Newborn screening

What is Newborn Screening?

When a baby is born, they have a series of tests in their first 6 to 8 weeks after birth to test for some rare conditions so that the baby can receive the most timely and effective treatment possible.

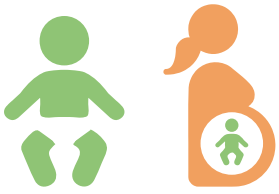
There are three types of tests:

Newborn physical examination

Newborn hearing screening

Newborn blood spot (heel prick) test

Newborn screening is different from antenatal (prenatal) screening which is carried out during pregnancy.



Learn more on the NHS website

The newborn blood spot (heel prick) test

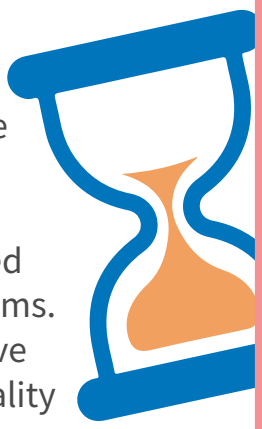
When a baby is about 5 days old, a healthcare professional will prick their heel and collect a few drops of blood on a special card. This is sent off for testing. The blood is tested for 9 rare conditions.

The blood spot test could identify that a child will develop a rare condition before the symptoms begin, which can give more treatment and planning options.

The 9 rare conditions screened for in the UK are:

- phenylketonuria (PKU)
- medium-chain acyl-CoA dehydrogenase deficiency (MCADD)
- maple syrup urine disease (MSUD)
- isovaleric acidaemia (IVA)
- glutaric aciduria type 1 (GA1)
- homocystinuria (pyridoxine unresponsive) (HCU)
- cystic fibrosis
- sickle cell disease
- congenital hypothyroidism

Why is the blood spot test important?

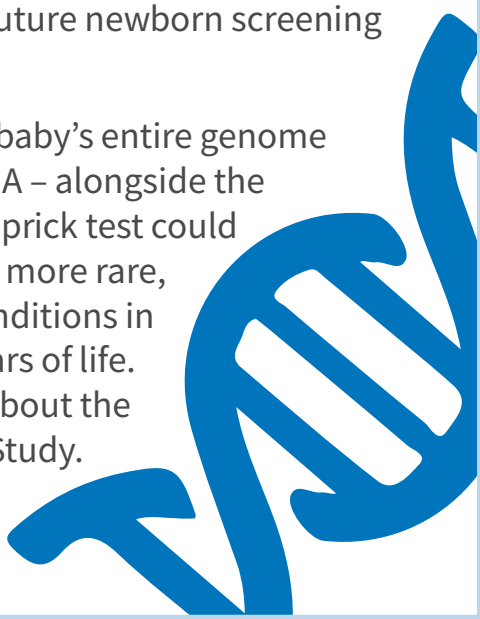


Early diagnosis of certain rare conditions can provide access to life-changing treatments which are most effective when administered before the onset of symptoms. They can drastically improve the life expectancy and quality of life for children with these conditions.

Blood spot tests also allow for the delivery of timely information to families and for the opportunity of choice and planning.

The future of newborn screening

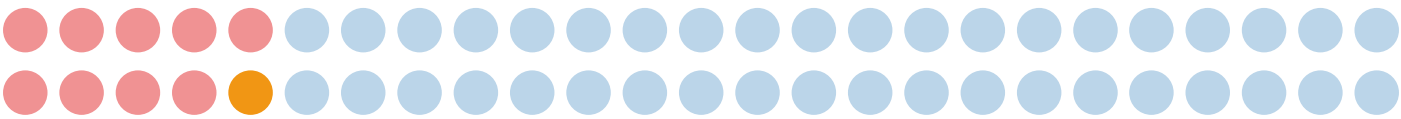
The Generation Study is an NHS-embedded research study which aims to understand whether sequencing babies' genomes can help to discover rare genetic conditions earlier. It aims to look at the DNA of over 100,000 babies and gather evidence to consider whether whole genome sequencing could be rolled out as part of a future newborn screening programme.



Screening a baby's entire genome – all their DNA – alongside the current heel prick test could detect many more rare, treatable conditions in their first years of life. Read more about the Generation Study.

The challenge

Newborn screening programmes in the UK test for much fewer conditions than programmes in other countries. The blood spot tests in the UK screens for **9 conditions** – soon to be **10** with the recent addition of tyrosinemia type 1. More than 20 European countries screen for more conditions than the UK, with most of them screening for more than 20 conditions whilst the US **screens for up to 50**.



The UK National Screening Committee (UK NSC) is responsible for assessing the evidence for national screening programmes and recommending which conditions should be screened for.

More information on newborn screening on the NHS website.

GENETIC ALLIANCE UK