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**Title: Newborn screening information supports public health more than informed choice**

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**Abstract**

**Objective:** To appraise information resources on newborn blood spot screening currently available for parents and health professionals internationally.

**Method:** Health information on newborn blood spot screening was sourced internationally through the internet and, in the United Kingdom, through health service providers and support organisations. An appraisal tool was developed based on DISCERN criteria for evaluating information about patient treatment choices and UK National Screening Committee criteria for determining whether a screening programme should be implemented. Two researchers independently applied the appraisal tool to all the leaflets describing the heel prick, then discussed and resolved discrepancies.

**Results:** The majority of leaflets supported the public health agenda by informing parents of the benefits of screening, including the significance of early detection and treatment for these conditions. Few leaflets, however, supported the informed choice agenda by mentioning either the limitations of screening, or choice, or by being easy to read.

**Conclusion:** Most health information about newborn blood spot screening portrays it in a positive light, and relatively few leaflets address the difficulties openly. We

recommend the use of criteria for high-quality patient information alongside topic or condition-specific guidelines as a starting point for developing patient information.

### **Introduction**

Population screening programmes are regarded as important public health priorities (1;2). However, screening programmes have their limitations in that they may provide false reassurance or anxiety through inaccurate results, or difficulties with equivocal results or results that label individuals with an inconsequential condition or genetic carrier status (3). The public health approach emphasises high uptake of screening rather than informed participation (4;5). New policies supporting realistic expectations and informed choice in healthcare, however, are driving the need for good quality research-based information for patients (6). Facilitating realistic expectations and/or informed choice means increasing openness about the difficulties and limitations as well as the benefits of screening, and providing understandable information for participants.

A changing balance in policy between supporting public health priorities and encouraging realistic public expectations or informed choice for screening is evident since the National Screening Committee for England (NSC) published its first criteria for appraising the viability, effectiveness and appropriateness of a screening programme (1). The NSC's additional criteria in the second report are included to assist people further in making informed choices about screening (2). Criteria added

in a more recent report of the NSC's Director relate specifically to the identification of unaffected carriers of the conditions and genetic testing (7).

All such issues are now being addressed by the UK Newborn Screening Programme Centre, funded by the Department of Health on behalf of all UK countries since April 2002, including the provision of information for parents of newborn babies. All newborns in the UK are offered screening for the serious but rare conditions of phenylketonuria (PKU) and congenital hypothyroidism and, in some areas, sickle cell disorders and/ or cystic fibrosis. This involves a heel prick to collect a series of bloodspots for laboratory testing. Parents are to be given evidence-based information, explaining the consequences of testing, investigation and treatment, as recommended by the National Screening Committee to assist them in making an informed choice about screening their baby (7).

This paper reports the findings of an international survey of leaflets on newborn blood spot screening for parents and health professionals, carried out as part of the process of developing evidence-based information resources (8).

## **Methods**

We sourced leaflets and information sheets through internet websites, health service providers, including screening laboratories, other screening programme co-ordinators such as the NHS Sickle Cell and Thalassaemia Screening Programme (9),

and parent support organisations. To match our objective of preparing pre-screening parent information, we included in the survey only those leaflets that described the process of taking the blood spots from a baby's heel, excluding leaflets that focussed only on the condition identified through screening.

To judge whether leaflets contained information about the difficulties as well as the benefits of screening, we developed an appraisal tool. The questions contained in the tool were adapted from DISCERN criteria for appraising information about patient treatment choices(10), and National Screening Committee criteria for determining whether a screening programme should be implemented (1;2;7). Two researchers refined the tool by applying it to a small number of leaflets until consensus was reached. The finalised tool was entered into specialist software, EPPI-Reviewer (11), and used to evaluate each leaflet (see Box 1).

Two researchers appraised each leaflet using the finalised appraisal tool, compared answers, and addressed discrepancies to reach consensus on the answers to each question. Using EPPI-Reviewer, we analysed how many leaflets addressed the positive aspects of screening (in support of the public health agenda) and how many also provided information about the limitations of screening, and were presented in a clear, accessible way (in support of informed choice).

## **Results**

We found over 300 information sheets and leaflets about newborn bloodspot screening. One hundred and six leaflets described the heel prick and were included in our analysis: 68 from the UK (64 per cent); 34 from the USA (33 per cent); and four from Australia (4 per cent).

We found leaflets usually described the benefits of screening, and sometimes described the limitations of screening and choices open to parents. They varied in terms of their explicit reference to evidence and their ease of reading. We describe this in more detail below.

#### *Benefits of screening*

Most leaflets favoured screening. The vast majority of them (88/106) explained the primary aim of screening: to identify babies who are at higher risk of some rare but serious conditions. Some presented specific information about the rarity of these conditions in a particular population (24/106). Three-quarters of the leaflets described the conditions for which newborns were being screened (79/106). Over two-thirds mentioned the treatment available for one or more of the conditions for babies identified through screening (73/106). Less than half mentioned the difficulties of preventing the conditions (43/106) thereby increasing the need for screening; only 39/106 explained that the conditions were inherited, and only 11/106 mentioned reproductive choice.

Generally, leaflets also portrayed a comprehensive screening service. The majority of leaflets described the screening process, outlining the procedures involved in screening: 93/106 how the blood sample is taken, and 72/106 how the sample is sent to the laboratory. Many of the leaflets also reassured parents that additional support was available for them (66/106); 48/106 referred to general practitioners, midwives or specialists, and 23/106 referred to support organisations within or external to the health service. Sixteen leaflets referred to counselling (including genetic counselling) and 16/106 referred to other services such as supplementary screening programmes. Over two-thirds of the leaflets (71/106) indicated that follow-up tests would be needed to confirm the results for babies thought to be affected by the conditions, almost all of which indicated in what circumstances these further tests would be needed (69/106).

*Difficulties/limitations of screening*

Leaflets were less forthcoming about technical difficulties with screening, inconsistencies in the communication of results to parents, costs or the controversial issue of retaining samples for use once screening was complete.

Few leaflets mentioned that the test would be uncomfortable for the baby (19/106) or how the pain might be eased (5/106), although 76 leaflets mentioned the possibility that a second blood sample might be required. Only 3/106 leaflets mentioned that the cards on which the blood spots are collected are stored, or provide any information about possible uses of these stored cards.

Fewer than half of the leaflets indicated when parents would receive results (45/106), and fewer than half of these mentioned both when parents might receive negative results ('all clear') and positive results (i.e. their baby may have a condition).

Only a few explained when parents would be told if there was a need for follow-up tests (9/71), when these tests might occur (12/71), or when the results might be available (1/71).

Few leaflets contained information about the limitations or possible harms of screening: 19/106 mentioned false-negative results (that some babies affected by the conditions would be 'missed' by screening); 22/106 mentioned false-positive results (that some healthy babies might be thought to be affected in error); and 41/106 leaflets mentioned other possible harms, such as worry caused to parents (although these were often expressed as exhortations not to worry).

Only 14/106 leaflets referred to both false-negative and false-positive results. Thirteen of these were produced in the United States: 10 for parents and 3 for professionals.

One was created for health professionals in the UK. Some parent information created by US state health departments was in connection with piloting supplementary screening programmes, where more information is provided on the risks and benefits of participating in the pilot. For example, the New England Screening Programme information sheet stipulates that:



If your baby has an abnormal screening result, it is possible that your baby actually does not have the disorder. To be sure, your baby's doctor may recommend further testing by a specialist who may take additional specimens.... If your baby has a normal screening result, there is still a chance that your baby has the disorder (i.e. for some reason, the disorder was not detected by the newborn screen).... This risk exists for all forms of screening (12).

This level of information on the risks of screening generally did not appear in leaflets from the UK. We found three exceptions. The first was the following health professional information found on the publicly available Contact a Family website:

For most tests, some people with a positive result will turn out not to have the disorder ('false positive') and some with a negative result will subsequently be shown to have the disorder ('false negative'). Those people who have a positive result on the screening test ('diagnostic test'), which will pick out much more accurately those who have the disorder (13).

Two other UK exceptions were parent leaflets provided in Scotland and Trent that addressed the limitations of screening, albeit only in relation to cystic fibrosis. The Scottish parent leaflet produced in 2002 states that:

Several factors other than cystic fibrosis can affect the test result and therefore not all babies with a high IRT [screen positive test result] will have cystic fibrosis.... Also, it should be noted that on rare occasions the IRT test will fail to identify a baby with cystic fibrosis (14).

The parent leaflet produced in 2001 by the Trent Region Neonatal Metabolic Screening Service states, "This disease [cystic fibrosis] affects roughly 1 baby in 3000 and the majority of cases will be picked up." Later in the leaflet, under the heading "The fine details" and in smaller print, it states, "Unfortunately, whilst it is the best that is available, this is not a very good test. Several factors other than cystic fibrosis influence the amount of immunoreactive trypsinogen in the blood [screen positive test result] and we have to have a second-stage to the screen"(15).

Fourteen leaflets (14/106) mentioned the costs of screening, and only 5/106 referred to the cost-effectiveness of screening. The majority of these were from the United States, where screening is often funded by health insurance companies.

*Explicit about choice*

Only a third of the leaflets indicated whether or not screening was mandatory (38/106). The other 68 mentioned nothing about choice at all. Sixteen leaflets (all from the United States) said that screening was mandatory, and 25 leaflets said that screening was a choice (three of these were referring to a choice over supplementary screening where the core screening is mandatory). Ten leaflets that said screening

was a choice also recommend screening to parents, whilst four stressed the negative implications of not screening.

*Clear communication of up-to-date evidence*

To facilitate realistic expectations or to inform choice, information provided should be clearly communicated to its target audience, at the right time and supported by up-to-date research evidence. The target audience for the information was clear in 98/106 of the leaflets. Sixty-one of the leaflets indicated when they would be relevant or when they should be given. Over half the leaflets indicated the date of development or review (56/106). Similarly, over half (56/106) were appraised as 'easy to read', although 36/106 included technical terms that were unexplained, and 14/106 needed expert knowledge to understand. Only 17/106 cited any sources of evidence for the information contained in them, referring to policy documents, research papers or medical specialists who had reviewed the content of the leaflet.

*Information about carriers and genetic testing*

Almost half of the leaflets (49/106) referred to carriers. About half of these (23/49) indicated that, as an outcome of screening, babies may be identified as carriers of cystic fibrosis (10), sickle cell disorders (9), PKU (2) or other disorders (3). Twelve leaflets mentioned that babies' DNA might be tested, and 19 mentioned the implications of carrier status for babies' health. Almost half of the leaflets mentioning carriers (24/49) indicated that parents might be carriers of sickle cell disorders (10), PKU (9), cystic fibrosis (8) or other disorders (4). The following

information relating to carrier status also appeared in some of the leaflets: the implications of being a carrier for family planning and reproductive choice (19); wider carrier testing for family members (10); uncertainty regarding diagnosis (8); the number of carriers in the population (8); the psychological implications (e.g. anxiety) (6); not all mutations are tested for (4); and the number of mutations tested for (3).

## **Discussion**

### *Summary of findings*

The majority of leaflets support the public health agenda by informing parents of the benefits of screening. These included: the aims of screening; information about the conditions screened; benefits of early treatment for babies found to have the conditions; how the screening test is carried out; and information about follow-up tests.

Many leaflets did not facilitate realistic expectations or support the informed choice agenda, either failing to mention limitations, or not mentioning choice, or being difficult to understand. Few leaflets mentioned alternatives to screening, how and when parents would receive results, or provided sources of evidence or described how the information was developed.

More leaflets from the US, compared with those from the UK, provided information on the limitations of screening, particularly the possibility of false results, although these were still a minority. Whilst there was a dearth of this type of information in UK leaflets, it appears that more recent leaflets are beginning to address this more openly. This is in the context of screening for cystic fibrosis where there is a greater risk of false results than for other conditions.

*Strengths and weaknesses of the study*

The study involved carrying out a wide search for information resources through the internet, and through a selection of health service and support organisations throughout the UK. Our searching strategy, however, was not exhaustive. In developing and using the tool, two researchers performed double data extraction independently and compared results. However, this is a very small-scale exercise, compared with the use of a large Expert Panel and network of testers in developing the original DISCERN instrument (10).

In developing our tool, we drew on both NSC criteria, which are topic specific and policy focussed, and DISCERN criteria, which are broader and more focussed on the patient/individual. The researchers were generous in applying the criteria. Some leaflets were judged to have met the criteria even if the information provided was extremely brief.

Some contradictions were evident in categorising information as either presenting the benefits or difficulties/harms of screening. For example, we have listed the possible need for repeat testing as a limitation of screening. This is because repeat testing can cause worry for parents, and highlights some of the uncertainties of screening. However, repeat testing could also be considered a benefit, in that it provides evidence of follow-up procedures to ensure that screening is done correctly and all affected babies detected.

Nevertheless, the appraisal tool was able to distinguish parent information in terms of the type and style of information provided, and to indicate its reliability in terms of how and when it had been produced.

*Relating to other surveys of patient information*

Other studies that have examined and evaluated patient information about screening have also concluded that the risks, limitations or difficulties were not openly discussed. An Australian survey evaluating 58 leaflets on screening for breast cancer found that the benefits of screening were reported only ever as relative risk reduction and never as absolute risk reduction, thereby exaggerating the benefit (16).

Another study of 27 websites compared information on the possible benefits and harms of screening for breast cancer provided by different groups (17). The authors concluded that information provided by professional advocacy groups and

government organisations were biased in favour of screening. In contrast, information on consumer websites was more balanced and comprehensive.

Other small qualitative studies have been carried out to evaluate existing patient information. One such study of information on gastroscopy procedures in seven different hospitals, found that the quality of information varied between units, most leaflets lacked vital information, or included information that was confusing or ambiguous, and that information about risks was included in only one leaflet (18).

An audit of informed consent procedures prior to surgical procedures found that the poorest area of information-giving was related to the potential complications of various procedures (19).

The Royal College of Anaesthetists (RCA) has also evaluated patient information materials with the specific aim of developing new information resources (20;21). Like us, they carried out an in-depth review of patient information with a view to developing new, better-quality patient information. In the process of evaluating the leaflets, the RCA also developed an appraisal tool based on the DISCERN tool for evaluating patient information, as well as other tools for evaluating health information (20).

Although we found that many of the leaflets in our survey did not provide detailed information, particularly about the limitations and possible harms of screening, this is in line with the findings of a study in which parents' views were sought about the

information they required – parents generally viewed screening as routine, and only wanted basic information (22).

It should be noted that the research evidence is in favour of screening either to improve health outcomes through early treatment, or to improve parents' experiences of the diagnostic period (3;7;23-25), and that informed choice for screening is a relatively new phenomenon. It is not surprising, therefore, that the leaflets are 'biased' towards the benefits of screening, and many do not support informed choice. With or without informed choice, in an increasingly legalistic climate there may be increased pressure to provide patients with detailed information to facilitate realistic expectations. The question also arises whether in the current climate of informed choice and with increasing patient information available some parents may begin to demand more information about screening (19).

#### *Conclusions and recommendations*

We found combining the DISCERN criteria for patient information about treatment choices with NSC criteria for implementing screening programmes provided a practical route for appraising parent information and identifying examples of clear and informative text. We recommend the use of criteria for high quality patient information alongside topic or condition-specific guidelines as a starting point for developing patient information.



This survey of leaflets and information sheets illustrates that most health information about newborn blood spot screening portrays it in a positive light, and relatively few leaflets address difficulties openly. We therefore conclude that this information generally supports the public health agenda of achieving high screening uptake, but that exemplar parent information resources facilitate realistic expectations or informed choice in newborn screening.

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**Box 1: Issues addressed when appraising leaflets**

***Purpose of Leaflet***

Is it clear whom it is for?

If so, how is this indicated in the leaflet?

Is it clear when the information would be given?

***Reasons for Screening***

Aims/reasons for screening

Description of the conditions screened

Prevention programmes/difficulties of prevention

***Process and Consequences of Screening***

Description of the heel-prick test

Indication of when parents will receive results

Information on whether screening is not compulsory or mandatory

The limitations of screening (e.g. false-negative, false-positive results)

Costs of screening

Cost-effectiveness of screening

***Follow-up to Screening***

Information on need for further testing for diagnosis

Treatment for the conditions screened for

Any related services

***Information about Carriers***

Babies may be identified as carriers

Babies' DNA may be tested

Mutations tested for

Implications of carrier status for babies' health

Parents may be identified as carriers

Wider implications for families

***Production of the Leaflet***

Sources of evidence

Specific information linked to evidence

Level of complexity of the leaflet

Indication of how the leaflet was developed