CASE REPORT

Juvenile polymyositis with unremitting pain and progressive loss of motor and bulbar function on a background of sickle cell disease

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SUMMARY

The diagnosis of acute autoimmune rheumatic disorders in sickle cell disease (SCD) can be challenging. Polymyositis is an inflammatory myopathy which, like SCD, may present with myalgia but is usually associated with proximal muscle weakness. We describe an adolescent boy presenting with limb pain, difficulty in mobilisation, with progressive loss of motor function and later bulbar weakness. Investigations showed massive elevation of creatine kinase, and MRI and muscle biopsy findings consistent with severe polymyositis. The patient was treated with corticosteroids, intravenous immunoglobulin and intensive rehabilitation therapy. He made a good recovery and was discharged on azathioprine and prednisolone. In the context of SCD, multisystem symptoms, unexplained muscle pain and weakness, unresponsive to conventional treatment in the presence of steady state haemoglobin, should alert the clinician to autoimmune phenomena. Key factors in making a diagnosis are an autoimmune screen and early discussion with a rheumatology expert.

BACKGROUND

Sickle cell disease (SCD) arises from inheritance of two abnormal β -globin chains, one of which is due to haemoglobin (Hb) S mutation (a substitution of valine for glutamic acid in the sixth amino acid of the β -globin gene).¹ Unlike the homozygous state of Hb S (sickle cell anaemia), double heterozygote states such as the combination with C (HbSC) patients tend to have a higher Hb level and generally have a less severe phenotype.¹ Affected Hb polarises in the presence of low oxygen tension resulting in polymers that disrupt red blood cell architecture and flexibility, causing microvascular obstruction, tissue ischaemia and acute pain.

Juvenile idiopathic inflammatory myopathies (JIIM) are fivefold less common than adult IIM, and dermatomyositis is the most common presentation in children (median age 7.5 years). Juvenile polymyositis (JPM) is seen in 4–8% of JIIM, and occurs mainly in teenagers who often have very high creatine kinase (CK) levels. As a result of muscle weakness, they present with frequent falls and their condition may be confused with other non-inflammatory myopathies such as muscular dystrophies. A muscle biopsy is therefore essential for the diagnosis of JPM.²

Autoimmune diseases may present with signs and symptoms similar to acute crises in SCD with case

Figure 1 MRI of lower legs: axial mid-thigh showing inflammation of all muscle groups.

reports highlighting the diagnostic challenges should the two conditions coexist.^{3 4}

To the best of our knowledge, this is the first case of SCD associated with JPM and highlights key clinical features that should alert the clinician to the possibility of autoimmune phenomena as a cause for atypical symptoms in SCD.

CASE PRESENTATION

A 15-year-old previously healthy boy with HbSC presented with a 4-week history of feeling generally unwell. The initial trigger was a febrile upper-

Figure 2 Flair images showing diffuse high signal through both lateral pterygoids and both temporalis muscles.

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respiratory tract illness, which was treated with fluoxacillin in the community. Over the next fortnight the patient developed increasing weakness and pain in upper and lower limbs, which was not relieved by paracetamol or non-steroidal anti-inflammatory agents. He developed progressive loss of muscle power in the lower limbs and was unable to walk or dress independently. Examination revealed marked proximal weakness of the lower limbs, more pronounced on the left side, with normal muscle tone, reflexes and sensation. There were no localising neurological signs and no bladder, bowel or bulbar symptoms

on admission. The patients had no skin rashes. He was initially managed for acute vaso-occlusive painful crisis and discharged home with crutches. Nevertheless, he insisted that the pain was different to the 'usual sickle pain' he experienced during a crisis.

Prior to this presentation he had no previous admissions with vaso-occlusive crises and had never required a transfusion. He had a subaortic stenosis, diagnosed at birth with surgical resection at age 7. His only treatment was a standard prophylactic regimen of penicillin and folic acid. He attended school regularly, and was

Figure 3 Electroneurography report.

academically and athletically proficient. He was not functionally limited before this admission.

INVESTIGATIONS

On admission, the patient's Hb was 10.7 g/L (consistent to his steady state), with a normal white cell count and C reactive protein (<5) (0–4mg/L). Urea and electrolytes were unremarkable. Liver function showed deranged liver enzymes (alkaline phosphatase and aspartate transaminase), erythrocyte sedimentation rate was raised at 23 mm/Hr and CK on admission was 34 821 IU/L international units.

On admission, an extensive infection screen showed positive nasal swabs for enterovirus/rhinovirus, positive cytomegalovirus (CMV) IgM and an elevated aspartate transaminase. A CMV viral load was negative.

MRI showed evidence of an inflammatory myopathy in the lower legs (figure 1) and in the facial muscles (figure 2). An electromyogram of tibialis anterior (figure 3) showed nerve conduction within normal limits. A muscle biopsy showed necrotic fibres and human leukocyte antigen class 1 overexpression, consistent with polymyositis.

DIFFERENTIAL DIAGNOSIS

Differential diagnosis included

- ▶ Vaso-occlusive sickle crisis
- ▶ Guillain-Barre Syndrome
- ▶ Myositis triggered by an infectious process (eg, CMV induced myositis)
- ▶ Autoimmune polymyositis

TREATMENT

On admission, the patient was started on a moderate dose of prednisolone, 20 mg, once a day.

OUTCOME AND FOLLOW-UP

On day 4 of admission, the patient developed nasal speech and had difficulty swallowing, symptoms consistent with bulbar weakness. His medication was changed to high-dose intravenous methylprednisolone 500 mg×3 doses, and he was kept nil by mouth and a nasogastric tube was inserted. On reassessment 1 day later there was persistence of bulbar symptoms with

evidence of ongoing nasopharyngeal regurgitation. In light of the progressive symptoms, intravenous immunoglobulin (IVIG) 0.4 g/kg/day for 5 days was given. MRI demonstrated extensive inflammatory changes on T2 imaging in the facial muscles (figure 1).

The patient's symptoms markedly improved, with a fall in CK after the course of IVIG subsequent to which the steroids were tapered and immunomodulating therapy (azathioprine) was started.

The patient was discharged a month after initial admission and has made a complete recovery.

DISCUSSION

Our report illustrates the importance of considering an autoimmune rheumatic disorder in the presence of SCD. The clinical presentation of severe painful muscle weakness associated with the development of bulbar symptoms shows the importance of early diagnosis and treatment in achieving a good outcome. A diagnosis of coexistent JPM was delayed because the symptoms were initially thought to be attributable to a SCD crises and the patient did not have the skin lesions typical of dermatomyositis.

Autoimmune rheumatic disorders reported in SCD include systemic lupus erythematosus,^{2 3} juvenile idiopathic arthritis⁵ and dermatomyositis.⁶ The diagnosis of inflammatory autoimmune conditions in SCD can therefore be delayed³ and clinicians should be vigilant of these possibilities. Early intervention may enhance patient recovery, resulting in earlier return to school and full physical function.² To the best of our knowledge, this is the first report of SCD associated with JPM.

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Competing interests None.

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Learning points

- ▶ Prolonged atypical pain associated with significant weakness in sickle cell disease should alert the clinician to consider alternative diagnoses, particularly autoimmune rheumatic disorders.
- ▶ In the presence of normal haemoglobin, an unremarkable white cell count and C reactive protein, an autoimmune screen, including creatine kinase, should be considered.
- ▶ Ideally, these patients should be managed in conjunction with an experienced sickle cell and rheumatology team.

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