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


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 a or b 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.


10 children had severe α1-antitrypsin deficiency (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, weight, and bone age were all 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 crystallloid 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, Godelweg 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 deficiency (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.
to differentiate this enzyme deficiency from debranching enzyme deficiency. The amount of collagen present in the liver tissue varied considerably in these two disorders. In cases of debranching enzyme deficiency, collagen fibres grossly disturbed the liver architecture. The degree of fibrosis varied considerably in the liver in phosphorylase deficiency. By light microscopy the presence of collagen fibres was not impressive in glucose-6-phosphatase deficiency, though by electron microscopy collagen was shown in the spaces of Disse.

Localized obliteration of bile ducts. H. B. Marsden. Department of Pathology, Royal Manchester Children's Hospital, Pendlebury, Manchester M13 0JH.

Limitation of growth potential of kidney by injury in early life. C. L. Berry, G. W. Slocombe, and P. R. Freeman. Department of Morbid Anatomy, Guy's Hospital Medical School, St. Thomas's Street, London SE1.

There is evidence to suggest that ureteric reflux and urinary tract infections in early childhood may affect the growth potential of the kidney. Since tubular growth is mainly a postnatal event in man, it is possible that permanent damage may result from a depletion of the tubular cell pool. This hypothesis was examined in the developing rat kidney, whose growth is similar to that of man. Tubular cell numbers were reduced using mercuric chloride. The effect of unilateral nephrectomy was also examined. DNA and RNA estimations were used to assess renal growth in terms of cell numbers, mean cell size, and RNA/DNA ratio. It was found that the kidney was capable of repairing extensive necrosis within 10 days. Kidneys stressed by unilateral nephrectomy showed evidence of hyperplasia and hypertrophy. Unilateral nephrectomy in addition to mercuric chloride was followed by a hypertrophic response alone and cell numbers failed to increase. These findings suggest that while the developing kidney can repair tubular injury, a sufficiently gross depletion of tubular cells during a period of rapid growth may result in a permanent reduction in kidney size.

Primary causes of neonatal mortality: prospective study of 100 cases. V. V. Joshi. Department of Pathology, Medical College of Virginia, Virginia Commonwealth University, Richmond, Virginia.

Many of the previous reports on causes of neonatal mortality have the following faults. They are retrospective studies, they mix primary and secondary causes of death, they do not breakdown causes of extrinsic perinatal hypoxia (EPH), and such important causes as intrauterine growth retardation and neonatal necrotizing enterocolitis are excluded. The list of primary causes of death used by the Perinatal Mortality Committee of Quebec Province, Canada, was modified to avoid the above criticisms. The methodology and results of 100 consecutive necropsies, excluding macerated stillbirths, are reported. Respiratory distress syndrome (RDS) 31%, congenital anomalies 28%, and EPH 17% were the three commonest causes of death. In the low birthweight infants (<2500 g) RDS was the most frequent cause (46%), and in full birthweight infants (>2500 g) congenital anomalies were the most frequent (57%). Maternal, placental, or labour and delivery problems were associated with about half the cases of EPH. In 2 cases the cause of death remained unexplained, though sudden infant death syndrome may be a possibility.

Postperinatal deaths and unexpected deaths in infancy: prospective study. J. L. Emery. The Children's Hospital, Sheffield.

Home unexpected deaths now constitute the largest group of postperinatal deaths. The state of babies at the time of birth, who had later been unexpected deaths, has been compared with control children and a scoring system devised in order to select children at increased risk of unexpected death (Proastos et al., 1973).

A prospective study using this criteria has been carried out in Sheffield which suggests that the method is functioning, and also identifies children having a doubled risk of requiring hospitalization (Carpenter and Emery, 1974). The study will be continued for a further year before a final report is made.

REFERENCES


Fatty change in brains of perinatal and unexpected deaths. D. R. Gadson and J. L. Emery. Children's Hospital, Sheffield.

Earlier work showed that the presence of fat-laden cells in the CSF of children with hydrocephalus is an indicator of severe brain damage (Chester, Emery, and Penny, 1971; Chester, Penny, and Emery, 1971). The findings of such cells in CSF of some unexpected cot deaths and perinatal deaths stimulated a survey of the brains of such children to ascertain the likely site of origin. This was found to be in the region of the corpus callosum, the septum pellucidum, fornix, and related regions of the lateral ventricles. A survey carried out of 200 brains of children dying in the postperinatal period indicated that the fatty change is principally related to two groups of children—early perinatal deaths associated with severe hypoxia and a number of the unexplained, unexpected deaths in infancy. Evidence was presented which indicated that this fatty change is pathological and suggests that it is a reaction to hypoxia in actively myelinating brain tissue.

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doi: 10.1136/adc.50.8.663-d

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