Paediatric Research Society

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Quantitation of small intestinal biopsies from children. J. W. Keeling and R. A. Risdon. The Hospital for Sick Children, London.

Small intestinal biopsies from children have been analysed quantitatively to obtain indices reflecting both the mucosal volume and surface-to-volume ratio.

In subjectively normal biopsies, correlation of the patients's ages with the surface-to-volume ratios showed the latter to be significantly reduced in younger children and some overlap occurred between these lower values and the range seen in biopsies showing 'partial villous atrophy'. However, in the abnormal biopsies additional mucosal abnormalities were invariably present. It is suggested, therefore, that slight changes in the villous pattern of biopsies from children under 2 years of age should be interpreted with caution and should not be regarded as necessarily pathological in the absence of other mucosal abnormalities. 'Flat' biopsies from children with untreated coeliac disease invariably showed smaller surface-to-volume ratios than controls, but the mucosal volumes of these biopsies were significantly increased. Specimens showing less marked abnormalities ('partial villous atrophy') had surface-to-volume ratios in an intermediate range, but, like the biopsies from untreated coeliacs, mucosal volumes were significantly increased. The most useful application of quantitative analysis is likely to be in following accurately the sequential changes in biopsies from the same patient. We have therefore applied this technique to serial biopsies obtained from a number of children suspected on clinical grounds of having coeliac disease, but in whom this diagnosis had not been confirmed histologically. Biopsies were obtained while the patients were receiving a gluten-free diet and also after a 'gluten challenge'.

Case of hyperammonaemia due to ornithine transcarbamylase deficiency. C. Morley and I. B. Sardharwalla. Willink Biochemical Genetics Laboratory, Royal Manchester Children's Hospital, Pendlebury, Manchester.

A 5-year-old female with normal mental and physical development presented with a 3-month history of episodic vomiting, lethargy, and nocturnal confusion, each lasting for about 48 hours. Spontaneous recovery occurred each time. In the final attack she became ataxic, confused, and lapsed into coma, which led to her admission. Examination revealed that she was deeply unconscious, pyrexial, and hypertonic with hepatomegaly. Investigations showed high blood and CSF ammonia levels ($>1000 \mu\text{g}/100 \text{ ml}$). In addition, the

urine contained increased amounts of pyrimidine derivatives, namely orotic acid, uridine, and uracil. Plasma and urine glutamine concentrations were raised. In an attempt to lower blood and CSF ammonia, peritoneal dialysis was carried out with standard dialysis solution and the ammonia levels returned to almost normal within 48 hours. The effectiveness of dialysis was shown by high levels of ammonia in the dialysate. Unfortunately the child died of cerebral oedema. Enzyme assay in the liver obtained at necropsy within 6 hours of death showed marked deficiency of ornithine transcarbamylase which was 3% of normal at pH 7. The other urea cycle enzymes were normal.

Osteodystrophy associated with prolonged cholestasis. J. F. T. Glasgow and P. S. Thomas. Institute of Clinical Science, Belfast.

Metabolic bone disease was studied in 9 infants (birthweight 1.02–3.53 kg) during the course of prolonged obstructive jaundice beginning in early infancy. 5 had proven biliary atresia and in 2 others cholestasis complicated severe rhesus isoimmunization. The occurrence of bone disease at 2 to 4 months of age was accompanied in only 3 patients by features suggesting this diagnosis, and jaundice was diminishing in those who did not have biliary atresia. Indeed, 2 patients were no longer clinically icteric. At the time of diagnosis each patient had a marked rise in serum alkaline phosphatase concentration and aminoaciduria. Serum calcium and phosphorus values were more variable, the former being closely related to the gestational age of the baby at birth. X-rays in the majority showed classical rickets with bone age retardation. One infant, however, had in addition to rickets of the scapula, severe demineralization of the axial skeleton and multiple rib fractures. The appendicular skeleton was normal. Rapid skeletal healing took place in those whose cholestasis subsided. In biliary atresia distinctly abnormal bones remained and one child of 13 months developed multiple long bone fractures. Various pathogeneses might be contributory to the osteodystrophy, including a shortened gestation.

Effect of B17-V inhalation on hypothalamic-pituitary-adrenal axis and on growth in asthmatic children. M. Friedman and J. Frears. University College Hospital, London.

The effect of the administration of 1600 μg daily of B17-V by aerosol was studied in 15 adult volunteers over a 7-day period. The daily plasma cortisol levels taken at