Propionicacidaemia in twins. F. Cockburn, M. D. Cohen, J. A. B. Darling, M. Giles, R. A. Harkness, and A. D. Nicol. (*Departments of Child Life and Health and Paediatric Biochemistry, University of Edinburgh, and Simpson Memorial Maternity Pavilion, Royal Infirmary, Edinburgh.*) A few infants with a serious metabolic disease associated with increased concentrations of propionic acid in the plasma have been reported. The majority of these infants have died in the neonatal period. Ketoacidosis and hypotonia have been presenting features. We describe such a syndrome in nonidentical twins.

The parents are first cousins and come from Pakistan. The mother's 4 previous pregnancies had resulted in an abortion, an anencephalic stillbirth, and two children who died in the neonatal period. One of these children was reported by Gompertz *et al.* (1970) to have had propionicacidaemia.

During pregnancy the mother's urinary oestriol concentrations had been consistently low. Labour occurred spontaneously at 37 weeks. Amniotic fluid from each sac and umbilical venous plasma from each twin had normal concentrations of amino acids and propionic acid. Twin I, a girl, weighed 1.7 kg, and twin II, a boy, 2.7 kg. Both followed an almost identical course of progressive hypotonia, apathy, and diminished reflexes, merging into coma and death. Twin I died on day 10 and twin II on day 13. Long chain ketones and acetone were present in their urine from the first day of life, in association with a mild metabolic acidaemia. Plasma propionic acid concentrations increased markedly, 6.0 mM in twin I and 4.8 mM in twin II, by day 7. Hydroxyproline, serine, asparagine, glycine, isoleucine, leucine, and ornithine concentrations were increased in the plasma of twin I. Asparagine, aspartic acid, glycine, citrulline, leucine, and lysine concentrations were increased in the plasma of twin II.

Propionyl coenzyme A carboxylase activity in liver mitochondria from each twin was less than 10% of control levels in human, rabbit, and rat liver mitochondria.

Biotin, pantothenic acid, and carnitine were without demonstrable clinical or marked biochemical effects.

It is suggested that the twins had clinical and biochemical abnormalities similar to cases previously described as having idiopathic ketotic hyperglycinaemia, methylmalonic acidaemia, and propionicacidaemia.

REFERENCE

- Gompertz, D., Storrs, C. N., Bau, D. C. K., Peters, T. J., and Hughes, E. A. (1970). Localisation of enzymic defect in propionicacidaemia. *Lancet*, 1, 1140.

Plasma copper and zinc in acute leukaemia. F. W. Alexander, H. T. Delves, and H. Lay (*introduced by C. G. D. Brook*). (*Institute of Child Health, 30 Guilford Street, London WC1N 1EH.*) The concentrations of copper and zinc have been measured in the plasma of leukaemic children before and after treatment and compared with healthy controls.

The plasma copper concentrations were higher and

the plasma zinc concentrations were lower for the untreated leukaemic children than for the other 2 groups. The concentrations of both metals were altered after treatment—the copper was lowered and the zinc increased—to values approaching the normal range.

The plasma Cu : Zn ratio discriminated well between the 3 groups of children and would be valuable both in the diagnosis and response to treatment of leukaemia in childhood. There was no correlation between this ratio and the total white cell or peripheral blast cell count. However, the ratio was proportional to the extent of the leukaemic process.

Further work is necessary to determine the roles which Cu and Zn play in leukaemia in children.

Familial hypobetalipoproteinaemia. A. S. Fosbrooke, S. Choksey, and B. A. Wharton (*introduced by B. A. Wharton*). (*Institute of Child Health, 30 Guilford Street, London WC1N 1EH.*) A 2-year-old boy admitted to hospital with gastroenteritis was further investigated because of small stature. He had low serum cholesterol (73 mg/100 ml) and reduced beta lipoprotein cholesterol (20 mg/100 ml). Faecal fat, jejunal biopsy, and red cell morphology were normal. Plasma growth hormone and thyroxine were also normal, and it was concluded that small stature was the result of hereditary and/or psychosocial factors.

A family study showed a similar lipoprotein abnormality in the patient's mother; her serum cholesterol was 79 mg/100 ml and beta lipoprotein cholesterol 34 mg/100 ml. Analysis of the beta lipoprotein fraction in both child and mother showed it to have abnormal lipid composition; the cholesterol/phospholipid ratio was 1.0 and 0.8 respectively (normal 1.7), and within phospholipid components the proportion of sphingomyelin was markedly reduced (11% and 5%; normal 30%). These findings differ from previous reports that the composition of beta lipoprotein is normal. Familial hypobetalipoproteinaemia has been shown to be inherited as an autosomal dominant, and our findings are in agreement.

Though a few individuals with this condition have been reported to have some though not all of the features associated with abetalipoproteinaemia, neither our patient nor his mother had any gastrointestinal, haematological, or neurological abnormalities.

Dystrophia myotonica in infancy and childhood. Victor Dubowitz. (*The Department of Child Health, University of Sheffield.*) Though characteristically a disease of adolescence and adult life, there have been several case reports in recent years of dystrophia myotonica in infancy and childhood.

Cases were presented illustrating the wide variation in clinical pattern. These included a boy, aged 10 at the time of diagnosis, with early features (myopathic facies), whose mother had a classical syndrome, thus making clinical diagnosis easy; a young man of 30 with a classical syndrome whose muscle symptoms date from birth; a girl of 13 presenting with scoliosis, who at birth had unexplained asphyxia and bilateral talipes, and