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Case Report

Aplastic Anaemia Presenting with Raised Intracranial Pressure Mimicking CNS Leukaemia

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Abstract

Introduction

Patients with aplastic anaemia most commonly present with symptoms of anaemia and skin or mucosal haemorrhage. Visual disturbance due to retinal haemorrhages is much less common.

Case presentation

A 13yr boy presents with an acute history of easy bruising, headaches, vomiting and a deterioration in his vision. He was found to be pancytopenic. On examination there was hepatosplenomegaly and papilledema with bilateral retinal haemorrhages. Urgent neuroimaging revealed raised intracranial pressure but no space occupying lesion. An urgent blood film revealed abnormal cells initially suggestive of acute leukaemia and hence a lumbar puncture performed, and intrathecal methotrexate given. A bone marrow aspirate and biopsy showed a hypocellular marrow with no evidence of leukaemia. A diagnosis of severe aplastic anaemia was made and further investigations including clotting screen, B12 and folate, Hb electrophoresis, cytogenetics, viral serology, immune function and Fanconi screen were all within normal range.

As the cause of his raised intracranial pressure was unknown, he had further neuroimaging including an MRA and lumbar puncture with saline perfusion studies. As these were abnormal, he was diagnosed with benign intracranial hypertension and started on acetazolamide. He also had a further three spinal taps to relief symptomatology.

Management

He continued to require blood product support weekly and was then admitted for antithymocyte globulin (ATG) two months after diagnosis. On day 15 he developed a widespread rash, fever and joint pains and was diagnosed with serum sickness. Hydrocortisone therapy was initiated, and these symptoms quickly resolved. He then started ciclosporin and discharge pending a donor search for potential bone marrow transplantation.

Conclusion

The differential diagnosis of aplastic anaemic includes a prodrome of acute leukaemia. Whilst CNS leukaemia should be considered careful assessment of patient and investigations is required.

Detailed case

Presentation and Diagnostic Work-up

The most common presentation of aplastic anaemia is nose bleeds, gum bleeds or subcutaneous haemorrhages. This was the case for this patient however he also had rapid onset visual loss in his left eye associated headaches and vomiting worsened by lying down. On examination there was no dysmorphology or café-au lait patches suggestive of any neurocutaneous syndrome. He had moderate hepatosplenomegaly and a few scanty small volume lymph glands throughout his cervical chain and groin. Neurological examination revealed bilateral retinal haemorrhages and papilledema. He was found to be pancytopenic with a haemoglobin of 5g/l, total white cell count of 0.6g/l and platelets of only 5. A blood film at the time was difficult to interpret but was felt to have abnormal cells initially suggestive of acute leukaemia. Urgent neuroimaging revealed raised intracranial pressure but no space occupying lesion. A lumbar puncture performed, and intrathecal methotrexate given. The cell count was 357 red blood cells and 7 white blood cells but no evidence of CNS leukaemia. A bone marrow aspirate and biopsy showed a hypocellular marrow with no evidence of leukaemia. A diagnosis of severe aplastic anaemia was therefore made and further investigations including clotting screen, B12 and folate, Hb electrophoresis, cytogenetics, viral serology, immune function and Fanconi screen were all within normal range. These investigations are required to confirm the diagnosis, exclude other possible causes of pancytopenia with a hypocellular bone marrow, exclude congenital aplastic anaemia, screen for underlying cause of acquired aplastic anaemia and document or exclude a co existing abnormal cytogenetic clone or a paroxysmal nocturnal haemoglobinuria (PHN) clone.

As the cause of his raised intracranial pressure was unknown, he had further neuroimaging including an MRA. This showed normal flow signal in the superior sagittal sinus and straight sinus. The transverse sinuses show flow gaps in their anterior segments which is suggestive of raised intracranial pressure without implying a particular cause. There are no features to indicate venous sinus thrombosis.

A lumbar puncture with saline perfusion studies showed abnormally high CSF pressure he was diagnosed with benign intracranial hypertension and started on acetazolamide (Fig 1 and 2). He also had a further three spinal taps to relief symptomatology. After then he had no further headaches and his visual loss stabilised.

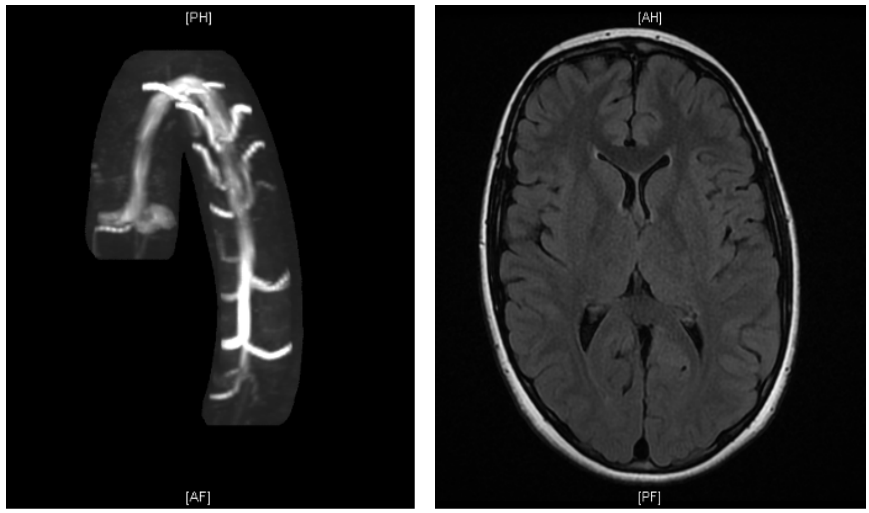

Figure 01 & 02 MRA and MRI respectively

Therapy, Supportive care and complications

In view of his marrow failure, he continued to require blood product support weekly both platelets and red cells. to maintain a safe blood count. Prediction of bleeding is difficult in an individual patient. Fatal haemorrhage, usually cerebral, is more common in patients who have <10 x 109/l platelets, extensive retinal haemorrhages, buccal haemorrhages or rapidly spreading purpura. However, cerebral haemorrhage may be the first major bleed in patients who have none of these other bleeding manifestations (Gordon-smith et al 1991). Apart from platelet transfusion support, other important practical measures to help prevent bleeding include good dental hygiene, the use of oral tranexamic acid and control of menorrhagia with norethisterone. A common problem with multi-transfused patients with aplastic anaemia, compared with leukemic patients, is the development of alloimmunization to leucocytes present in red cell and platelet transfusions by generating HLA or non-HLA antibodies. Prior to the introduction of leucocyte depletion in the UK around 50% of patients with aplastic anaemia developed alloimmunization (Killick et al 1997). As yet our patient ahs not had reactions to blood products.

Immunosuppressive therapy using the combination of ATG and ciclosporin associated with response rates of between 60 – 80% with current 5-year survival rates of around 75% (Bacigalupo et al 2000). Response to ATG and ciclosporin is delayed and response usually does not start much before 3 months. In the UK the standard preparation of ATG is horse ATG although rabbit is also used. It is given over five days as a daily infusion over 18 hours. The immediate side effects include fever, rigours, rash and hypotension.

For this patient rabbit antithymocyte globulin (ATG) was administered two months after diagnosis. On day 15 he developed a widespread rash, fever and joint pains and was diagnosed with serum sickness.

Hydrocortisone therapy was initiated, and these symptoms quickly resolved. He then started cyclosporin and discharge pending a review with regards to response to ATG therapy. The response to ATG can be several months.

Key learning points

1. Diagnostic workup of children with bone marrow failure
2. Unusual presentation and association of aplastic anaemia with raised intracranial pressure
3. Treatment strategies for aplastic anaemia and rationale

Medico-legal implications

Treating a patient with chemotherapy without a confirmed cancer diagnosis raises significant medico-legal concerns because it violates core standards of medical practice, informed consent, and patient safety. Chemotherapy involves inherently toxic agents with well-known risks such as myelosuppression, organ damage, secondary malignancies and in the case of intrathecal chemotherapy neurological toxicities. Clinically, physicians are expected to establish a clear, evidence-based rationale supported by pathology, imaging, or molecular testing before initiating such high-risk treatment. Legally, initiating chemotherapy without adequate diagnostic confirmation may expose clinicians and institutions to claims of negligence, malpractice, or battery, particularly if the patient experiences preventable harm. Understandably in this case presented there was an urgency due to loos of vision to think quickly and proceed at risk however standard protocols, guidelines need to be adhered to avoid diagnostics and therapeutic errors. Overall, deviation from diagnostic standards places the clinician at substantial legal, professional, and ethical risk.

References

1. Acquired aplastic anaemia. In: Haematology. Basic Principles and Practice (ed by R Hoffman)
2. Pilot study of HLA alloimmunisation after transfusion with pre-storage leucodepleted blood products in aplastic anaemia. Lillick et al 1997 BJH 97, 677-684
3. Guidelines for the diagnosis and management of acquired aplastic anaemia. BJH 2003 123, 782-801.