Intestinal obstruction in utero. G. Molz. Anatomishes Institut der Universitat, CH-8006 Zurich.

Absence of islets of Langerhans in pancreas of a newborn. K. M. Laurence and J. A. Dodge. Department of Child Health, Welsh National School of Medicine, Cardiff.

A small-for-dates male infant developed respiratory distress and increasing metabolic acidosis with rapid deep respirations and a rapid thready pulse which failed to respond to the usual measures. At 36 hours he was found to have a blood glucose of 800 mg/100 ml, with acetone in the urine and was diagnosed as having diabetes mellitus with dehydration. He was given two injections of 2 units of insulin which reduced the blood glucose to only 680 mg/100 ml, and an attempt at rehydration led to cardiac arrest and death at 40 hours. At necropsy there was patchy haemorrhagic consolidation of the lungs, dilatation of the heart, thymic exhaustion, severe fatty change of liver, and cerebral oedema. Complete absence of islets of Langerhans was noted in the pancreas, and there were no isolated α or β cells, though some collections of lymphocytes were present. Argentophil cells in the gut were scanty.

This was the fourth child of healthy unrelated parents, whose first child, a boy born severely small-for-dates, died under similar circumstances at 48 hours but no histological examination of the pancreas was undertaken. It is suggested that both boys had the same underlying pathology and that this might be a previously undescribed recessively or X-linked inherited condition.

Clinical observations and histopathology of the liver in infants and children with α_1-antitrypsin deficiency. J. D. Elema, R. Boersma, E. Reerink-Brongers. Pathologisch-Anatomisch Laboratorium der Rijksuniversiteit, Groningen, Oostersingel 63, Holland.

10 children had severe α_1-antitrypsin (AAT) deficiency of ZZ phenotype, 2 had partial AAT-deficiency of MZ phenotype, and 5 who had died before the study, had probably suffered from AAT deficiency. All had presented with neonatal jaundice and most had hepatomegaly and failure to thrive. Alkaline phosphatase, bilirubin, and SGOT and SGPT levels were moderately increased. There was a spontaneous gradual recovery. Biopsies taken between 2 and 4 months after birth showed portal fibrosis. Giant cells were not a common finding. PAS-positive inclusions sometimes were only minimal, as was immunofluorescence. 7 children developed cirrhosis of the liver, all of whom underwent laparotomy during infancy. One of these children had an MZ phenotype. None of the children not undergoing laparotomy has developed cirrhosis so far. It is concluded that the number of PAS-positive inclusions can sometimes be small and this is probably related to other factors than phenotype only. The clinical picture of neonatal jaundice is not related to AAT storage and laparotomy may have an ill effect on the prognosis of the liver disease.

Hepatic ultrastructure in a child with carbamyl phosphate synthetase deficiency, hyperornithinaemia, hyperammonaemia, and homocitrullinuria. M. Daria Haust and P. D. Gatfield. Departments of Pathology and Biochemistry, Children's Psychiatric Research Institute and University of Western Ontario, London, Ontario, Canada.

To date only 2 children have been reported with hyperornithinaemia, hyperammonaemia, and homocitrullinuria. In the case we report there was, in addition, a deficiency of mitochondrial carbamyl phosphate synthetase I (CPS I), the enzyme involved in the first step of the Krebs-Henseleit urea cycle. The enzyme is believed to be localized largely in hepatic mitochondria. The patient, a 9-year-old severely retarded boy, had a history of vomiting, aversion to food, episodes of lethargy, EEG changes, and raised SGOT since early infancy. He has been followed since the age of 7 years when increased urinary excretion of ornithine and lysine and increased blood ornithine and ammonia were discovered. Restricted protein intake resulted in slight improvement in growth, but height and weight remain below the 3rd centile. He has speech impairment, increased tendon reflexes, markedly delayed bone age, and a slightly enlarged and firm liver. His blood ornithine (up to 550 μmol/l) and ammonia (up to 270 μgN/100 ml), and urinary excretion of homocitrulline (up to 284 μmol/g creatinine) are all markedly increased. There is total absence of CPS I (and CPS II) in leucocytes and a decrease to 20% of normal activity in the liver.

Electron microscopy of biopsied liver tissue showed that in the nonreticulated cells mitochondria were either strikingly enlarged and filled with crystalloid structures, or small with numerous abnormal cristae. Many mitochondria showed a peculiar periodicity at the level of the inner membrane. The morphology of the mitochondria was thought to correlate with the biochemical data.

Morphology of liver glycogenosis. W. C. de Bruijn, J. Fernandes, J. Huber, and J. F. Koster. Medical Faculty, Erasmus University, Rotterdam; Sophie Children's Hospital and Neonatal Unit, Gordelweg 100, Rotterdam, The Netherlands; and Department of Pathology, Hospital for Sick Children, Toronto, Ontario, Canada.

Morphology of the liver was compared by light and electron microscopy of 13 children with glycogen storage disease. The results were correlated with biochemical enzyme assays of liver tissue and of leucocytes to determine whether ultrastructural criteria could be used to differentiate glucose-6-phosphatase deciency (type I), debranching enzyme deficiency (type III), and deficiency of the phosphorylase system (type VI). Marked steatosis of the hepatocytes and the characteristic ultrastructure of the lipid droplets clearly differentiated the glucose-6-phosphatase deficiency from the two other deficiencies. The presence of slightly electron dense areas between the glycogen particles found in phosphorylase deficiency may be used