Postmortem examination unexpectedly showed concurrent cytomegalovirus pneumonia. This is not uncommonly seen in immune suppressed patients. Cytomegalovirus was not grown from the maternal cervix, and although the possibility that the infection was acquired from one of several top up transfusions was considered, this could not be confirmed.

In any neonate with non-responsive neonatal pneumonia, congenital tuberculosis should be considered.

We thank Dr Maeve Keaney for constructive advice, the Department of Medical Illustration at Hope Hospital, and Jacqueline Buckley for secretarial help.

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Received 4 February 1986

Specific malabsorption of vitamin B₁₂ in Down's syndrome

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SUMMARY A 3 year old girl with Down's syndrome became lethargic and withdrawn, and investigations showed a specific malabsorption of vitamin B₁₂ without proteinuria.

Megaloblastic anaemia due to specific malabsorption of vitamin B₁₂ is usually associated with proteinuria. Urban *et al* reviewed the published reports in 1981 and found only six cases without proteinuria,¹ and Conway *et al* reported a further case with neuropathy but without proteinuria.² There is often delay in diagnosing a new problem in a child with Down's syndrome, and we report this association to emphasise the need to take seriously the development of new symptoms, rather than attributing them to the syndrome.

Case report

A 3 years and 4 months old girl of unrelated parents who was under regular review because she had Down's syndrome presented with new symptoms developing over one month. She suffered one episode of unproven haematuria followed by two weeks of watery diarrhoea. She then developed diurnal enuresis, cold cyanosed peripheries, leth-

argy, irritability, anorexia, and a scaly erythematous non-itching rash over an elbow and became withdrawn. She had lost 350 g in six months, her weight being 9650 g (2000 g below the third centile). Her height was just below the third centile with a normal height velocity. On examination she had typical features of Down's syndrome and, in addition, was pale and miserable and had an area of discoid eczema over her left elbow.

The following investigations yielded normal results: plasma urea, creatinine, electrolytes, calcium, phosphate, albumin, glucose, bilirubin, and thyroxine concentrations, alkaline phosphatase, alanine transferase, and γ -glutamyltransferase activities, urine analysis and culture, throat and cough swabs, faecal microscopy and culture, and chest x ray films.

The peripheral blood film showed a pancytopenia with macrocytic normochromic red cells (haemoglobin=5.9%, mean corpuscular volume 107.6 fl, mean corpuscular haemoglobin 35.9 pg, white cell count $3.41 \times 10^9/l$, neutrophils $0.550 \times 10^9/l$, platelets $47 \times 10^9/l$). Erythrocyte sedimentation rate was 9 mm in the first hour. The bone marrow was megaloblastic. Serum total cobalamins were 80 ng/l (normal range 300-1100 ng/l), red cell folate 240 mcg/l (normal range 130-600 mcg/l), and serum folate 15.4 mcg/l (normal range 2.6-14 mcg/l). Intrinsic factor antibody, parietal cell antibody, and

autoantibody screen on serum yielded negative results. Intrinsic factor type I (blocking) antibody and parietal cell antibody were not detected in gastric juice. Resting gastric juice pH was 2, but difficulties in collection of gastric juice prevented a satisfactory pentagastrin test. A Schilling test showed intestinal malabsorption of vitamin B₁₂ with 3.56% of free ⁵⁸CoB₁₂ and 3.06% of intrinsic factor bound ⁵⁷CoB₁₂ excreted into urine over 24 hours. Plasma concentrations of ⁵⁷CoB₁₂ and ⁵⁸CoB₁₂ were assayed between 7 and 11 hours after oral administration, and both showed a peak concentration of 30% of the predicted normal. Repeated testing for proteinuria (now 19 months after presentation) has yielded negative results. Stool microscopy did not show parasites (including fish tapeworm) or fat globules. Transcobalamin concentration was normal. Urinary indican was low. Barium study of the stomach and small bowel yielded normal results, apart from showing a hiatus hernia.

She has been treated with five 1 mg doses of hydroxocobalamin followed by 1 mg every other month. She made a rapid haematological response, with a normal haemoglobin and blood film one month after starting treatment. All her symptoms have disappeared and she has gained 2500 g in six months.

Discussion

The diagnosis of specific malabsorption of vitamin B₁₂ was confirmed by the low serum vitamin B₁₂, impaired intestinal absorption of vitamin B₁₂, the presence of normal gastric acid, and the absence of intrinsic factor antibody and parietal cell antibody in serum and gastric juice. The rapid clinical response to treatment with vitamin B₁₂, the lack of

steatorrhoea, the normal small bowel barium study, and the low urinary indican make more general malabsorption unlikely, and further investigation was not thought to be justified.

Specific malabsorption of vitamin B₁₂ was described by Imerslund³ and has an autosomal recessive inheritance. It is associated with proteinuria in 90% of cases,⁴ but proteinuria may not occur for 18 months.⁵ Small bowel electronmicroscopy and histology is normal,⁴ and aetiology is unknown. Diagnosis is made by showing intestinal malabsorption of vitamin B₁₂ in the absence of general malabsorption.

Doctors tend to stereotype Down's syndrome children,⁶ symptoms of lethargy and mental slowness being interpreted as part of the syndrome. It is important to consider other causes and to be as assiduous in tracking down the diagnosis as one would be for otherwise normal children.

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Received 13 January 1986

Neonatal malaria and obstructive jaundice

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SUMMARY A case is presented of transplacental malaria in an infant associated with persistent conjugated hyperbilirubinaemia and liver disease.

Malaria presenting in the neonatal period is a rare disease even in endemic areas.¹

Case report

A female term infant was born in Bradford, England, weighing 2800 g to Indian parents of a consanguineous marriage who had two other healthy children weighing 5600 g at term and 1700 g at 32 weeks. The mother had spent four months of the pregnancy in India, and two days after delivery a