UK NSC Gaucher disease recommendation

Following a review of the evidence against strict criteria, the UK NSC does not currently recommend universal screening for Gaucher disease in newborn babies.

Gaucher disease is a rare inherited genetic condition which can affect a wide range of the body’s organs and tissues. The signs and symptoms vary and currently the condition is divided into three classifications (type 1, 2 and 3) based on the severity and age that symptoms develop.

There is evidence that enzyme replacement therapy (ERT) is effective in treating the symptoms of type 1 Gaucher disease. However it is not clear whether earlier treatment brings any benefit over later treatment once symptoms have developed.

A screening programme would also identify newborn babies with type 2 and type 3 Gaucher disease.

Key findings supporting the UK NSC recommendation

- There are a number of uncertainties with Gaucher disease; specifically around predicting how severely an individual, detected through screening, might be affected by the condition. It is not currently possible to identify who will be severely affected by the condition and who will never experience any problems.

- It is unclear whether earlier treatment following a screening test would be more beneficial than current medical practice of identification and treatment when symptoms develop.

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 Gaucher disease recommendation, please visit:  

www.screening.nhs.uk/gauchers