Paediatric Research Society

15th Meeting, The Children's Hospital, Sheffield, March 28-29, 1969

Abstracts of Papers

pH Changes in Children with Burns. F. Harris and J. Black. (*Department of Child Health, University of Sheffield*). As part of a study in depth of metabolic changes in burned children, serial acid-base studies were done. In order to evaluate the findings further, a group of children admitted for elective minor surgery had as part of the pre-operative assessment, an analysis of the acid-base status. A small number had serial post-operative analyses done. The results in patients with burns of varying severity were presented and compared with the data obtained from children undergoing minor elective surgery. The clinical significance of these findings was discussed.

Observations on Fat and Carbohydrate Metabolism in Generalized Lipodystrophy. M. Segall and J. Lloyd (*Institute of Child Health, London*). Investigations of fat and carbohydrate metabolism were made on a 10-month-old girl with generalized lipodystrophy; her parents were second cousins and her appearance was abnormal from birth. Subcutaneous tissue obtained by open biopsy showed no macroscopical fat; histologically a few isolated areas of adipose tissue were seen and the cells contained little lipid. Analysis of lipid extracted from the tissue showed a greatly reduced triglyceride content (0.4% wet weight), with a low percentage of linoleic acid (3.0% of total fatty acids).

Fasting plasma non-esterified fatty acid (NEFA) concentrations were normal (0.52–1.15 mEq/l.); the percentage of linoleic acid was reduced (2.3–4.6%). After subcutaneous adrenaline (0.01 mg./kg.) plasma NEFA concentrations showed only a transient increase. After oral glucose (2 g./kg.) the plasma glucose and insulin levels rose but the normal reduction in plasma NEFA did not occur; the glucose tolerance curve showed no diabetic features. Fasting plasma triglyceride concentrations were variable (82–648 mg./100 ml.) but were usually raised, and paper electrophoresis showed a prominent pre- β -lipoprotein fraction. Triglyceride fatty acids showed an increased percentage of palmitic acid (41.5–46.7%) and reduced percentages of oleic acid (35.1–41.3%) and linoleic acid (0.8–3.4%). After oral glucose there was an increase in plasma triglyceride from 84 mg./100 ml. to 146 mg./100 ml.; this is an abnormal response. An oral fat

load test (2.2 g./kg.) showed delayed clearing of plasma triglyceride, and the post-heparin lipolytic activity was very low (0.08 μ Eq fatty acid/ml. plasma per min.). Fasting plasma triglyceride and pre- β -lipoprotein were reduced by a low fat diet. These findings show that fasting plasma NEFA is maintained at normal concentrations despite the gross depletions of adipose tissue; however, the regulation of plasma NEFA by glucose and insulin appears abnormal. The hypertriglyceridaemia results mainly from a defect in peripheral triglyceride clearance.

Nephrotoxic Effect of Vitamin D Therapy in Vitamin D-Refractory Rickets. M. Moncrieff (*Introduced by R. H. R. White*). Published in full, in *Archives of Disease in Childhood* (1969), 44, 571.

Incorporation of Methionine Sulphur into Cysteine *In Vitro* by Fibroblasts Deficient in Cystathionine Synthetase. P. F. Benson, J. L. Hamerton, and V. Young (*Paediatric Research Unit, Guy's Hospital Medical School, London*). The main pathway for the incorporation of methionine sulphur into cysteine involves its transference to the carbon residue of serine. A step in the metabolic pathway involved is catalysed by the enzyme cystathionine synthetase.

Though the activity of cystathionine synthetase of cultured fibroblasts from patients with homocystinuria was considerably reduced (4 controls 1.7 to 2.3; 2 homocystinurics 0.2, 0.4 μ mole of cystathionine formed per hr. per g. protein), the rate of incorporation of 35 S-methionine into protein as 35 S-cysteine and 35 S-methionine was similar to both types of cells.

The utilization of an alternative transulphhydrase pathway by homocystinuric cells was investigated. In this pathway homocysteine desulphhydrase catalyses the conversion of methionine sulphur into hydrogen sulphide. The latter then reacts with ammonia and pyruvic acid, catalysed probably by a reversal of the cysteine desulphhydrase reaction. In cells from subjects with homocystinuria a raised activity was found of cysteine desulphhydrase (4 controls 8.3 to 14.6 units; 2 homocystinurics 23.6, 48.3 units). Homocysteine desulphhydrase activity was similar in controls and patients (4 controls 8.9 to 12.5 units; 2 homocystinurics 10.1, 11.1 units).

The characteristic biochemical abnormalities in