UK NSC recommendation on newborn screening for organic acid oxidation disorders propionic acidaemia and methylmalonic acidaemia

Following a review of the evidence against strict criteria, the UK NSC does not currently recommend introducing a national screening programme for introducing a newborn screening programme for the two organic acid oxidation disorders propionic acidaemia (PA) and methylmalonic acidaemia (MMA).

These two rare conditions are both passed on from parents and mean that the child’s body is unable to break down parts of proteins called amino acids. This leads to a build-up of chemicals that can cause damage. The severity of symptoms and the age they develop can vary. It has been suggested that screening would be beneficial because early treatment can potentially prevent serious damage from happening.

Key findings supporting the UK NSC recommendation

• the quality of evidence from studies is not high enough to understand whether screening is of long term benefit
• not enough is known about PA to identify those who will be severely affected and those who will not
• it is also unclear from the guidelines how to treat babies who are detected with PA and MMA at screen that show no signs of being ill
• the screening test for PA and MMA detects other conditions that may not be suitable for screening

The UK NSC regularly reviews its recommendations on screening for different conditions in the light of new research evidence becoming available.

To find out more about the UK NSC’s newborn screening for organic acid oxidation disorders propionic acidaemia and methylmalonic acidaemia recommendation, please visit:

http://legacy.screening.nhs.uk/organicacidmetabolism

The UK National Screening Committee (UK NSC) advises ministers and the NHS in the 4 UK countries about all aspects of screening and supports implementation of screening programmes.

Find out more about the UK National Screening Committee at www.gov.uk/government/groups/uk-national-screening-committee-uk-nsc. The UK NSC evidence review process is described at www.gov.uk/government/publications/uk-nsc-evidence-review-process and a list of all UK NSC recommendations can be found at legacy.screening.nhs.uk/recommendations

The UK NSC secretariat is hosted by Public Health England (www.gov.uk/phe).