14/388

UK National Screening Committee

Screening for Congenital Adrenal Hyperplasia in Children

19 November 2015

Aim

1. To ask the UK National Screening Committee to make a recommendation, based upon the evidence presented in this document, whether or not screening for Congenital adrenal hyperplasia (CAH) meets the NSC criteria to support the introduction of a population screening programme.

   This document provides background and reviews the evidence on screening for CAH in newborns.

Current Recommendation

2. The current UK NSC recommendation, developed in 2012, is that universal screening for CAH in newborns is not recommended.

   This recommendation is based mainly on the conclusions from a systematic review (Khalid et al., 2010) undertaken on behalf of the UKNSC. The systematic review highlighted uncertainties around the screening test, with additional concerns about the UK prevalence and acceptability of the test.

This Review

3. This condition is being reviewed as part of the UK NSC's triennial cycle of reviews. Dr Glen Wilson was commissioned to do the review in December 2014. The 2015 UK NSC update focuses on the three clinical questions highlighted in the 2012 review.

4. The conclusion of this current review is that population screening for CAH in newborns should not be recommended. The key findings to support this conclusion have been summarised below:

    a. <u>The condition</u>; A British Paediatric Surveillance Unit (BPSU) study estimates the UK incidence of CAH to be approximately 1:18000 births. This is within the range reported internationally. However the higher incidence of CAH in those of Asian ethnicity in the UK was not a finding identified elsewhere. This, and the study's relatively short duration, introduces some uncertainty into the review's evaluation of the evidence relating to the epidemiology. **Criterion 1 is partially met.**

    b. <u>The test</u>; There is uncertainty over the suitability of the screen test with evidence suggesting the test has poor accuracy in positively predicting cases of CAH. Due to this uncertainty, the test does not meet the criteria to be considered a simple, safe, precise and validated screening test. **Criterion 5 not met.**

Consultation

5. A three month consultation was hosted on the UK NSC website. Communication of the consultation was promoted through both PHE Events and the PHE Screening Twitter platform. Direct emails were sent to stakeholders of whom 10 organisations were contacted directly. Annex A

6. One response was received from the consultation from the Royal College of Paediatrics and Child Health (RCPACH), which is attached in Annex B.

The comments were supportive of the review conclusion but suggested that amendments to the plain English summary may help readability.

Recommendation

7. The committee is asked to approve the following recommendation:

Universal screening for Congenital adrenal hyperplasia in newborns is not recommended

The absence of a suitable screening test is significantly detrimental to the viability of a screening programme.

Based upon the 22 point criteria set to recommend a population screening programme, CAH screening in newborns did not meet the following primary requisites:

	Criteria	Met/Not Met/ Partially Met
Section 1 - Criteria for appraising the viability, effectiveness and appropriateness of a screening programme		
The Condition		
2.	The epidemiology and natural history of the condition, including development from latent to declared disease, should be adequately understood and there should be a detectable risk factor, disease marker, latent period or early symptomatic stage	Partially Met
The Test		

5.	There should be a simple, safe, precise and validated screening test	Not Met (clear evidence not met) 
6.	The test should be acceptable to the population	Not Met (evidence uncertain) 
The Screening Programme		
14.	There should be evidence that the complete screening programme (test, diagnostic procedures, treatment/ intervention) is clinically, socially and ethically acceptable to health professionals and the public.	Not Met (evidence uncertain) 

UK National Screening Committee

14/388

Annex A

List of stakeholders who were contacted directly;

1. Children Living with Inherited Metabolic Diseases
2. Clinical Genetics Society
3. Genetic Alliance UK
4. Rare Disease UK
5. Royal College of Midwives
6. Royal College of Paediatrics and Child Health
7. Save Babies Through Screening Foundation UK
8. NHS England
9. Genomics England
10. Department of Health rare diseases team.

UK National Screening Committee

14/388

Annex B – CAH Consultation Comments

1.

Name:	Comments provided on behalf of the following: • Dr Eugen Strehle • Dr Jane Hawdon	**Email address:**	**xxxx xxxx**
Organisation (if appropriate):	Royal College of Paediatrics and Child Health		
Role:	**NA**		
Do you consent to your name being published on the UK NSC website alongside your response? **Yes** ☒ **No** ☐			

Section and / or page number	Text or issue to which comments relate	
All		We agree with the conclusion that currently screening for CAH is not beneficial for all newborns.
All		We feel that the plain English section is not at all plain English for the lay person.