

# BLOODSPOT SCREENING

## INTRODUCTION

- Screen babies on day 5 of age (date of birth = day 0) for the following conditions:
  - sickle cell disease
  - phenylketonuria (PKU)
  - congenital hypothyroidism (CHT)
  - cystic fibrosis
  - medium chain acyl co-A dehydrogenase deficiency (MCADD)
  - maple syrup urine disease (MSUD)
  - isovaleric acidaemia (IVA)
  - glutaric aciduria type 1 (GA1)
  - homocystinuria (HCU)
  - severe combined immunodeficiency (SCID)

***Obtain pre-transfusion bloodspot samples as previous blood transfusions can falsify results***

## TIMING

Co-ordinate with other tests when possible to minimise invasive procedures

### **If transfused before day 5**

- Collect first bloodspot card before transfusion
  - fill 1 circle
  - mark card 'pre-transfusion'
- Collect second bloodspot card at aged 5–8 days and  $\geq 72$  hr after blood transfusion
  - fill 4 circles
  - record whether plasma or red cells transfused
- Staple pre-transfusion and second bloodspot card together and send to West Midlands screening centre via courier service after validation check
- If baby has not received a blood transfusion the pre-transfusion bloodspot card can be discarded appropriately

### **Multiple transfusions between aged 5–8 days**

- Collect 4 bloodspots within this window. Complete with as much time-lapse as possible from any transfusion
- If  $\leq 3$  clear days between the last transfusion and routine sample, a repeat sample to be taken  $\geq 3$  clear days after the last transfusion

### **No transfusions before day 5**

- Collect routine bloodspot card at day 5
  - fill 4 circles and send to West Midlands screening centre via courier service after validation check, irrespective of milk feeds or gestational age

***Preterm babies  $\leq 31$  weeks and 6 days will require repeat sample at 28 days or discharge home, whichever is the sooner, for CHT. Collect 2 bloodspots. Mark the sample 'CHT preterm' and write the gestational age on the card***

## CONSENT AND INFORMATION

- Person undertaking procedure must:
  - offer parents an informed choice about screening for their baby, to gain consent and prepare them for the blood sampling procedure
  - provide national pre-screening leaflet at least 24 hr before procedure
  - parents must understand that they are consenting to storage of residual bloodspots, these can be used for:
    - checking screening results

- monitoring and improving the screening programme and health research that does not identify babies
- It is mandatory to include baby's NHS number on the bloodspot card
- If screening declined:
  - for all conditions – send completed card to screening laboratory (without blood sample) clearly marked DECLINE – ALL CONDITIONS
  - inform GP, health visitor and Child Health Records Department, in writing, of conditions baby not screened for
    - template letters available from: <https://www.gov.uk/government/publications/declined-newborn-blood-spot-screening-template-letters>

**Further information**

Detailed information regarding the UK Newborn screening programme can be sourced from:

- Newborn bloodspot screening programme handbook:  
<https://www.gov.uk/government/publications/health-professional-handbook-newborn-blood-spot-screening>
- Standards for NHS newborn bloodspot screening:  
<https://www.gov.uk/government/publications/standards-for-nhs-newborn-blood-spot-screening>
- Newborn bloodspot screening sampling guidelines:  
<https://www.gov.uk/government/publications/newborn-blood-spot-screening-sampling-guidelines>