SHORT COMMUNICATION

Congenital asplenia and group B streptococcus sepsis in the adult: case report and review of the literature

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Abstract

Asplenia is associated with an increased incidence of fatal and life-threatening sepsis caused by encapsulated pathogens. Isolated congenital asplenia is a very rare condition, with only 33 cases reported in the literature. The authors report another case of this condition complicated by overwhelming Group B streptococcus sepsis secondary to paronychia that was managed successfully.

Introduction

Asplenia is an uncommon condition that may be acquired (following surgery), functional or congenital. It is associated with an increased incidence of life-threatening sepsis caused mainly by encapsulated bacteria. Functional asplenia is seen in conditions such as sickle cell (SC) disease, hemoglobin SC disease and sickle β-thalassemia. Most children with these hemoglobinopathies are functionally hyposplenic in the first year of life and anatomically asplenic (due to autoinfarction) before the age of 20 (Lane *et al.*, 1995). Congenital asplenia is usually seen in the context of recognized malformation syndromes (Alonso *et al.*, 1995; Casey *et al.*, 1996). Conversely, isolated congenital asplenia is rare, with only 33 cases reported in the literature (Gilbert *et al.*, 2002; Halbertsma *et al.*, 2005; Hummler *et al.*, 2005). Most of these presented in the early years of life and only seven cases were diagnosed in the adult. The authors report another case of this condition in a 28-year-old man presenting with Group B streptococcus (GBS) sepsis, and review the literature.

Case report

A 28-year-old Caucasian male presented with a few hour history of abdominal pain, nausea and vomiting. He had a past history of alcohol abuse and also ulcerative colitis that was in remission for more than a year. He was not on any medications and had never undergone any surgical procedures.

On examination he was found to be pyrexial at 38.9 °C, tachycardic, hypotensive, hypoxic and olyguric. His abdomen was mildly distended with minimal diffuse tenderness. Blood tests revealed severe leucopenia (white cell count $2.3 \times 10^9\,L^{-1}$), markedly deranged renal function tests (serum creatinine 268 μmol L^{-1}, urea 20 mmol L^{-1}), elevated serum amylase (680 IU L^{-1}) and metabolic acidosis with a base excess of −8.3 and a lactate of 2.3 mmol L^{-1}. The patient was noted to have a paronychia of the right big toe. Despite aggressive resuscitation and antibiotic therapy, he deteriorated rapidly and developed multiorgan failure requiring circulatory, ventilatory and renal support.

A provisional diagnosis of severe alcohol-related pancreatitis was made. However, a CT scan of the abdomen and

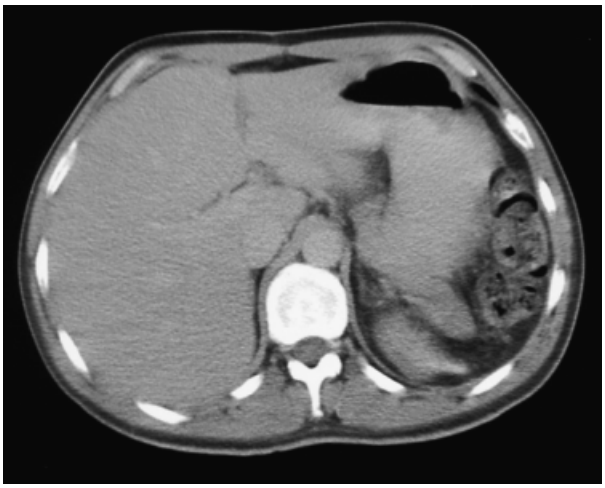

Fig. 1. Nonenhanced CT scan of the abdomen: the absence of a spleen is noted on the left; the colon occupying the empty splenic fossa.

pelvis revealed a normal pancreas and the absence of a spleen (Fig. 1). The presence of Howell Jolly bodies on a blood smear was consistent with asplenia. No family history for this condition was reported.

Serum lipase 2 days following admission was normal. Blood cultures yielded GBS. The paronychia was incised and drained. Microbiology cultures from the pus yielded no bacterial growth.

Over the following 2 weeks, the patient gradually improved and was weaned off the ventilator with a tracheostomy. Nevertheless, he complained of persistent abdominal pain, especially on increasing enteral feeding. A repeat abdominal CT-scan showed a dilated large bowel with no signs of obstruction. The pancreas was normal. A flexible sigmoidoscopy was unremarkable up to the splenic flexure.

In view of persistent abdominal pain and inability to tolerate feeding, it was decided to carry out an exploratory laparotomy. This showed only mild thickening of the sigmoid colon consistent with previous colitis. The spleen was found to be absent. A feeding jejunostomy was fashioned. In the absence of any other infective focus, a final diagnosis of GBS-related sepsis secondary to paronychia was made.

The patient continued to improve postoperatively and made an uneventful recovery. He was vaccinated against encapsulated organisms (*Pneumococcus*, *Meningococcus* and *Haemophilus*) and started on long-term penicillin prophylaxis.

Discussion

Congenital asplenia is most often found in association with other anomalies and is present in *c.* 3% of neonates with structural heart disease and in 30% of patients who die from cardiac malposition (Rose *et al.*, 1975; Katcher, 1980). The most common of these anomalies is Ivemark syndrome. This is characterized by visceral heterotaxy with bilateral right-sidedness. The right-sided organs are duplicated and organs that are normally present on the left side are absent. Infants with this condition usually present with cyanosis and respiratory distress caused by complex cardiac malformations resulting in a high mortality rate in the early months of life (Ruttenberg *et al.*, 1964; Rose *et al.*, 1975; Dyke *et al.*, 1991).

Asplenia syndrome has been related to both autosomal and X-linked inheritance patterns, although most cases occur sporadically. *ZIC 3* is an X-linked gene identified in both sporadic and familial cases: affected males typically have situs ambiguous phenotype, and females have either situs solitus or situs inversus (Gebbia *et al.*, 1997).

Other conditions associated with asplenia include Pearson syndrome (pancreatic insufficiency, sideroblastic anemia), Stormorken syndrome (thrombocytopenia and miosis), Smith–Fineman–Myers syndrome (mental retardation, short stature, cryptorchidism) and ATR-X syndrome (α-thalassemia and mental retardation (Kevy *et al.*, 1968; Katcher, 1980; Leahy *et al.*, 2005). Isolated congenital asplenia is rare and only 33 cases have been reported in the literature. Of these, 20 (54%) were shown to be familial and 13 (46%) sporadic. Clinical presentation was: pneumococcal sepsis (9), pneumococcal meningistis (6), sepsis of unknown origin (5), thrombocytosis (4), *Haemophilus influenzae* sepsis (2) and *Haemophilus influenzae* meningitis (2). Three patients were asymptomatic and diagnosed incidentally. Only seven patients were older than 20 years of age. Mortality after acute presentation with sepsis was 56% (Gilbert *et al.*, 2002; Halbertsma *et al.*, 2005; Hummler *et al.*, 2005).

Other series looking at patients with asplenia have reported up to an 80% mortality arising from sepsis (Dyke *et al.*, 1991; Beytout *et al.*, 2003). These patients deteriorate very rapidly, with death occurring within several days or even hours. This accelerated course of the disease is related to reduced response to antigen (particularly polysaccharides), impaired phagocytosis and decreased levels of tuftsin and properdin (Lynch & Kapila, 1996; Waghorn, 2001).

Streptococcus pneumoniae is responsible for up to 80% of cases. Other organisms include *Haemophilus influenzae* type b, *Neisseria meningitidis, Escherichia coli*, *Staphylococcus aureus* and other streptococci. Gram-negative bacilli including *Salmonella* species, *Klebsiella* species and *Pseudomonas aeruginosa* are less common causes of bacteremia (Waghorn, 2001).

In the case reported GBS was the cause of sepsis. This pathogen is a source of infection especially in peripartum patients. Nonperipartum infections usually occur in the elderly and in patients with underlying systemic diseases (e.g. diabetes, malignancy or alcoholism). The most frequent sites of localized infection are skin or soft tissue infections, followed by pneumonia and urinary tract infections (Schuchat, 1998). GBS produces a toxic shock-like

syndrome associated with a high mortality rate (Schlievert *et al*., 1993).

There have been very few reports of GBS as a pathogen in asplenic patients. Tremlett *et al*. (1994) reported the case of a rapidly fatal septic shock caused by GBS 16 years postsplenectomy. Sims & Barton (2006) reported a case of GBS septic shock in a 42-year-old man with previous splenectomy. Of 456 patients included in two population-based studies of adult GBS disease, only two patients were reported to have asplenia (Bisno, 1971; Farley *et al*., 1993).

In the reported case, absence of a spleen with the presence of Howell Jolly bodies confirmed the diagnosis of congenital asplenia. In the absence of any other infective foci, paronychia remains the most likely source of sepsis. Cultures from the pus were negative, but this was probably the result of antibiotic treatment. Elevated amylase was likely to be the result of the patient's renal failure.

In conclusion, isolated congenital asplenia should be considered in patients with no previous history presenting with overwhelming sepsis. Any focus of infection should be dealt with immediately and aggressive resuscitation initiated to reduce the high mortality rate associated with this condition.

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