subsequent delay in motor milestones, and her 10-year-old sister with bilateral pes cavus and talipes equinovarus, otherwise symptom-free, who was also found to have facial weakness and EMG evidence of myotonia; two floppy infants with delayed milestones whose related mothers have a subclinical (almost) involvement; and a 6-year-old boy who presented with bilateral facial palsy and whose symptom-free mother and grandfather have EMG evidence of myotonia.

The typical EMG pattern is readily missed, unless carefully searched for in distal as well as more proximal muscles. Muscle biopsy may be histologically normal in these children, but may show selective type 1 fibre atrophy with histochemical enzyme reactions.

Late onset form of globoid cell leucodystrophy. Desmond Patrick and John Wilson (introduced by John Wilson). (*Department of Biochemistry, Institute of Child Health, 30 Guilford Street, London WC1N 1EH.*)

Elevated erythrocyte 2,3-diphosphoglycerate concentration in primary trisomic Down's syndrome. Elizabeth Nelson and Philip F. Benson. (*Department of Clinical Haematology, University College Hospital Medical School, London W.C.1, and Paediatric Research Unit, Guy's Hospital, London S.E.1.*) The level of 2,3-diphosphoglycerate (DPG) in erythrocytes is an important factor in regulating oxygen delivery to the tissues. It does this by binding to deoxyhaemoglobin and has the effect of lowering the haemoglobin affinity for oxygen. DPG is a glycolytic intermediate and its concentration is dependent upon the activity of the enzymes in the pathway. In Down's syndrome there is a marked increase in the activity of erythrocyte phosphofructokinase, a unidirectional enzyme which plays an important role in the regulation of glycolytic rate.

We have therefore investigated the possibility of a concomitant rise in erythrocytic DPG concentration.

The DPG level was higher in 20 subjects with primary trisomic Down's syndrome (12 males, 8 females, mean age 17.6 years, range 9 to 26 years; mean $5.37 \mu\text{moles/ml RBC}$; SD 0.625) than in 20 matched controls (mean $4.32 \mu\text{moles/ml RBC}$; SD 0.368 ; $P < 0.001$).

Changes in DPG concentration may be influenced by red cell pH and conditions of hypoxia as well as glycolytic activity. Anaemia was excluded in our patients (mean PCV = 43.1% ; SD 3.4 ; mean Hb concentration 14.7 g/100 ml ; SD 1.498).

One can calculate that the observed increase in DPG concentration would produce a 14% increase in P_{50} when associated with Hb-A. Though there would not be such a significant difference with Hb-F we do not know the effect of DPG on embryonic Hbs and further studies are necessary to determine the DPG level in Down's fetuses and to assess any possible effects that changes might have on fetal growth and development.

Creatine phosphokinase (CPK) in the CSF: its value in the management of children with

myelomeningocele and hydrocephalus. Margaret B. Drummond and Neville R. Belton (introduced by J. Keith Brown). (*Department of Child Life and Health, and Royal Hospital for Sick Children, Edinburgh.*) A need exists for a reliable test which will indicate 'brain damage' during the course of acute or chronic neurological disease. A number of studies have previously investigated the level of enzymes and other suitable substances in the CSF.

Creatine phosphokinase is present in high concentration in brain as well as in skeletal and heart muscle. Previous studies (Herschkowitz and Cumings, 1964; Nathan, 1967) have suggested increased CPK activity in a number of neurological diseases, particularly in patients with progressive hydrocephalus and symptomatic epilepsy. Sherwin, Norris, and Bulcke (1969) have shown that CSF contains only the brain isoenzyme of CPK.

In this study, which is part of a wider investigation of CSF-CPK in children with neurological disorders, all 65 children studied had a myelomeningocele and hydrocephalus. CPK was estimated, along with routine bacteriological and biochemical estimations, and pressure measurements taken whenever a ventricular tap was indicated clinically. CSF was withdrawn during the investigation and treatment of increased intracranial pressure, ventriculitis, and blocked shunts. Serum CPK levels were estimated concurrently in 11 cases.

The main findings were: (1) No correlation was found between serum and CSF levels of CPK, or between CSF-CPK and protein levels or pressure readings. (2) Newborn infants with myelomeningocele have increased levels of CPK in CSF. (3) Increased CSF-CPK levels are also found in most cases of raised intracranial pressure and in ventriculitis where they tend to parallel the clinical course of the infection. (4) CPK levels in the myelomeningocele lesion fluid were much higher than those in ventricular CSF on concurrent specimens.

Thus there are indications that CSF-CPK determinations can be useful in the management of blocked or malfunctioning shunts but may not add additional information in the management of ventriculitis.

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Fat absorption in children with chronic liver disease.* J. F. T. Glasgow (introduced by I. J. Carré, Belfast). The digestion and absorption of dietary fat in 15 children (aged 2-90 months) with varying degrees of chronic liver disease has been investi-

*Carried out in co-operation with Drs. J. R. Hamilton and A. Sass-Kortsak, The Hospital for Sick Children, Toronto, Canada.