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UK NSC screening for LCHADD and MTP deficiency recommendation

Following a review of the evidence against strict criteria, the UK NSC does not currently recommend screening for Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHADD) / mitochondrial trifunctional protein (MTP) deficiency.

LCHADD and MTP deficiency are rare conditions which stop the body from changing some fats to energy. Babies with LCHADD and MTP deficiency can suffer from lack of energy, low blood sugar and feeding difficulties. If untreated, the condition can affect the body's organs and may cause early death. Specialist care and a low fat, high carbohydrate diet can help with the management of LCHADD and MTP deficiency.

Newborn blod spot screening happens 5 days after babies are born. Adding LCHADD and MTP deficiency to the newborn screening programme may help to identify children before symptoms begin. Dietary management could then begin earlier in the hope this may stop the baby from becoming very ill.

Key findings supporting the UK NSC recommendation

The results of this review are that:

- 1. LCHAD/MTP deficiency is very rare with about with only 5 or 6 babies affected each year
- 2. It is not clear what the best test is or whether screening on day 5 would identify affected babies before symptoms begin
- 3. People diagnosed before they have symptoms of LCHADD/MTP deficiency appear to be healthier than those treated after symptoms begin. But the studies which looked at this were small. Because of this, it is not certain that they are accurate.

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 LCHADD recommendation, please visit:

https://legacyscreening.phe.org.uk/lchadd

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/uknsc. 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).