

CONGENITAL ASPLENIA IN A SURGICAL PATIENT

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ABSTRACT

Congenital asplenia is rare and could occur sporadically or less commonly as part of a syndrome. Sporadic or isolated congenital asplenia is not usually associated with significant morbidity. Congenital isolated asplenia can be present as septicaemia, commonly pneumococcal, hence the importance of screening the peripheral blood for Howell-Jolly bodies. Abdominal imaging becomes necessary if screening is positive. An isolated case of congenital asplenia is discovered by chance in a 69 year-old man, after falling off a ladder from a height of about 15 feet. Standard surgical procedure was followed in managing the patient. Here, abdominal ultrasound commented on the absence of the spleen followed trauma. Fracture of the spleen was suspected hence exploratory laparotomy was performed. To the best of our knowledge, this report is probably an isolated abnormality and most likely a sporadic case.

Keywords: *Congenital isolated asplenia, laparotomy, Howell-Jolly bodies, trauma*

INTRODUCTION

Congenital asplenia is rare and could occur sporadically or less commonly as part of a syndrome. Sporadic or isolated congenital asplenia is not usually associated with significant morbidity. Studies have however shown a higher incidence of sepsis in childhood in those with congenital asplenia (especially with the syndrome) when compared with the control population (Waldman *et al.*, 1977). In asplenia, encapsulated bacteria cause the majority of infections, and adults are at lower risk than children be-

cause they are more likely to have antibody to these organisms. *Streptococcus pneumoniae* infection is most common, causing 50 to 70% of cases, but the risk of infection with *Haemophilus influenzae* or *Neisseria meningitidis* is also high. Severe clinical manifestations of infections due to *Escherichia coli*, *Staphylococcus aureus*, group B streptococci, *Pseudomonas aeruginosa* as well as some species of *Capnocytophaga*, *Babesia*, and *Plasmodium* have been reported (Kasper *et al.*, 2005)

The functional asplenia seen in sickle cell disease may contribute to severe mycoplasma disease as well as pneumococcal infection. Severe respiratory distress and large pleural effusions may occur. Serious infections are most common during the first five years following splenectomy, and particularly during the first five years; recurrent infections occur in about 20 per cent of those affected. Approximately 50 per cent of infections are meningitis or bacteraemias, and most of the remainder are pneumonias (Warrel *et al.*, 2003).

Congenital isolated asplenia may arise as a minor form of situs abnormalities or result from an unrelated specific defect of spleen development. It is a rare life-threatening condition and pneumococcal sepsis is often the first sign of the disease. A case of congenital asplenia discovered by chance is hereby reported.

CASE REPORT

A 69 year-old man was admitted after falling off a ladder from a height of about 15 feet. He sustained injury to the head, back and abdomen with a brief loss of consciousness. On admission, he was complaining of lower back pain. He was restless, pale, tachycardic with a small pulse volume and reduced blood pressure. There was no detectable abnormality in the chest but abdominal examination revealed tenderness in the left lumbar region with a large bruise. He was also tender over the L3/L4 region. Bowel sounds were reduced. He was resuscitated with fluids and blood. X-rays revealed fractured L3 and left acetabulum. Both were stable and undisplaced. The left psoas shadow was also absent. Ultrasound revealed intra-abdominal clot (mostly retroperitoneal) with no visible spleen. Kidneys were normal. An exploratory laparotomy revealed a huge retroperitoneal haematoma with a smaller collection in left subphrenic space, a tear in the tail of pancreas (which was resected) and an absent spleen. The clots were evacuated. There was nothing significant in the past medical history. There was no history of any haemolytic disorder or infective episodes in childhood.

He had only one daughter and none of her previous investigations suggested asplenia. Echocardiography did not reveal any abnormalities.

DISCUSSION

Asplenia usually occurs as an isolated abnormality (Lindor *et al.*, 1995) or less commonly as recognized syndrome - a rare congenital complex of splenic agenesis, cardiac malformation, malposition of abdominal viscera and various gastrointestinal abnormalities including biliary atresia, duodenal stenosis, imperforate anus, hischurungs disease, hiatus hernia, etc. (Berman, 1982; De Mingo *et al.*, 1992; Hausdorf, 1983; Hutchins *et al.*, 1982; Shirotani *et al.*, 1983; Tunauglu *et al.*, 1991; Wang, 1991; and Wang, 1993).

Congenital isolated asplenia may arise as a minor form of situs abnormalities or result from an unrelated specific defect of spleen development. It is a rare life-threatening condition and pneumococcal sepsis is often the first sign of the disease (Gilbert *et al.*, 2002). Most cases of asplenia are thought to be sporadic but some have suggested the possibility of either an autosomal dominant (Lindor, 1995; Devrient *et al.*, 1994) or recessive (Toriello *et al.*, 1986) inheritance.

This report is probably an isolated abnormality as there were no other defects as excluded by echocardiography, abdominal ultrasound or laparotomy. It is also likely to be a sporadic case since the daughter did not have any anomaly. Studies have shown a higher incidence of sepsis in children in those with congenital asplenia (especially with the syndrome) when compared with the control population (Waldman *et al.*, 1997). Establishing the diagnosis of congenital isolated asplenia in the case of pneumococcal sepsis can be achieved by searching for Howell-Jolly bodies on blood smear, which indicate severe impairment of splenic function and performing ultrasound examination of the abdomen to look for the spleen. In the case of congenital isolated asplenia, use of appropriate antibiotic prophylaxis and (pneumococcal vaccination is recommended (Gilbert *et al.*, 2002).

Sometimes, the first presentation of congenital isolated asplenia can be life-threatening pneumococcal septicaemia and even sudden unexpected death hence the importance of checking for Howell-Jolly bodies in peripheral blood smears (Germing *et al.*, 1999; Kanthan *et al.*, 1999). If this is positive, the patient should undergo further screening with abdominal ultrasound, CT-scan, or scintigraphy which could confirm the presence or the absence of the spleen. The ultrasonic findings of asplenia in this case of abdominal trauma suggested a fractured spleen but laparotomy confirmed it to be a congenital absence. Luckily, this is rare anomaly and should not constitute any problem in diagnosis.

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