Press release: Screening consultation on rare but serious condition in babies

The UK's independent expert screening committee is today (4 August 2017) launching a consultation to look at whether screening for Severe Combined Immune Deficiency (SCID) in babies should be tried within the NHS.

SCID is a rare inherited condition which makes it more difficult for babies to fight off infections. An infection is not serious for most babies but can be life-threatening for those with SCID, with around 15 to 25 babies born with the condition every year.

The consultation findings will provide valuable information about whether SCID should be added to the existing NHS newborn blood spot screening programme, which currently checks for 9 rare but serious health conditions by taking a blood sample from a baby's heel.

Screening for SCID would use blood from the current heel prick test to check if a baby has a low white blood cell count, which may make them more likely to have infections.

Before a decision can be taken, more needs to be known about:

- whether screening will save lives
- the number of healthy babies found to have low numbers of white blood cells
- what care and treatment is best for babies who are found to have low numbers of white blood cells for reasons other than SCID

Dr Anne Mackie, Director of Programmes for the UK National Screening Committee (UK NSC), said:

> We currently test babies for 9 rare but serious health conditions as part of the NHS newborn blood spot screening programme. This consultation will consider key organisations' and the public's views on how testing for SCID would work practically within the NHS. We need this information before the Screening Committee can make a recommendation on including SCID as part of the newborn programme.

The consultation will run until early November.

The committee also recommended against screening pregnant women for vasa praevia, a rare condition that can cause heavy bleeding during birth. It had been suggested that screening all pregnant women for the condition could be useful in finding which women might benefit from a caesarean.

Screening was not recommended for a number of reasons, including concerns over the accuracy of the test and that screening would find other, more common, conditions which could lead to a larger number of pregnancies being wrongly considered at risk. The inaccuracy of the test means that some women would be offered an early caesarean when unnecessary.

As part of a regular review process all of the recommendations will be looked at again in 3 years – or earlier if significant new evidence becomes available.

The latest screening recommendations were made at the UK NSC's meeting on 23 June 2017. Read the minutes which were published today (4 August 2017).

All recommendations will now be considered by ministers.

This press release is issued on behalf of the UK NSC, an independent organisation, by Public Health England (PHE). For more information please call the PHE screening press office:

1. The UK National Screening Committee is independent of, but supported by PHE.
2. Information on how to participate in the public consultation on SCID is available at: legacyscreening.phe.org.uk/screening-recommendations.
3. The committee did not recommend screening for 2 further conditions: tyrosinaemia type 1 (TYR1) in newborns and thrombophilia in all ages.
    - TYR1 is a very rare, inherited condition which prevents the body breaking down an amino acid called tyrosine from food which can lead to the build-up of toxic substances in the blood. If these substances are left untreated they can cause damage, particularly to the liver, kidneys and the nervous system. Screening was not recommended because although the screening test finds most babies with TYR1, there is not enough information to know if the test misses other affected babies. The UK NSC is continuing to work with stakeholders to understand more about the issues.
    - Thrombophilia increases a person's risk of developing blood clots which can be dangerous if they break away and block blood flow to important organs such as the heart, lungs or brain. The condition can cause stroke, heart attack and deep vein thrombosis. Screening pregnant women was considered because women with thrombophilia may be at increased risk of blood clots in their leg, complications such as high blood pressure or the birth of an early or small baby. However, there was no evidence of benefit compared with current practice and inconclusive findings on the safety and effectiveness of treatment. Screening was also not recommended for newborn babies and adults as there was insufficient evidence to suggest that a screening programme would offer more benefit than current practice. There was also not enough evidence about how good the treatment is.
4. The blood spot test screens for the following 9 rare but serious conditions:
    - sickle cell disease
    - cystic fibrosis
    - congenital hypothyroidism

    - phenylketonuria (PKU)
    - medium-chain acyl-CoA dehydrogenase deficiency (MCADD)
    - maple syrup urine disease (MSUD)
    - isovaleric acidaemia (IVA)
    - glutaric aciduria type 1 (GA1)
    - homocystinuria (pyridoxine unresponsive) (HCU)
5. Detailed summaries of the recommendations for all of the conditions mentioned can be found at legacy.screening.nhs.uk/screening-recommendations.
6. Public Health England exists to protect and improve the nation's health and wellbeing, and reduce health inequalities. It does this through world-class science, knowledge and intelligence, advocacy, partnerships and the delivery of specialist public health services. PHE is an operationally autonomous executive agency of the Department of Health. Follow us on Twitter: @PHE_uk and our blog phescreening.blog.gov.uk.