UK NSC recommendation on newborn screening for biotinidase deficiency

Following a review of the evidence against strict criteria, the UK NSC does not currently recommend newborn screening for biotinidase deficiency

Biotinidase is an enzyme that the body needs to recycle a vitamin called biotin. Biotinidase deficiency is a rare genetic condition where people do not make enough of this enzyme. Babies can inherit the condition from their parents if both parents carry a mutation in the biotinidase gene. There are 2 forms of the condition: a severe or ‘profound’ deficiency and a milder ‘partial’ deficiency.

Without treatment, people with severe deficiency may develop problems with the nervous system, coordination problems and seizures. People with the milder deficiency may only develop symptoms if they have other illnesses, like an infection.

Biotin supplements are the highly effective, lifelong treatment, with no known side effects. Screening may help to identify individuals earlier before symptoms develop.

Key findings supporting the UK NSC recommendation

A national screening programme for biotinidase deficiency is not recommended in the UK because:

• it is not known how many children in the UK are affected
• it is still not clear if all screen-detected children need treatment
• the optimal screening test threshold and the timing of the test have not been clarified
• most children are treated at diagnosis so evidence is not available to inform which screen-detected children would develop symptoms and need biotin treatment, or the optimal dose to give
• it is not known whether screening improves outcomes

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 recommendation on newborn screening for biotinidase deficiency, please visit:

legacyscreening.phe.org.uk/biotinidasedeficiency

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).