The excretion of the total 17-hydroxycorticosteroids and the α-ketonic metabolites of cortisol (tetrahydrocortisol, allo-tetrahydrocortisol, tetrahydrocorticosterone) increases from infancy to adult life and when expressed per 100 kg of body weight show a close relationship. The corticosterone metabolites (tetrahydrocorticosterone, allo-tetrahydrocorticosterone, tetrahydro-11-dehydrocorticosterone) when expressed similarly show a marked fall during the first few years of life.

G. Komrower (Manchester). 'Experiences with the Scriver Technique as a Screening Procedure for Phenylketonuria and Other Amino Acid Disorders.'

D. N. Raine introduced by DR. MARGARET I. GRIFFITHS (Birmingham). 'Screening for Several Amino Acidopathies in Neonates in Birmingham by Plasma Chromatography.'

A screening programme based on the plasma chromatographic method described by Scriver, Davies, and Cullen (1964) was established in Birmingham (population 1 million, 20,000 births per year) two years ago. The organization at domiciliary, laboratory, and City Health Department levels will be described. As a result of specific studies it has been found that:

1. The programme can be conducted with posted specimens as well as those analysed on the day of collection.
2. The time of collection of blood in relation to feeding, although leading to differences, does not present special problems.
3. Chromatography of plasma is superior to that of discs punched from paper soaked with whole blood collected for the Guthrie test.
4. When, at 6–9 days, tyrosinaemia is the only abnormality, further tests should be delayed until the age of 6 weeks when 95% will have become normal.
5. The cost of Scriver testing is similar to that of Guthrie testing up to 30,000 tests/year; above that, Guthrie testing will probably be cheaper.

Such a screening programme will lead to about 40 additional outpatient visits per year and 12 additional admissions for further investigations and treatment. Though this system can detect up to 19 amino acid disorders, the efficiency of this method and the optimal time for testing for those other than phenylketonuria has not yet been established.

REFERENCE


MARGARET I. GRIFFITHS (Birmingham). 'Implications for Clinical Implementation of Results of Metabolic Screening for Amino Acidopathies in the Newborn.'

This paper discussed some of the problems that have arisen in the clinical follow-up of neonates examined in Birmingham, as described by D. N. Raine.

It was found essential that where there was any question of persisting aminoacidemia that the babies should be examined and that they should be seen regularly at a follow-up clinic. The following aminoacidemias presented clinical problems:

1. Children with methioninemia. These comprise a group of 18 children, the majority of whom were immigrants, and the majority of whom were premature. The implications of this were discussed. In addition, some of the babies showed rickets and/or abnormalities in the liver function tests. The significance of this was discussed.

2. Infants with histidinemia. So far three of these infants have been detected. As there is doubt as to the effect of histidinemia all three have been put on a histidine-free diet and their speech and language development is being recorded.

3. Prolineaemia. Three children with prolineaemia have been found.

The implication of abnormal aminoacidemia in early life is not yet clear. It is considered that it is most important that these children should be carefully followed up and, particularly, should be assessed at school age. It has been suggested (Woolf, 1968) that because the incidence of phenylalaninemia is very much higher in the newborn than in the population in subnormality hospitals that many undetected cases of phenylalaninemia are of normal intelligence. The converse may well be true and it may be that children who have had transient abnormalities of amino acid metabolism during the early months of life, may show permanent retardation for which no cause can be found, if they are only examined at a later age. This hypothesis was discussed.

REFERENCE


BARRABA E. CLAYTON (London). 'Experience with a screening service, using the Guthrie Test, in the North-West and North-East Metropolitan Regions.'

Infants in two metropolitan regions have been screened by the Guthrie test for raised phenylalanine (Phe) levels in blood. The results obtained in 117,446 infants were:

1. 8 infants had classical phenylketonuria (PKU), i.e. an incidence of 1 in 14, 680 births. One of these had a hitherto undiagnosed PKU sib who was very retarded, and two had sibs already receiving dietary treatment for classical PKU.

2. 1 infant had atypical PKU with plasma phenylalanine levels of 20–27 mg/100 ml. He had a hitherto undiagnosed sib of normal mentality with Phe levels of 12–20 mg at age 2.2 to 6 years.

3. 391 infants (i.e. 0.35–3% of the total) had raised values when first tested, but on repeating about 1 week later these had fallen to normal.

4. 26 infants had raised values persisting after the second test. The maximum value was 15 mg and all except 2 infants had tyrosinaemia associated with prematurity, excessive protein intake, etc.
None of the infants raised difficult diagnostic problems.

Among 82,407 infants tested for raised methionine levels:

1. 1 had homocystinuria.
2. 31 had raised values when first tested but these were normal at the second test.
3. 6 had more persistent methioninaemia and in 1 this was still present after 14 weeks and associated with rising levels of alkaline phosphatase and transaminase.

The designated screening laboratory forms part of the Department of Chemical Pathology, so that biochemical investigation of positive results presents no administrative problems, and there is complete paediatric cooperation for the further diagnosis and clinical care of infants in whom this is indicated. This is the pattern advised in a Government paper HM (69) 72.

J. MELLON introduced by Dr. V. J. MARRIAN (Dundee). ‘Plasma Amino Acid Screening in the Newborn.’

A new microscreening method has been developed to detect phenylketonuria and the plasma aminoacidopathies using one-dimensional thin layer chromatography on cellulose layers. Amino acids are accurately and rapidly determined in a 2 μl sample of plasma without prior deproteinization or desalting of the plasma.

To obtain a true picture of the incidence of the plasma aminoacid disorders, it was necessary to broaden the screening programme by testing for a large number of abnormalities. It has now been shown that previous estimations of the incidence of the plasma aminoacid disorders were probably inaccurate. 2.5% of the total number of infants in the screening programme had plasma aminoacid abnormalities. Disorders of tyrosine metabolism, followed by disorders of phenylalanine metabolism represent the greatest incidence of individual abnormalities and together these represented 93% of the 2.5% incidence of all the aminoacid disorders detected.

A Seminar on The Management of Myelomeningocele was held on Saturday morning, 24 April, in the Royal Hotel, Scarborough, with Professor S. D. M. Court as Chairman. The following speakers took part: Mr. R. B. Zachary (Sheffield) — ‘The Ethics of Treatment’. Mr. B. McKibbin (Sheffield), by invitation — ‘Orthopaedic Aspects’. Mrs. B. M. Freeston (Sheffield), by invitation — ‘Stress in Family Life’. Dr. J. Lorber (Sheffield) — ‘Long-Term Results: The Indications for and the Case for Selection in Treatment’.

GEORGE FREDERIC STILL MEMORIAL LECTURE. The Lecture was delivered on 22 April by Professor Harry Harris (Galton Professor of Human Genetics) — ‘Genetic Heterogeneity and the “Inborn Errors of Metabolism”.’ Members and guests of the Association attended a Civic Reception at the Town Hall on the evening of 22 April, by invitation of the Mayor and Corporation of Scarborough.

The Ulster Cup competition was held at the Ganton Golf Club on 23 April, and was won by Dr. R. J. Young. The U.C.H. Squash Cup competition was held at the Scarborough Lawn Tennis and Squash Club and won by Dr. K. Vernon Jones.

The Annual Dinner was held on the evening of 23 April, with His Grace the Archbishop of York as guest of honour.