Finding harmony in newborn blood spot screening

Supporting greater
certainty for families
of newborn babies.
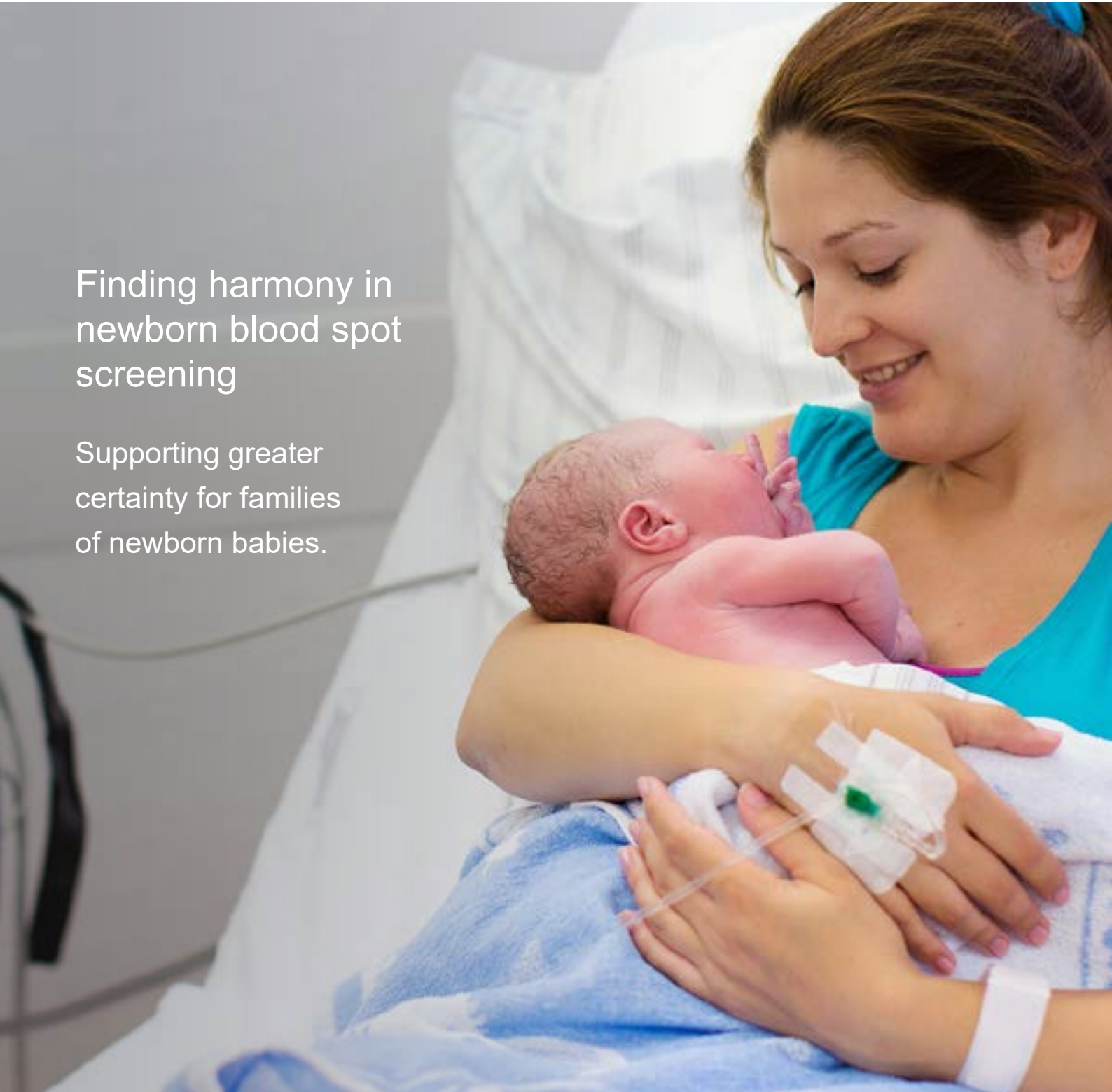
Background

Currently, all babies born in the UK are offered blood spot screening (heel prick test) in the first few days of life. This test detects nine conditions and inherited diseases, including cystic fibrosis, congenital hypothyroidism, and sickle cell disease. The goal is to detect and treat these rare conditions as early as possible, before they cause severe developmental problems and unnecessary suffering to the children.

Most babies (99 %) will have a normal result, which means it's unlikely that they have any of the conditions. However, a small number of babies will screen positive for one of the conditions and will be referred to a specialist for more tests. This is a very stressful and challenging time for the families. It is crucial to minimise the false positives – those where the secondary test results come back negative – as it has been shown that this experience can have significant long-term effects on the entire family, with new parents showing increased stress scores and greater numbers of hospital admissions in the first few years of the child's life.

With a baby born every 40 seconds in the UK, nearly 775,000 babies are tested each year using nationally agreed cut-off values for the different levels of hormones or amino acids in the blood. However, the 16 testing laboratories use a variety of different protocols and so ensuring harmonisation between them is vital.

Impact

We partnered with Dr Rachel Carling, Clinical Lead for Biochemical Sciences and Director of South East Thames Newborn Screening Laboratory, Viapath, Guys & St Thomas' NHS Foundation Trust, as part of the NHS England CSO's Knowledge Transfer Partnership (KTP) to investigate potential measurement improvements to support the Newborn Blood Spot Screening programme.

The KTP brought together the mass spectrometry expertise at the NML with Rachel's clinical experience as one of the country's foremost authorities on newborn screening. We helped further improve the methodology underpinning the screening programme, supporting harmonisation of the results. Through a workshop with the newborn screening laboratories, we discussed current best practice for statistical and mass spectrometry-based approaches to clinical measurement and are now developing a Best Practice Guide for use across the UK.

These improvements, leading to greater harmonisation of the methods used by the 16 laboratories involved in the screening programme, are aimed at reducing the possibility of false positives occurring. This will ensure parents are given the correct results, minimising unnecessary stress, and clinical resources are not wasted on unnecessary testing. The outcomes of this project will provide a framework within which more analytes can be added to the UK's screening programme to increase the range of diseases that can be tested for at birth.

Together, we are helping to deliver greater certainty for the Newborn Blood Spot Screening programme, impacting on every child born in the UK.

"I am delighted to be working with the National Measurement Laboratory, and by extending the collaboration to include the UK Newborn Screening Laboratory Network, I am confident that we can further improve the methodology which underpins the screening programme."

- Dr Rachel Carling, Consultant Clinical Scientist, Director of Service and Clinical Lead, Viapath, Guys & St Thomas' NHS Foundation Trust.

The **NHS Chief Scientific Officer's Knowledge Transfer Partnerships** (KTP) is a 12-month program that runs in partnership between NHS England, three UK National Measurement System (NMS) laboratories (LGC NML, NPL and NIBSC), UKAS and EPSRC FAST. It provides clinical leaders in healthcare science access to the unique capabilities within the National Measurement Laboratories to identify new approaches to measurement within their fields of expertise, ultimately helping to improve patient care.

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