

News release

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Evidence to support expanded screening of newborn babies in the UK

A new report produced by the PHG Foundation, and funded by the National Institute of Health Research - Collaboration for Leadership in Applied Health Research and Care - South Yorkshire (CLAHC-SY), examines the evidence for using modern screening technology to test newborn babies for a wider range of conditions.

Newborn screening tests babies shortly after birth for the presence of rare diseases where prompt medical intervention can protect them from serious and often irreversible harm. A technique called tandem mass spectrometry (MS/MS) was recently introduced to the UK newborn screening programme to allow testing for the inherited metabolic condition medium chain acyl-CoA dehydrogenase deficiency (MCADD). Improved treatment options and the recognition of the importance of early diagnosis has resulted in pressure from parent groups and clinicians to expand the range of conditions included in the newborn screen.

Inherited metabolic diseases (IMDs) are typically rare genetic defects that interfere with the function of a single protein involved in normal metabolic processing of foods in the body. These mutations can have devastating effects, causing a critical imbalance of products essential for normal body function, which in turn can lead to organ damage and death or severe physical and learning disabilities, often at a very early age.

The implementation of the MS/MS technique for screening makes it feasible to simultaneously test for the presence of additional rare inherited metabolic diseases for little additional cost. However, determining whether or not such screening would be beneficial is not straightforward, since these diseases are very rare, and can also be variable in their effects, making the gathering of robust evidence impossible. Despite this, a number of international programmes are now established with some reported success in reducing death and disability from these conditions.

The PHG Foundation report examines in detail all the available evidence to support the potential benefit of newborn screening for five inherited metabolic disorders considered to be strong candidates by scientific and medical experts:

- Maple Syrup Urine Disease
- Homocystinuria
- Glutaric Aciduria Type I
- Isovaleric Acidaemia
- Long-chain 3-hydroxyacyl CoA dehydrogenase deficiency (LCHADD)

The analysis considers the probable benefits, harms and costs of expanding newborn screening to include each of these five conditions. It concludes that there is potential to reduce death and severe disability caused by these conditions in a cost-effective manner. Experience from other programmes shows that there can, however, be downsides of screening, including some anxiety in parents that have an initial false positive result, and, occasionally identification of milder variants who might not need treatment. These effects can be minimized by good communications and rapid expert management of all who test positive.

Broadly, the Report concludes that a pilot study is required for a practical assessment of expanded newborn screening in the UK. Such a study should include not only measures to track cost-effectiveness, quality standards and patient outcomes following early diagnosis, but also take into account issues such as the availability of appropriate specialist care for babies diagnosed with these IMDs and the need for resources to support patients and families.

The Report, which was presented to the UK National Screening Committee for their consideration earlier this year, is freely available from the PHG Foundation website, www.phgfoundation.org

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Notes to Editors

PHG Foundation (www.phgfoundation.org)

- The PHG Foundation is a policy think-tank and health service development NGO based in Cambridge, UK.
- Our mission is *making science work for health* - identifying the best opportunities for 21st century genomic and biomedical science to improve global health, and to promote the effective and equitable translation of scientific innovation into medical and public health policy and practice.
- We provide knowledge, evidence and ideas to inform and educate, and to stimulate and direct well-informed debate.
- We also provide expert research, analysis, health services planning and consultancy services for governments, health systems, and other non-profit organisations.

The PHG Foundation is the working name of the Foundation for Genomics and Population Health, a charitable company registered in England and Wales, charity no. 1118664 / company no. 5823194.

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- The National Institute for Health Research (NIHR) Collaboration for Leadership in Applied Health Research and Care, South Yorkshire (CLAHRC-SY) is one of nine CLAHRCS across England, which commenced in October 2008 with funding for five years.
- The three primary functions of the CLAHRCS are the conduct of high quality applied health research; implementing findings from research in clinical practice; and increasing the capacity of NHS organisations to engage with and apply research, including continuing professional development for professionals and managers.
- CLAHRC-SY's mission is to undertake high quality, strategic, applied research and related education in order to enable a 'step change' in the way research is delivered and services are designed in South Yorkshire; and to foster knowledge transfer that will improve the quality and effectiveness of health care delivery across South Yorkshire.

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