



UK NSC amino acid metabolism disorders recommendation

Following a review of the evidence against strict criteria, the UK NSC does not currently recommend screening for the amino acid metabolism disorders; Tyrosinaemia type 1, Citrullinaemia (CIT) and Argininosuccinate lyase (ASL) deficiency in newborn babies

Tyrosinaemia type I, Citrullinaemia and Argininosuccinate lyase deficiency are amino acid metabolism disorders. They are rare conditions which are caused by altered genes from both the mother and the father. The three conditions can disrupt the processes through which amino acids (the building blocks of proteins) are broken down. These disorders lead to a build-up of harmful chemicals that can cause damage to vital parts of the body, such as the liver.

Newborn screening has been suggested as it might find babies with these disorders before they become ill with the aim of treating the baby to improve its health and the experience of the family.

Key findings supporting the UK NSC recommendation

- Babies with CIT often develop symptoms before the results of the screening test would be available. And the test would falsely identify some healthy babies as having the condition.
- The long term effects of ASL Deficiency, such as liver disease and brain damage, appear to be unaltered by early treatment.
- The review, and public consultation, suggested that screening for tyrosinaemia type 1 may be possible. The UK NSC is undertaking further work to explore the issues raised in the consultation. This includes evaluating the accuracy of the test and effectiveness of the treatment.

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 amino acid metabolism disorders recommendation, please visit:

www.screening.nhs.uk/aminoacidmetabolism

The UK National Screening Committee (UK NSC) is chaired by the Deputy Chief Medical Officer for England, advises Ministers and the NHS in the four UK countries about all aspects of screening and supports implementation of screening programmes. Using research evidence, pilot programmes and economic evaluation, it assesses the evidence for programmes against a set of internationally recognised criteria covering the condition, the test, the treatment options and the effectiveness and acceptability of the screening programme. Assessing programmes in this way is intended to ensure that they do more good than harm at a reasonable cost. The UK NSC also sets up practical mechanisms to oversee the introduction of new programmes in the English NHS and monitors effectiveness and quality of these programmes.

Find out more about the UK National Screening Committee at www.screening.nhs.uk. The UK NSC evidence review process is described at www.screening.nhs.uk/policyreview and a list of all UK NSC recommendations can be found at www.screening.nhs.uk/recommendations

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