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Isolated Congenital Asplenia in an Asymptomatic Patient: A Very Rare Diagnosis

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ABSTRACT

Isolated congenital asplenia is a rare condition that mostly manifests in the early years, usually due to fatal systemic infections. In this paper, however, we present a case of a 36-year-old asymptomatic patient who was referred for suspected hyposplenism, with no history of splenectomy. There were no significant changes on physical examination. Blood analysis revealed leukocytosis and thrombocytosis as well as moderate anisopoikilocytosis and red blood cells with Howell–Jolly bodies. No spleen or other malformations were identified on imaging. Individuals with isolated congenital asplenia have an increased susceptibility to invasive infections and sepsis, with rapid clinical decline and a high mortality rate despite treatment.

LEARNING POINTS

- Isolated congenital asplenia is underdiagnosed in adults and should be excluded in patients with Howell–Jolly bodies in a peripheral blood smear, leukocytosis or/and thrombocytosis.
- Febrile episodes may present initially in these patients with mild symptoms; however, rapid progress to septic shock can occur. As a result, a delay in initiating broad-spectrum antibiotics may compromise their survival.
- Prevention with an individual vaccination plan and patient education is paramount.

KEYWORDS

Isolated congenital asplenia, hyposplenism, sepsis

CASE DESCRIPTION

A 36-year-old female Caucasian patient was referred by the attending physician for a haematology consultation for suspected hyposplenism. There was no relevant past medical history or history of splenectomy. The patient was taking no regular medication. She was asymptomatic and the physical examination appeared normal, namely, without characteristic physiognomic alterations, cardiac auscultation without murmurs, and without palpable masses.

Blood tests revealed mild leukocytosis and thrombocytosis (GB 12,400/μL with 58.6% neutrophils and 31.9% lymphocytes, platelets 491,000/μL) and haemoglobin of 12.9 g/dL. Peripheral blood smear confirmed these findings, as well as marked anisopoikilocytosis with some Howell–Jolly bodies. No haemoglobinopathies (HbA 97.7%) were found.

Given these findings, an abdominopelvic computed tomography (CT) scan (*Fig. 1*) was carried out, which did not identify the spleen. An upper abdominal magnetic resonance imaging (MRI) scan (*Fig. 2*) was performed to clarify this result, confirming absence of the spleen as well as no signs of an infarcted spleen or splenosis. The imaging tests also excluded changes in organ rotation or other abnormalities.

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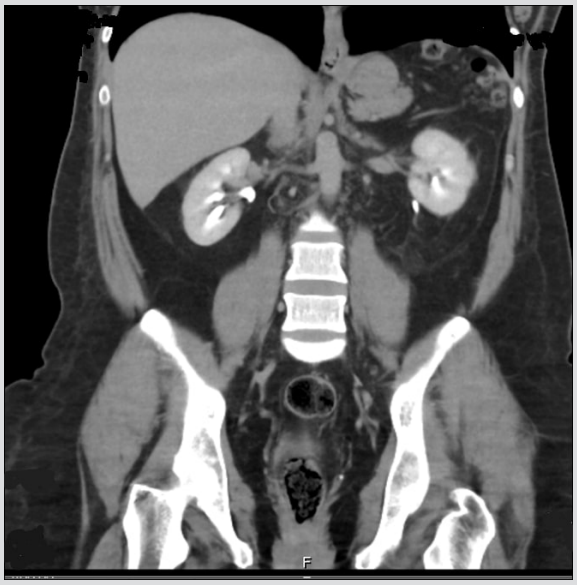

Figure 1. Coronal section of the abdominopelvic CT scan

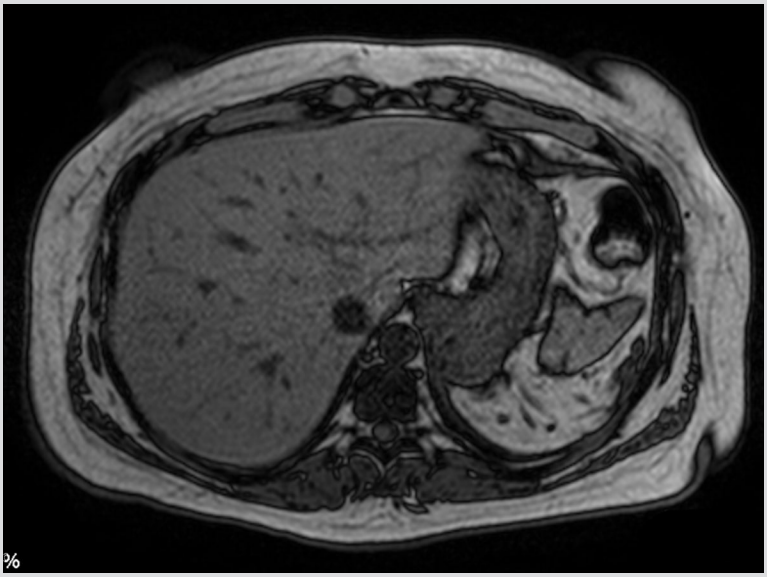

Figure 2. Cross-sectional MRI view of the upper abdomen

The study was completed with transthoracic echocardiography that excluded congenital heart diseases; a myelogram was performed, showing normal bone marrow cellularity, with a maintained myeloid/erythroid ratio.
Thus, the patient was diagnosed with isolated congenital asplenia (ICA) and recommendations were made, namely, an individual vaccination schedule should be put in place and that education about the disease and its warning signs should prompt immediate evaluation.

DISCUSSION

The spleen has a unique structure: it allows the capture of pathogens that are then destroyed by resident macrophages, through phagocytosis and specific cytokine production, and it is the main lymphocytic reservoir, with activated B-cell clonal expansion zones in response to antigens present on the surface of encapsulated bacteria[1].
Asplenia refers to the absence of the spleen, where the most frequent cause is surgical splenectomy[2]. Congenital asplenia is more associated with other abnormalities, specifically Ivemark syndrome, characterised by visceral heterotaxy, with duplication of the right-side organs and absence of the left-side ones, along with high mortality in the first months of life[3].
The diagnosis of ICA becomes a challenge due to the absence of other abnormalities[4]. Despite being extremely rare, it is thought that ICA may be underdiagnosed. Two studies point to an estimated prevalence of 1/600,000 to 0.51 per million births[1, 5].
However, there are few published cases, which complicates an understanding of this disease. A 2017 article identified 73 ICA cases between 1956 and 2016[4]. Of these, 32 were identified as sporadic cases. In 78% of cases, the first manifestation of ICA was severe invasive bacterial infection, with a total mortality rate of 48% and mortality in the first 24 and 48 hours of admission of 58% and 75%, respectively. Of the total episodes of sepsis recorded, the causative agent was identified as Streptococcus pneumoniae in 62% of cases, 21% as Haemophilus influenzae and 7% as Escherichia coli. Regarding familial cases, the transmission pattern is heterogeneous, although dominant transmission with incomplete penetrance seems to prevail[1]. Apart from our case, only three other patients with accidental diagnosis have been described[3, 5].
A 2018 study has linked different mutations in the RPSA gene, which encodes a ribosomal protein, that appears to account for at least 50% of previously described ICA cases[5]. We encouraged our patient to be referred to this study.
Regarding age of presentation, only eight cases were detected in adults[3]. In fact, ICA tends to be more symptomatic and life-threatening during the first 2 years of life, by which time the immune system has not yet peaked. After this period, an immune adaptive response develops, but the underlying mechanism is unknown[2].
The diagnosis should be suspected by the presence of Howell–Jolly bodies, intraerythrocytic remnants normally removed in the spleen, associated with other analytical changes, such as monocytosis, lymphocytosis and/or thrombocytosis[2]. Imaging should be performed to confirm the absence of the spleen (ultrasonography, CT, MRI or splenic scintigraphy with Tc-99m), while congenital haemoglobinopathies

and heart diseases, especially those that affect the pulmonary and venous circulation, should be excluded[1, 2, 4].

Prevention is key to the survival of these patients and can be managed by choosing an individual vaccination plan, especially against encapsulated bacteria. Annual vaccination against the influenza virus is recommended[1]. In addition, patients should be educated about the associated risks (e.g. animal biting and travel to foreign countries) and protective measures. Also, they must be instructed to attend a health facility for observation in case of febrile episodes, for immediate broad-spectrum antibiotic treatment while awaiting the result of the cultures[3,5]. These patients may initially have mild and non-specific symptoms but rapid progression to septic shock and disseminated intravascular coagulation can occur[1–4]. Thus, a treatment delay of hours might even compromise survival[2]. It is believed that this rapid clinical deterioration may be explained by reduced antigen response levels, absence of immunoglobulin M (IgM) memory B cells and by low levels of tuftsin and properdin, two important surface proteins for the promotion of phagocytosis[1–3].

Finally, investigation of the immediate family members of the affected patients, as well as all children or young adults presenting to the emergency department with severe invasive infection, should also be considered[1].

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