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