



UK NSC screening for spinal muscular atrophy recommendation

Following a review of the evidence against strict criteria, the UK NSC does not currently recommend introducing a national screening programme for carrier or newborn screening for spinal muscular atrophy.

Spinal muscular atrophy (SMA) is a genetic disease that makes muscles weak. It gets worse over time. SMA can be fatal if it affects the muscles that control breathing. This is due to progressive degeneration of nerve cells called alpha motor neurons in the spinal cord, which are required to transmit impulses from the brain to the muscles in the chest and limbs. There are 5 different types of SMA, from type 0 (the most severe which affects newborn babies and it is often fatal before the age of 6 months) to type 4 (stable and mild disease, affects adults and usually only causes mild problems). About half of people with SMA have type 1.

Key findings supporting the UK NSC recommendation

- there was very limited evidence about how acceptable a screening programme would be
- there was no evidence on how to support individuals who need to make difficult decisions following carrier screening
- there was a lack of information on the reliability of screening tests for SMA
- no effective treatments for SMA were identified in an unsymptomatic population, currently only palliative support can be offered - however, the review identified evidence on a new treatment for SMA, called nusinersen, that can improve symptoms in children with SMA

However, this evidence review did not find information on the effectiveness of nusinersen in children without symptoms, and there is no evidence on the long term effects of this drug.

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 SMA recommendation, please visit:

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

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).