Repeated fits occurred in 32 children in this study; in 13 before hospital admission. The repeated fits were of short duration (mean 7.75 minutes, range 0.75 to 20). The incidence of repeated fits (16.1%) compares with 16.2% in the study by Nelson and Ellenberg.¹

Diagnosis of the cause of fever was helped by our active investigative policy of excluding meningitis. Although we are aware of the differing views on the value of routine lumbar punctures in studies reviewed by Kudrjavcev,² our policy has been adopted as a safety measure in a hospital with relatively inexperienced resident staff. The results of this study suggest that it is safe to discharge children (even if still febrile) when they have been observed for 24 hours after a febrile convulsion—provided, of course, they are reasonably well in themselves and a

diagnosis of cause of the fever established. The risk of recurrent febrile convulsions 24 hours after admission seems to be very low (0/199).

We thank Dr C S Livingstone for his permission to report details of some of his patients in this study.

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Type V glycogen storage disease

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SUMMARY We describe three children with type V glycogen storage disease, who were reluctant to climb hills. We suggest that this condition, usually described as being of adult onset, can often be diagnosed in childhood.

Myopathy associated with defective glycogen breakdown was first described by McArdle in 1951.¹ Absent muscle phosphorylase activity was subsequently shown.² Standard texts suggest that the condition rarely presents in childhood and diagnosis is usually established in adolescence or later. We describe three children with type V glycogen storage disease, one diagnosed at the age of 8 years and two at the age of 10.

Case histories

Case 1. An 8 year old girl was referred with a four year history of disinclination to walk more than a short distance, particularly uphill. Short rests seemed helpful in allowing her to continue. She had always been considered clumsy as a small child and fell often.

Examination showed a mild tremor of both hands and pain with contraction of finger flexors within 20

seconds of beginning exercise. Creatine kinase activity was raised at 1500 IU/l and 3234 IU/l on consecutive measurements. Standard test for ischaemic lactate concentrations showed an inappropriately low rise in blood lactate concentration. No muscle phosphorylase activity was detected by muscle biopsy.

Case 2. This 10 year old girl had a long history of slow walking (particularly uphill) and associated leg cramps. Physical examination was unremarkable. Creatine kinase activity was raised initially at 7548 IU/l, though it was subsequently measured at 374 IU/l. Test for ischaemic lactate concentration showed no appreciable rise in blood lactate values, and no muscle phosphorylase activity was detected by muscle biopsy.

Case 3. A 10 year old girl was referred with a long history of pain and weakness below the knee. This had become more pronounced in the preceding 18 months, and she needed rests on walking any great distance. No abnormalities were noted on examination. Creatine kinase activity was raised at 650 IU/l, and tests for ischaemic lactate concentration showed no appreciable rise in blood lactate values. Muscle phosphorylase activity was absent in the muscle biopsy.

Methods

Ischaemic lactate concentration test. Patients fasted for 12 hours before the test and remained at rest throughout. A cannula was introduced into a forearm vein and heparinised. Two resting samples of venous blood were taken for estimation of lactate concentration and creatine kinase activity. A sphygmomanometer cuff was then inflated to 200 mm Hg and the patient instructed to squeeze the bulb of a second unattached cuff once per second for at least 30 seconds followed by a rest period of two minutes. The cuff was then released and further venous samples for estimation of lactate concentration and creatine kinase activity were taken immediately and after two, four, and 10 minutes after cuff release. The final sample was taken two hours after cuff release. For estimation of lactate concentration 1 ml of blood was mixed with an equal volume of ice cold, 15% perchloric acid immediately after sampling, and was transferred to the laboratory. Lactate concentration was measured using a standard nicotinamide-adenine dinucleotide linked spectrophotometric assay adapted for the Cobas Bio centrifugal analyser. Creatine kinase activity was measured at 37°C using an optimised kit method (Boehringer Diagnostics, Mannheim, Germany) in a centrifugal analyser (Cobas Bio, Roche Diagnostics, Welwyn Garden City).

Phosphorylase activity. Determination of muscle phosphorylase activity was by the technique described by Godlewsky.³

Discussion

The existence of a group of inherited enzyme defects involving glycogen metabolism in the liver or muscle or both is now well recognised. Von Gierke's disease was the first in which the specific enzyme deficiency was identified. Traditionally, glycogen

storage diseases (as they became known) were classified by the allocation of a number, but many prefer a classification based on the identified absent enzyme with or without the appropriate eponym.

Muscle phosphorylase activity, absent in type V glycogen storage disease, catalyses the reaction that breaks down intracellular glycogen. During exercise consumption of adenosine triphosphate increases dramatically. Glycogenolysis is important in providing additional adenosine triphosphate, and failure of satisfactory glycogenolysis may become clinically evident during exercise.⁴

Type V glycogen storage disease may present in a variety of ways in childhood. It is characterised typically by exercise related muscle pain and cramps, which resolve quickly on stopping the exercise. In young children pain may be less obvious—rather a disinclination to walk more than a short distance, particularly uphill. Weakness is often, but not always, present. Surprisingly, two of our patients (cases 2 and 3) were able to sprint well for short distances without discomfort.

Diagnosis is established by reduced production of venous lactate concentrations after ischaemic exercise—the result of failure of glycogenolysis and glycolysis—together with absent muscle phosphorylase activity by muscle biopsy (Table). Biopsy is generally considered mandatory for confirmation. Recently, it has been suggested that the phosphorous Nuclear Magnetic Resonance technique could establish the diagnosis of type V glycogen storage disease both reliably and non-invasively by monitoring changes in tissue pH and phosphocreatinine concentration in response to exercise.^{5 6} Phosphofructokinase activity deficiency (glycogen storage disease type V11, Tauri's disease) may have identical though usually milder presenting features to deficiency of muscle phosphorylase activity, and there is also poor lactate response after exercise. Myoglobinuria, seen in many adults with type V glycogen storage disease, is not essential for diagnosis

Table Creatine kinase activity and lactate concentrations before and after exercise test in three patients with type V glycogen storage disease

Case no	Creatine kinase activity (IU/l) ¹							Lactate concentrations (mmol/l) ²						
	Time before cuff release (min)		Time after cuff release (min)					Time before cuff release (min)		Time after cuff release (min)				
	5	0	0	2	4	10	120	5	0	0	2	4	10	
1	395	390	395	395	395	395	943	0.52	0.50	0.51	0.75	0.74	0.76	
2	267	280	289	280	267	267	250	1.43	1.48	0.89	1.25	0.81	0.63	
3	489	489	489	503	499	492	1536	0.69	0.66	0.82	0.78	0.71	0.77	

¹ Normal activities (at rest) 24–195 IU/l with no significant rise after exercise test.

² Normal concentrations (fasting) 0.56–1.5 mmol/l. Expected rise in normal subjects of two to threefold two minutes after cuff release.

sis. The dramatic variation in values of creatine kinase activities in case 2 was unexplained. It was not obviously related to varying degrees of exertion before testing. This phenomenon has been described previously and could lead to diagnostic confusion. The condition is generally regarded as inherited on an autosomal recessive basis, but there is evidence to suggest genetic heterogeneity.

These three patients' symptoms, dating from much earlier in childhood, confirm that type V glycogen storage disease may be diagnosed at an earlier age than often reported. In all three symptoms were related to a disinclination to walk up hills. No more specific symptoms than this occurred within the histories. The test for ischaemic lactate concentration is an important and fairly straightforward procedure that should be considered in a child presenting with any of the features detailed in this report. An earlier diagnosis may be particularly important if the recent preliminary report of improvement in symptoms related to a high protein diet is confirmed in other patients.⁷

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Eosinophilic gastroenteritis

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SUMMARY We report two children with eosinophilic gastroenteritis—a 14 month old atopic boy with persistent vomiting and aspiration pneumonitis illustrates the mucosal variety of the disorder, and a 9 year old boy with eosinophilic ascites typifies serosal involvement.

Eosinophilic infiltration of the gut wall may affect the mucosa, muscularis layer, or the serosa. Each has a distinctly different clinical presentation and the two children we now report illustrate some features of the clinical spectrum.

Case reports

Case 1. This boy was fully breast fed and thrived until he was 6 months old. Two weeks after introducing cows' milk and various solid foods he

developed persistent vomiting and subsequent poor weight gain. Physical examination, chest radiograph, and urine culture were normal. Barium meal at the age of 11 months showed mild gastro-oesophageal reflux but no obstruction. Neither milk exclusion nor 'Gaviscon' improved his vomiting. At age 14 months he collapsed after vomiting and at bronchoscopy food material was removed from both main bronchi. Mechanical ventilation, together with antibiotics and hydrocortisone, were required to treat his aspiration pneumonitis. On reintroducing a normal diet, vomiting recurred. Further questioning revealed two previous episodes of wheeze after vomiting and also facial and lip swelling after certain foods (apple, rhubarb, baked beans, and egg). His mother and maternal grandmother have asthma.

A peripheral eosinophilia ($3.09 \times 10^9/l$) was present on hospital admission and persisted. His serum IgE concentration was 1890 IU/l (normal less than 10), and specific IgE antibodies to milk, egg