. They are often misdiagnosed, because these deposits of bilirubin can be easily broken down in the gallbladder. Gallstones develop in less than half of patients with mild HS and do not always cause symptomatic biliary tract disease.

Decisions regarding splenectomy during cholecystectomy remain controversial, not only because of the postoperative complications, such as overwhelming post-splenectomy infections (OPSI) and thrombo-embolic events, but also because surgeons need a good hematological control of the disease when the decision is made.

Case presentation

We present the case of a 66-year-old woman, who presented at the emergency room for pain in the right upper quadrant, jaundice, anemia and loss of appetite. The patient was admitted to our clinic 6 months before, for acute pancreatitis, jaundice and severe anemia. During her last admission, she was transferred to the intensive care unit for blood transfusions.

Ultrasound showed an enlarged gallbladder, with several deposits located in the fundus, common biliary duct measuring 11.3 mm and splenomegaly of 212 mm longitudinal axis; complete blood count showed leukocytosis 17.43*1000/uL, anemia 8.40 g/dL, and high levels of total bilirubin 8 g/dL. The level of bilirubin was too high to be considered only in the context of an acute abdomen[5].

The clinical exam was normal, revealing only a mild discomfort at palpation of the abdomen in the right upper quadrant.

We decided to delay the surgery and to begin the antibiotic treatment, because two days after admission, the ultrasonography showed no gallbladder stones and the common biliary duct measured 5 mm. In this case, usually when the process of hemolysis stops, the gallstones are broken down. After 3 days of antibiotics, the blood count showed a normal count of WBC, and higher levels of total bilirubin (17 g/dL).

We took into consideration the transfer of the patient to a hematological ward for treatment, and to decide if a splenectomy is necessary. Meantime, the symptoms worsened, with constant pain in the epigastric area, right upper quadrant, nausea and fever; the blood count revealed very high levels of bilirubin (40 g/dL).

We decided to perform an open cholecystectomy. We had an anterior approach of the gallbladder and also performed a cholangiography, that showed choledocolithiasis. After common bile duct stones were extracted, we maintained an external drainage of the bile for 3 days post-op. Although the patient had an enlarged spleen, we could not perform a splenectomy without the hematological indication.

Patient had a fast recovery, on the 3rd day post-op a contrast cholangiography on the external drainage was performed, that showed no blockages between the common bile duct and the duodenum, so the drain was removed. The bilirubin levels improved, the blood count revealed no signs of infection or severe anemia and the patient was discharged after 8 days.

Discussion

The disease was first described in the second half of the 19th century. In 1900, Oskar Minkowski published his observations on familial clusters. Hereditary spherocytosis belongs to the congenital hemolytic anemias, named after the microscopic aspect of spherocytes in a blood smear[6].

Surgical implications are very unclear regarding HS, cholecystectomy and splenectomy being the most commonly performed interventions. In both cases, often the surgeons need a hematologist indication, because of the peri- and postoperative complications. Patients that suffer from HS are diagnosed early and the symptoms are manageable with proper treatment, surgery being a last option in cases with severe anemia and obstructive jaundice.

In most cases, jaundice is only transitory, and when a patient with pain in the right upper quadrant, jaundice and a normal ultrasound (no blockages in the gallbladder or common bile) presents at the emergency room, the diagnosis of a hemolytic disorder must be confirmed by a hematologist.

Recent literature demonstrates that splenectomy during a cholecystectomy in a HS patient is not the recommended treatment. It was previously suggested that there is an increased risk of intra-hepatic choledocholithiasis following splenectomy[7] and the 2004 guidelines suggested that for children with HS who require cholecystectomy, the spleen should always be removed[8-13]. This recommendation was based on expert opinion, despite little supportive data in the literature; the recommendation was changed in subsequent guidelines to indicate that this issue remains controversial[14-17]. In a recent study, of 32 pediatric patients with HS, who underwent cholecystectomy, 27 patients underwent synchronous splenectomy. However, none of the five patients who underwent cholecystectomy without splenectomy experienced signs or symptoms consistent with gallstones over a median follow-up of 15.6 years[18].

Also, given the role of the spleen in immune capacity and blood filtration, there is a risk of OPSI (overwhelming post-splenectomy infections), which is highest with encapsulated microorganisms such as *Streptococcus pneumoniae, Neisseria meningitidis* and *Hemophilus influenza*[19]. The risk of post-splenectomy sepsis may vary according to the indication for splenectomy (intermediate risk in spherocytosis and higher in other inherited anemias)[20], patient's age at the time of surgery (highest before the age of 5 years)[21,22], and time since the splenectomy was performed (highest risk during the first year after the intervention). However, the risk probably remains elevated for the entire life. Given the high risk of OPSI at a young age, splenectomy should not be performed before 5 years of age.

Conclusions

Although hereditary spherocytosis is not a disorder often encountered in the surgical specialties, a surgeon must know the correct way of approaching the disease in a manner most beneficial for the patient.

Routine ultrasonographies must be performed in all HS patients, to exclude gallbladder and common bile duct stones. In case of splenomegaly, splenectomy must be performed only after the hematologist's indication in patients transfusion-dependents or suffering from severe anemia, especially if they are still in the teenage years.

There have been no evidenced based studies to support the idea that in HS patients cholecystectomy must be performed routinely.

Compliance with Ethics Requirements:

„The authors declare no conflict of interest regarding this article"

„The authors declare that all the procedures and experiments of this study respect the ethical standards in the Helsinki Declaration of 1975, as revised in 2008(5), as well as the national law. Informed consent was obtained from the patient included in the study"

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