



UK NSC recommendation on screening for tyrosinaemia type 1 (TYR1) in newborns

Following a review of the evidence against strict criteria, the UK NSC does not currently recommend screening for tyrosinaemia type 1.

TYR1 is a very rare, inherited condition which prevents the body breaking down an amino acid called tyrosine from food. This leads to the build-up of toxic substances in the blood. If these are left untreated they can cause damage, particularly to the liver, kidneys and the nervous system.

TYR1 can occur as:

- early onset, within the first months of life
- chronic, which is slower to develop

The treatment for TYR1 is a drug called nitisinone combined with a strict diet. A liver transplant is performed in patients who do not respond to nitisinone or who develop liver cancer. Newborn screening has been suggested to identify babies with TYR1 before they become ill in order to give early treatment.

Key findings supporting the UK NSC recommendation

The most recent evidence review showed that the screening test finds most babies with TYR1. But there is not enough information available to know if the test misses affected babies.

Treatment with nitisinone results in improved outcomes in TYR1 cases. But there were a number of problems in how the research had been carried out which made it difficult to be sure that there was more benefit from early rather than late treatment.

The UK NSC is continuing to work with stakeholders to understand more about these issues. The Committee will commission a modelling exercise to estimate whether screening is more beneficial than prompt treatment of babies with nitisinone following the start of symptoms.

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 tyrosinaemia type 1 recommendation, please visit:

legacyscreening.phe.org.uk/aminoacidmetabolism

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).