



Dear Colleague

NEWBORN SCREENING FOR GALACTOSAEMIA

I am writing to remind you that, as already announced in NHS HDL(2001)73 issued on 8 October 2001, **the routine testing of newborn bloodspot specimens for Galactosaemia will cease at the end of March 2002.** The decision is based on the advice of the National Screening Committee, who have endorsed the findings of an HTA report. A systematic review of the evidence concluded that there is no evidence to support a newborn screening programme for Galactosaemia and any current newborn screening for Galactosaemia should be discontinued. Screening for this condition no longer takes place in England and Wales.

Cessation of this routine test on all newborns means that it is even more important that Galactosaemia is included in the differential diagnosis for any newborn baby or small infant who presents with the relevant clinical features. As you are aware early diagnosis and treatment is of the essence in the successful management of this condition. It is therefore of paramount importance that affected infants are recognised and referred for specialist investigation and care at the earliest possible opportunity. As an aide memoir, the clinical features of Galactosaemia are detailed in Appendix 1.

The screening of all newborn infants in Scotland for Phenylketonuria (PKU) and Congenital Hypothyroidism will continue as usual. At a later date tests for Cystic Fibrosis will be added to the programme. Although NHS HDL(2001)73 announced that newborn screening for Cystic Fibrosis would be introduced from April 2002, it has been decided to delay for a short time the implementation of this testing programme. This will allow more time to enable the development of all the appropriate training arrangements and organisational infrastructure necessary for the delivery of a really robust screening programme.

**From the
Chief Medical
Officer**

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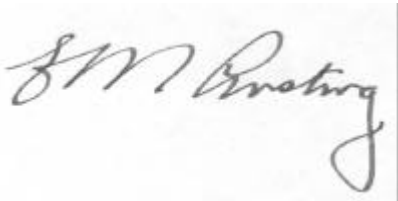
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A programme of work is underway to further develop and strengthen all aspects of the newborn screening programmes. One of the first changes to be introduced will be the distribution of freepost envelopes for the return of the Guthrie Card to the screening laboratory after the blood spot specimen has been obtained from a baby. It is hoped that the introduction of this measure will help to achieve the earliest possible return of the Cards by removing the need to batch Cards before posting. It is important that the return of the Cards is not delayed in order to batch cards because again the earliest possible diagnosis and treatment is vital for the prognosis of infants with PKU or Congenital Hypothyroidism. We will keep you informed of further developments with the programme as they occur.

Yours sincerely

A handwritten signature in black ink, appearing to read 'E M Armstrong', is written on a light-colored background. The signature is cursive and somewhat stylized, with a vertical line to its right.

DR E M ARMSTRONG

ANNEX 1

Galactosaemia

It is important to be aware of this rare condition as the early lethal consequences are treatable. However, **recognition** has to precede **treatment**.

How does it present?

Most affected children, fed on breast milk or on lactose-containing infant formula, become very unwell toward the end of the first week of life. There is vomiting, jaundice, lethargy and failure to gain weight because of liver disease. There may be bruising or frank bleeding, hepatomegaly and ascites. A faint central "oil drop" cataract may be visible by ophthalmoscope. Sepsis with coliform bacteria may coexist. In the fully established disease there will be liver failure, renal tubular disease and perhaps cerebral oedema, and there may, but not commonly, be hypoglycaemia. A number of children, may present later, at 1 to 6 months old, but this is unusual in the United Kingdom. Prolonged jaundice in an unwell baby should prompt consideration of galactosaemia.

How is it diagnosed?

The only diagnostic test is measurement of galactose-1-phosphate uridyl transferase in red blood cells.

Are other tests useful?

Galactose may be present in urine. However simple tests for "reducing substances" in urine by "Clinitest" tablets are unreliable for several reasons

- 1 Galactose will be found in urine only if the baby has been fed breast milk or a lactose containing infant formula in the preceding few hours and has not vomited.
- 2 Galactose will not be found in the urine of an ill baby on intravenous fluids.
- 3 Galactose and glucose may both be present while the infant is fed with milk. If a positive "Clinistix" test is found (which is specific for glucose), then the coexistence of galactose in urine may be overlooked.
- 4 Cephalosporin antibiotics may cause false positive "clinitest" for reducing substances.

Urine sugar chromatography, red cell galactose-1-phosphate, quantitative blood galactose or urine galactitol are not routine tests, take time and are not widely available.

Whom should I ask for help?

If you suspect galactosaemia, the child should be referred urgently to a paediatrician to be seen that day.

What should the paediatrician do?

Think of galactosaemia in a child with the symptoms and signs above. Remember that E. coli sepsis and galactosaemia may occur together. (The differential diagnosis of the list of symptoms given above is wide and not the primary purpose of this notice)

Stop lactose-containing milk feeds until the diagnosis can be excluded, or confirmed. Remember it is possible to be misled by examining urine for reducing substances.

Send the correct specimen to a hospital laboratory which is either able to assay galactose-1-phosphate uridyl transferase in red cells, or is able to process the specimen properly, and arrange transport to another capable laboratory. The only laboratories currently offering this assay are at the Royal Hospital for Sick Children in Edinburgh and the Royal Hospital for Sick Children in Glasgow.

What specimen is needed?

1.0 ml blood in a lithium heparin bottle, avoiding blood clots, sent without delay, after alerting the laboratory. The laboratory may be able to do a *rapid qualitative test* (reported as *normal* or *reduced* activity) or a *quantitative test* (if a *numerical* result is needed).

Some particular situations

The presence of donated red cells may delay reliable testing for 3 months, until all donated cells have been replaced. It may be advisable to take and store blood samples for analysis before blood is given or exchange transfusions are carried out in babies in whom there is reason to suspect galactosaemia. Where there is a strong suspicion galactosaemia may be inferred from measurement of the enzyme in blood from both parents. This will require a *quantitative test* and must be discussed with the laboratory