Introduction

Asplenia is a rare but clinically important phenomenon because it renders patients highly vulnerable to fulminant sepsis caused by encapsulated organisms. It may occur secondary to splenectomy, or very rarely there is congenital absence of the spleen. The latter is usually associated with interference of the establishment of normal left-right symmetry (situs) during embryogenesis.

Case

A 36 year old Caucasian gentleman presented in shock, with a 2 day history of malaise, and sudden severe epigastric pain with haematemesis on the morning of admission. Born with a univentricular heart, he had undergone childhood cardiac surgery to insert a Fontan circulation, leaving him on lifelong warfarin. He denied any other past medical or surgical history. This hospital did not hold his medical records, as had returned to the country only briefly to visit family here.

On examination, a facial petechial rash was noted. Haematological investigations revealed leucopenia, thrombocytopaenia and severe disseminated intravascular coagulation. After commencement of broad spectrum antibiotics and fluids, he was sent for a CT chest, abdomen and pelvis. This showed no pathology, but revealed right isomerism and absence of a spleen. Howell-Jowell bodies were present on his blood film and he was diagnosed with overwhelming sepsis of unknown cause secondary to asplenia. He deteriorated and, despite intensive care, died 20 hours after admission. After death, polymerase chain reaction showed evidence of a pneumococcal sepsis. His post mortem confirmed Ivemark Syndrome or right atrial isomerism.

Discussion

Situs or ‘position’ abnormalities can broadly be split into inversus abnormalities, or situs ambiguous/heterotaxy resulting in a spectrum of right and left tendencies of the organs. Asymmetry in the body is established during gastrulation, at 3 weeks of gestation. Cilia driven nodal flow is thought to be an essential epigenetic cue that initiates left-right asymmetry. (Figure 2).

Ivemark Syndrome is a heterotaxy resulting in right atrial isomerism, with a combination of bilateral trilobed lungs, midline liver and asplenia, as well as situs inversus affecting other organs. It has been associated with truncating mutations in the growth/differentiation factor-1 gene. Most with the syndrome will not survive beyond the age of one owing to severe cardiac disease. For those who do survive, lifelong prophylactic antibiotics (penicillins or macrolides) and Pneumococcal and Hib vaccinations are required. All patients must be taught to recognise the symptoms and signs of infections and should keep an emergency supply of antibiotics in case of infection. It is important that they carry an alert card/bracelet.

Conclusion

To the best of our knowledge, this gentleman was unaware that he was asplenic. Not all patients will know they are without a spleen, and may not understand its consequences. Sepsis is a common presentation in AAU. It would be prudent for clinicians to consider both the presence of asplenia (especially if there is a history of cardiac abnormality), and risk factors for functional hyposplenism in their assessment of a septic patient.

References

Written consent from next of kin was gained for use of case and image.