Paediatric Pathology Society

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Aminoacidopathies

At about 8 months gestation she developed lupus nephritis and was treated with prednisolone and frusemide. She delivered a male infant spontaneously at 36 weeks.

The infant was ‘small-for-dates’, developed hypoglycaemia, and died on the fourth day. At necropsy extensive pulmonary haemorrhage was found. There was calcification of the fat in the neck, the intima of the pulmonary arteries, the aorta, and the circle of Willis. There was hyperplasia of the kidneys. There was hyperplasia of the islands of Langerhans of the pancreas and also of the adult cortex of the adrenal. The kidney showed fibrosis of the subcapsular zone, destruction of some tubules, and dilatation of others. Large foci of calcification were seen. The bones showed depressed osteogenesis.

Liver Damage after Treatment with THAM via Umbilical Catheter. J. D. Elema (Pathologisch-Anatomisch Laboratorium, Oostersingel 63, Groningen, Holland).

Symposium on the Early Detection of Phenylketonuria

The Organization of a Large-scale Guthrie Screening Programme. S. F. Cahalane (Department of Pathology, The Children’s Hospital, Temple Street, Dublin, Eire).

Early Detection of Phenylketonuria and Other Aminoacidopathies in a Large City Using Plasma Chromatography. D. N. Raine (Department of Biochemistry, The Children’s Hospital, Birmingham B16 BET). Capillary blood has been collected from infants, mainly at 6 to 9 days of age, born in the City of Birmingham (population 1 million; 20,000 births per year), for the past 18 months and the plasma subjected to paper chromatography as described by Scriver, Davies, and Cullen (1964). Midwives take the samples to one of 12 collecting centres and all are delivered to the laboratory by midday. All subsequent work is done by one person whose only laboratory training has been in this procedure. The interpretation is checked by a biochemist.

Studies have established that the results on specimens posted to the laboratory, although different from those delivered immediately, can be interpreted satisfactorily. The optimum age for repeating initial tests which show only increased tyrosine has been established as six weeks, irrespective of whether the infant is premature or not. Administration of ascorbic acid has not shortened this period by a useful amount.

In 18 months the diagnoses that have been made include phenylketonuria 3, histidinaemia 3, prolinaemia 2, and hyperlipidaemia 1. There have been 14 infants showing hypermethioninaemia, mostly transient, but four required admission and one died with a possible diagnosis of ‘tyrosinosis’.

The initial test needed to be repeated in 6% of all cases. Three-quarters of the repeats were required because the initial pattern was abnormal, and 66% of the abnormal patterns were due to tyrosine. The remaining one-quarter were repeated for technical reasons (haemolysis, specimen collected before 6 days, and laboratory mishaps).

Reference


Screening for Phenylketonuria on a District Hospital Scale. L. R. Davis (Department of Pathology, The Belgrave Hospital for Children, Clapham Road, London S.W.9). A report was presented on the experience gained in the first year's work on screening infants born in the King's College Group of Hospitals and the neighbouring Borough of Southwark. The reasons for undertaking small scale screening were given. Over the year 3500 Guthrie tests had been performed. The procedures for minimizing the clerical and technical work were described. Technical difficulties had been confined to attempting quality control of the results because of the variation in the amount of growth with different batches of medium and on different plates. Ways of allowing for this variation were suggested. Only two positive tests were discovered, but about 0.5% of tests gave results about the upper limit of normal and required repeating.

No true cases of phenylketonuria were detected during the year.

Management of Hyperphenylalaninaemia (HPA) in Northern Ireland. Nina A. J. Carson (Nuffield Department of Child Health, The Queen's University of Belfast, Grosvenor Road, Belfast BT12 6BJ). For a period of 1 year and 9 months, from 1 January 1969 to 30 September 1970, 56,257 newborn infants were screened in Northern Ireland for phenylketonuria (PKU) by the use of the Guthrie inhibition assay.

Specimens were collected on the 7th day of life by midwives, health visitors, or hospital staff and all tests were carried out in one centre in Belfast under the direction of the Consultant Bacteriologist of the Royal Victoria Hospital, Dr. George Gibson.

In all babies with phenylalanine levels of 4 mg/100 ml and above, repeat tests were requested and those with rising levels were investigated further. In these infants venous blood was examined for aminoacids by the use of an automatic aminoacid analyser, and a sample of urine examined for aminoacids and phenylketones. 17 infants showed levels persistently above 4 mg/ml. 8 infants were found to have classical PKU with phenylalanine blood levels in excess of 25 mg/100 ml, and phenylketones in the urine. A daily intake of phenylalanine of less than 300 mg was required to keep their blood phenylalanine levels between 2–8 mg/100 ml. 9 infants had persistently high blood levels varying from 12–23 mg/100 ml. On initial testing, urinary phenylketones were not found in this group. All were started on treatment, those with lower levels being given a low natural-protein diet; those with levels nearer 20 mg/100 ml, a hydrolysate mixture as in classical PKU.

Up to the time of writing, three of these children could tolerate a normal diet and the remaining six were still pregnant.
Early detection of phenylketonuria and other aminoacidopathies in a large city using plasma chromatography.

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