The excretion of the total 17-hydroxycorticosteroids and the α-ketolic 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 Scrivener Technique as a Screening Procedure for Phenylketonuria and Other Amino Acid Disorders.'

D. N. Raine introduced by Dr. Margaret I. Griffitts (Birmingham). 'Screening for Several Amino Acidopathies in Neonates in Birmingham by Plasma Chromatography.'

A screening programme based on the plasma chromatographic method described by Scrivener, 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 Scrivener 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. Griffitts (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 methioninaemia. 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 histidinaemia. So far three of these infants have been detected. As there is doubt as to the effect of histidinaemia all three have been put on a histidine-free diet and their speech and language development is being recorded.
3. Prolineaemia. Three children with prolinaemia 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 phenylalaninaemia is very much higher in the newborn than in the population in subnormality hospitals that many undetected cases of phenylalaninaemia are of normal intelligence. The converse may well be true and it may be that children who have had transient abnormalities of aminoacid 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:

Barbara 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 2·6 years.
3. 391 infants (i.e. 0·35% 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.