



UK NSC screening for Gaucher disease recommendation

Following a review of the evidence against strict criteria, the UK NSC does not currently recommend introducing a national screening programme for Gaucher disease in newborns.

Gaucher disease is a rare genetic disease. There are 3 types of Gaucher disease (type 1, type 2 and type 3). Most people with Gaucher disease have type 1 disease.

Symptoms can range from very mild, to life threatening. Gaucher disease can cause death in early childhood.

Typical symptoms include enlarged liver and spleen, bone pain, fatigue, and easy bruising. Symptoms can appear in childhood or adulthood, and some people with type 1 Gaucher disease remain unaffected.

Individuals who develop symptoms in early childhood generally go on to have more severe symptoms than those who present later in adulthood.

Enzyme replacement therapy (ERT) and substrate reduction therapy (SRT) are used to reduce the severity of symptoms in children with type 1 Gaucher disease.

Key findings supporting the UK NSC recommendation

This review looked at whether children with type 1 Gaucher disease benefit from treatment before symptoms develop.

The review did not find any research into whether there is benefit in treating patients with type 1 Gaucher disease before symptoms develop. Because of this, it is unclear whether earlier treatment after screening, but before symptoms develop, would deliver more benefit than later treatment after 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:

<https://legacyscreening.phe.org.uk/gauchers>

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).