after a rehabilitative procedure. This was a 5-year-old girl with excellent sitting balance and discrete walking autonomy who had severe upper extremity tremor and spasticity one year after a vigorous programme of neck and head exercise. She became hemiparetic after sedation for neurological studies (electroencephalogram and neuroimaging). She partially recovered after surgical stabilisation (Gallie fusion).

Our patient had progressive neuromotor degeneration after 12 months of an intensive programme of neck, head and trunk exercise; her symptomatology, which rapidly progressed, confining her to a wheelchair and finally to bed, was almost certainly triggered by physical therapy procedures, as a radiograph obtained before her enrolment in the special school showed a normal atlantoaxial gap in all neck positions. Several studies have demonstrated that radiological status can change over time because of variations in atlantoaxial instability.1

The aforementioned case reports2-5 indicate that many patients with symptomatic AAI may have symptoms and signs of cervical spinal cord compression for weeks or years before they are recognised as having neurologic disease. Current evidence suggests that these neurological abnormalities may be more predictive of potential progressive injuries than the abnormalities or radiography themselves in asymptomatic patients.

Final diagnosis

Atlantoaxial dislocation with acute symptomatic compression to the spinal cord secondary to physical therapy procedures in a child with Down’s syndrome and atlantoaxial instability.

Keywords: Down’s syndrome; atlantoaxial instability; spinal cord compression; neurologic complications; rehabilitative procedures


Fever, thrombocytopenia, and diarrhoea

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A 40-year-old man was admitted to hospital with a 10-day history of fever, malaise, profuse diarrhoea and intermittent macular rash affecting the chest and legs. On arrival, he had a fever of 39.5°C, blood pressure 100/70 mmHg and pulse rate 110 beats/min. There was no lymphadenopathy. The abdomen was tender but not distended and the rectum empty and non-tender. Admission blood count revealed haemoglobin 9.8 g/dl, total white cell count 1.4 x 10⁹/l and platelets 74 x 10⁹/l. The rectum appeared inflamed at sigmoidoscopy and rectal biopsy revealed a mild inflammatory infiltrate. Barium enema was unremarkable. The patient was treated empirically for bacterial gastroenteritis with intravenous ciprofloxacin but a series of stool and blood cultures were negative and over the next 10 days he continued to have a high swinging fever and between 10 and 20 loose stools daily. Empirical steroid therapy (prednisolone 30 mg/day) was followed by a gradual improvement in his condition and at the time of discharge the diarrhoea had lessened and his haemoglobin was 10.5 g/dl, total white cell count 4.8 x 10⁹/l and platelets 239 x 10⁹/l. When reviewed three months later he was taking 10 mg prednisolone daily but his weight had fallen by 12 kg with intermittent severe diarrhoea. Increased steroid dosage did not alter his symptoms. Colonoscopy revealed no abnormality and colonic biopsies were normal. Serial stool cultures were again negative and microscopy revealed no ova, cysts or parasites. Blood count showed a haemoglobin of 11.9 g/dl, white cell count 3.9 x 10⁹/l and platelets 63 x 10⁹/l.

Questions

1 What acute infection might explain the thrombocytopenia and diarrhoea?
2 Which investigations would confirm the diagnosis?
3 What treatment should be offered?
Answers

QUESTION 1
Acute primary human immunodeficiency virus (HIV) infection (HIV seroconversion illness or stage I HIV infection)

QUESTION 2
The clinical diagnosis can be confirmed by polymerase chain reaction (PCR) detection of HIV in almost all patients with an acute seroconversion illness. The HIV antibody is less reliable and may not become detectable until several days or weeks after the onset of a seroconversion illness or when the symptoms of acute infection have declined. This patient had a negative antibody test when he first presented but a positive test three months later.

QUESTION 3
Combination antiretroviral therapy is now being offered to patients with acute HIV infection – a development which underlines the importance of early diagnosis.

Discussion

The illness associated with primary HIV infection was first described as a clinical entity in 1984, following the infection of a nurse from a needle-stick injury. Subsequent reports have indicated the disease's protean manifestations, the most typical of which is an acute mononucleosis-like illness with fever, rash, lymphadenopathy, arthralgia and pharyngitis.

In contrast to the other mononucleoses, gastrointestinal symptoms are often present, with diarrhoea reported in over 30% and oral ulceration in 13% of patients. The duration of a seroconversion illness is rarely longer than three weeks and this patient's symptoms ran an unusually protracted course. A prolonged or severe seroconversion illness may be associated with a rapid progression to acquired immunodeficiency syndrome (AIDS) and death.

The absence of any opportunistic infection in our patient, together with the ultimate spontaneous resolution of his symptoms, would suggest that the diarrhoea was a feature of his primary HIV infection. This patient's presentation indicates the need to include HIV seroconversion illness in the differential diagnosis of chronic, noninfectious diarrhoea, weight loss and unexplained thrombocytopenia. The diagnosis of acute HIV infection may be confirmed by PCR detection of the virus several weeks or months before the development of antibodies. Early diagnosis may be of importance as there is evidence to suggest benefit from antiretroviral treatment at the onset of HIV infection.

Final diagnosis

Acute primary human immunodeficiency virus infection.

Keywords: fever; thrombocytopenia; diarrhoea; human immunodeficiency virus infection

8 Niu MT, Stein DS, Schnitman SM. Treatment trials for primary human immunodeficiency virus type 1 infection. J Infect Dis 1993;168:1601–2.
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