

UK National Screening Committee

Is there evidence to alter the current UKNSC recommendation to offer a national screening programme for cystic fibrosis in newborn babies? A pilot of the triage approach.

Topic: Newborn screening for cystic fibrosis (CF)

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1. Background to the triage reports

This report is a rapid (triage) assessment of evidence relating to whether the existing national screening programme (NSP) for cystic fibrosis in newborns should be continued.

For conditions for which population screening programmes are recommended by the National Screening Committee (NSC) the triage process focuses on whether there is new evidence suggesting that the NSP should be stopped.

It consists of an externally produced report on a literature search undertaken to identify whether any papers have been published:

- addressing screening programme cessation
- reporting harms from screening
- reporting balance of harms and benefits from screening

The aim of these reports is to identify any “red flags” that suggest that an NSP needs to be reviewed in greater detail. They do not aim to identify all new literature relating to screening for the condition; instead they focus specifically on evidence relating to the three areas specified above.

If no papers are identified on the above, a recommendation to continue the programme is made. If papers on programme cessation or harms from screening are identified, the UK NSC will consider whether further work is necessary before making a final recommendation on the topic.

Stakeholders will be contacted for comments on the recommendation and a three month consultation will be hosted on the UK NSC website.

Based on the triage report and stakeholder comments the Committee decides whether to recommend that the issue is considered in more depth. Where further evaluation is considered appropriate, the options may include primary research, systematic review, cost effectiveness assessment, modelling or further rapid reviews.

2. Executive summary

This triage assessment identified one study with potential relevance to the three questions above, and that related to the possible harms of cystic fibrosis (CF) screening. This study discussed the effect of early diagnosis with CF on parental depression.¹

There were no studies identified which reported on the cessation or reported on the balance of harms and benefits of the newborn CF screening programme. One small study suggested that earlier diagnosis of CF may increase the risk of parental depression, but it was unclear from the abstract whether this earlier diagnosis was a result of screening rather than clinical diagnosis, or whether the results might have been influenced by the child's level of symptoms.

Recommendation: The findings do not provide sufficient evidence to suggest that the evidence supporting the national newborn CF screening programme needs to be reviewed in more depth or that the programme should be stopped.

3. Introduction to the condition

The current NSP being assessed is newborn bloodspot screening for cystic fibrosis (CF).

Cystic fibrosis is a genetic condition where the lungs and the digestive system are clogged with a thick sticky mucus. The mutation causing the condition is found in the cystic fibrosis transmembrane conductance regulator (CFTR) gene which, which normally regulates the levels of sodium and chloride in cells. The defect results in the sticky mucus which damages the lungs, digestive system and other organs. This damage causes the symptoms of cystic fibrosis.

CF is one of the diseases currently screened for as part of the NHS newborn blood spot (NBS) screening programme. This is offered for all newborn babies, and blood is usually taken 5 days after birth (in exceptional cases it can be taken between Day 5 and Day 8). The screening test for CF examines level of blood immunoreactive trypsinogen (IRT).

Early diagnosis allows the maintenance of optimal nutrition which may improve the long-term prognosis. Babies with CF are treated as soon as they are diagnosed, the aim is to improve nutrition with the use of supplements containing enzymes to aid digestion and to reduce chest infections with physiotherapy and antibiotics. Such treatment can slow down the effects of CF.

Screening for CF is performed by an initial analysis of the levels of IRT. Babies identified as higher risk of CF based on their screening result will go on to have DNA testing to look for CF-causing alterations in the CFTR gene.

This external review has searched the literature published between 2006 up to February 2016, and reviewed at title and abstract level whether there is evidence:

- indicating that other countries have terminated CF screening
- reporting harms from CF screening
- reporting balance of harms and benefits from CF screening

4. Description of the evidence

Fifty-two publications were selected at the first pass sift as being potentially relevant to these three questions based on title and abstract. These were reviewed more closely at abstract level at a second pass appraisal.

There were no studies identified which reported on the cessation or reported on the balance of harms and benefits of the newborn CF screening programme. The excluded studies were mainly surveys of screening and management practice (including in the UK), studies of parental experiences of screening, health technology assessments and cost effectiveness studies.

One study reported on parental depression following early diagnosis of CF.¹ The findings were that parents may be more likely to experience depression if their child is diagnosed with a life shortening condition within the first few months of life (9 months or younger). This implied that those with a later diagnosis were less likely to experience depression, but it was not clear from the abstract whether the figures presented were a comparison versus the control group of parents of children without CF, or versus those with a later diagnosis of CF. The abstract also did not specify whether children were screen or clinically detected, or whether parents would still prefer to have an earlier diagnosis.

Of the publications that did not meet the inclusion criteria, some did raise issues relating to harms, but did not directly indicate harms of screening or a balance of harms against benefits that suggested programme cessation should be considered.

One was a qualitative assessment of the psychosocial impact on parents and families of infants receiving a false positive screening result.² The findings were that genetic tests with abnormal results can affect the whole family. The paper did not offer any indication of the number of parents or families negatively affected by false positive results or whether the parents felt this outweighed any benefit of the screening programme. There were a number of other studies that looked at the impact on parents of a positive or false positive result, with some of these suggesting that parents were mostly satisfied with the screening process,^{3, 4} and would go ahead with the test for another child (even if their newborn had received a false positive result),⁵ and that the anxiety in parents of newborns with a false positive result does not cause long term anxiety after the child is found not to have CF.^{5, 6}

One report discussed the balance of risks and benefits of a CF newborn screening programme and the implications for policy in the US.⁷ This report mainly noted that the balance “not tipped dramatically in 1 direction” so mandatory screening of all newborns might not be appropriate, and discusses modifications which might affect or improve the balance. This was not included as it did not state that harms outweighed benefits, merely highlighted influencing factors.

A few papers reported on the prevalence and features of infants receiving an “indeterminate/ uncertain diagnosis” where test results fall into a borderline or uncertain range and the infant is asymptomatic. This is sometimes referred to as CFTR-related metabolic syndrome (CRMS). These infants are asymptomatic, but their long term prognosis is not known and some develop *Pseudomonas aeruginosa* infection. For this reason, the papers generally suggested that they need monitoring and treatment depending on results,⁸⁻¹¹.

Table 1 Details of relevant studies identified

| Publication details | Study details | Population | Intervention/ test and comparator | Main findings | Comments |
|---|--|--|---|--|---|
| Screening programme cessation | | | | | |
| No studies identified | | | | | |
| Harms from screening | | | | | |
| Glasscoe et al. 2007 ¹ | Matched cohort study in north west England | 45 couples with a child diagnosed with CF were compared with 45 control couples matching for age, sex, and position in the family of the index child | Parental depression based on the Beck Depression Inventory (BDI-II) with a clinical cut-off ≥ 13 for dysphoria (mild depression) | The study suggests “Parents with a child with CF ≤ 9 months of age at baseline had an elevated prospective risk of depression.” Estimated risk ratios were 2.6 for mothers (95% CI 1.05 to 6.42), and 2.26 for fathers (0.97 to 5.28) | <ul style="list-style-type: none"> • There was no indication in the abstract of whether children were screen or clinically detected • Many children with CF are likely to be clinically diagnosed within the first year of life whether screen detected or not (particularly those with more severe symptoms). • The severity of the child's symptoms may be linked to parental depression, and it was not clear from the abstract whether this was taken into account. • Not screening may only prevent detection at <1 yr of children with less severe symptoms, and parents of these less severely affected children may be less likely to experience depression. |
| Balance of harms and benefits from screening | | | | | |
| No studies identified | | | | | |

5. Methodology

It is intended that the triage process for each NSP will be performed every three years. This review is the first triage review for CF and includes literature published in the last 10 years.

Sifting was carried out in two stages. The first pass sift was conducted by an information specialist at title and abstract level, to remove clearly non-relevant material e.g. animal studies, or studies of different screening programmes. The second pass sift was performed by a health research analyst and this sift examined the results more closely at title and abstract level to remove those studies clearly not relevant, and select those meeting inclusion criteria for summary.

The reports focus on high quality studies, i.e. systematic reviews, randomised controlled trials, non-randomised controlled trials, cohort studies or screening programme evaluations that appear at abstract level to have covered potential harms of the NSP, the balance of harms and benefits, or screening programme cessation. Lower level evidence such as case series and case reports, non-systematic reviews, editorials or opinion pieces are not included unless they clearly highlight potential harms of the NSP indicating the need for further evaluation.

Studies on any issues other than the three questions of interest are not included. For example, studies examining cost effectiveness (unless relevant to the UK and highlighting the balance of benefits and harms), or studies assessing modifications to an existing screening programme (e.g. changing age at screening, screening test used, screening interval etc.) would be excluded. Studies evaluating management of the condition are also excluded - unless they indicate that the existing treatment is ineffective or harmful, which may suggest that harms of screening outweigh any benefits.

These triage reports are rapid assessments to identify any “red flags” which indicate the need for further assessment of the NSP. They are complemented by consultation with stakeholders to identify any additional issues which may not be represented in the literature identified.

6. Search strategy

We searched the following bibliographic databases:

- Medline (via Embase.com)
- Embase
- The Cochrane Library: including the Cochrane Database of Systematic reviews; Cochrane Central Register of Controlled Trials (CENTRAL); Database of Abstracts of Reviews of Effects (DARE); Health Technology Assessment Database (HTA); NHS Economic Evaluation Database (EED)

The searches were limited by date to include studies published since 2005. No language limits were used. Methodological filters were not used as they would not have been appropriate given the focus of the research questions.

The search strategy was developed through testing to identify the best balance between sensitivity and specificity that was fit for purpose. The search strategy used both indexing terms and text words as relevant records could have been indexed in different ways (or not indexed at all). The Embase search strategy was translated for the other databases and adapted to take into account the databases size, coverage and available indexing terms.

The search strategy was based on the PICO framework and combined three major concepts: the population (condition), neonatal screening, and harms from screening or screening programme cessation. (See table below)

Search strategy (Embase.com)

| Concept | Search strategy |
|---------------------|---|
| Population | <ol style="list-style-type: none"> 1. 'cystic fibrosis'/de 2. 'cystic fibrosis':ab,ti 3. cf:ab,ti OR mucoviscidosis:ab,ti 4. ((fibrocystic OR fibrosis) NEAR/3 pancreas):ab,ti 5. cystic:ab,ti AND pancre*:ab,ti 6. 1 or 2 or 3 or 4 or 5 |
| Screening | <ol style="list-style-type: none"> 1. 'newborn screening'/de 2. ((neonat* OR newborn*) NEAR/2 screen*):ab,ti 3. 'mass screening'/de 4. 'newborn'/de 5. 3 and 4 6. 1 or 2 or 5 |
| Programme cessation | <ol style="list-style-type: none"> 1. ceas*:ab,ti OR cessation:ab,ti OR stop:ab,ti OR stopped:ab,ti OR continu*:ab,ti OR discontinu*:ab,ti 2. appropriate*:ab,ti OR inappropriate*:ab,ti OR unnecessary:ab,ti OR question*:ab,ti 3. harmful:ab,ti OR harm*:ab,ti OR adverse:ab,ti 4. benefit*:ab,ti AND (risk*:ab,ti OR harm*:ab,ti) 5. 'side effect'/exp) 6. (side NEAR/1 effect*):ab,ti 7. overdiagnosis:ab,ti OR 'over diagnosis':ab,ti 8. 'patient safety'/exp 9. 'risk assessment'/de 10. 'risk benefit analysis'/exp 11. 1 or 2 or 3 or 4 or 5 or 6 or 7 or 8 or 9 or 10 |

Search results

| Databases searched | Dates searched | Number of hits |
|---|-----------------|----------------|
| Medline and Embase (Embase.com) | 2005-24/02/2016 | 406 |
| Cochrane Database Syst Rev (Cochrane Library) | 2005-24/02/2016 | 4 |
| CENTRAL (Cochrane Library) | 2005-24/02/2016 | 39 |
| NHS EED (Cochrane Library) | 2005-24/02/2016 | 5 |
| HTA (Cochrane Library) | 2005-24/02/2016 | 6 |
| Total number of hits | | 460 |
| Total number after de-duplication | | 435 |
| Total number after first appraisal | | 52 |

Embase.com search strategy

| | | |
|-----|--|-----------|
| #1 | 'newborn screening'/de | 13,746 |
| #2 | ((neonat* OR newborn*) NEAR/2 screen*):ab,ti | 12,206 |
| #3 | 'mass screening'/de | 49,630 |
| #4 | 'newborn'/de | 498,406 |
| #5 | #3 AND #4 | 2,463 |
| #6 | #1 OR #2 OR #5 | 19,026 |
| #7 | ceas*:ab,ti OR cessation:ab,ti OR stop:ab,ti OR stopped:ab,ti OR continu*:ab,ti OR discontinu*:ab,ti | 1,265,546 |
| #8 | appropriate*:ab,ti OR inappropriate*:ab,ti OR unnecessary:ab,ti OR question*:ab,ti | 1,495,204 |
| #9 | harmful:ab,ti OR harm*:ab,ti OR adverse:ab,ti | 609,591 |
| #10 | benefit*:ab,ti AND (risk*:ab,ti OR harm*:ab,ti) | 166,742 |
| #11 | 'side effect'/exp | 398,996 |
| #12 | (side NEAR/1 effect*):ab,ti | 268,067 |
| #13 | overdiagnosis:ab,ti OR 'over diagnosis':ab,ti | 3,449 |
| #14 | 'patient safety'/exp | 68,643 |
| #15 | 'risk assessment'/de | 369,811 |
| #16 | 'risk benefit analysis'/exp | 43,498 |
| #17 | #7 OR #8 OR #9 OR #10 OR #11 OR #12 OR #13 OR #14 OR #15 OR #16 | 3,964,755 |
| #18 | #6 AND #17 | 4,066 |
| #19 | 'cystic fibrosis'/de | 56,075 |
| #20 | 'cystic fibrosis':ab,ti | 49,246 |
| #21 | cf:ab,ti OR mucoviscidosis:ab,ti | 50,390 |
| #22 | ((fibrocystic OR fibrosis) NEAR/3 pancreas):ab,ti | 806 |
| #23 | cystic:ab,ti AND pancre*:ab,ti | 12,286 |
| #24 | #19 OR #20 OR #21 OR #22 OR #23 | 96,127 |
| #25 | #18 AND #24 | 513 |
| #26 | #18 AND #24 AND [2005-2016]/py | 406 |

Cochrane Library search strategy

| | | |
|-----|--|-------|
| #1 | MeSH descriptor: [Cystic Fibrosis] this term only | 1178 |
| #2 | (cystic next fibrosis):ti,ab,kw | 3500 |
| #3 | (cf or mucoviscidosis):ti,ab | 2303 |
| #4 | ((fibrocystic or fibrosis) near/3 pancreas):ti,ab | 6 |
| #5 | (cystic and pancre*):ti,ab | 280 |
| #6 | #1 or #2 or #3 or #4 or #5 | 4409 |
| #7 | MeSH descriptor: [Neonatal Screening] this term only | 287 |
| #8 | ((neonat* or newborn*) near/5 screen*):ti,ab | 446 |
| #9 | MeSH descriptor: [Mass Screening] this term only | 4625 |
| #10 | MeSH descriptor: [Infant, Newborn] explode all trees | 14298 |
| #11 | #9 and #10 | 123 |
| #12 | #7 or #8 or #11 | 628 |
| #13 | #6 and #12 Publication Year from 2005 to 2016 | 54 |

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Included after second pass sift (n=1)

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