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# **What is known about communication with parents about newborn bloodspot screening?**

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**UK Newborn Screening Programme Centre**

**'Quality through partnership in newborn bloodspot screening.'**

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A partnership between Great Ormond Street Hospital for Children NHS Trust, The Institute of Child Health, and The Institute of Education. Funded by the Department of Health on behalf of the UK.

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A glossary of terms used in this report is available from the UK Newborn Screening Programme Centre website at <http://www.newbornscreening-bloodspot.org.uk/>

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## **1.0 Abstract**

### **1.1 Background**

Two major trends are influencing the development of newborn bloodspot screening. First, technological advances are allowing bloodspot testing to identify a wider range of conditions. Second, raised ethical expectations are demanding that screening programmes adopt policies to support informed choice, as outlined in the second report from the National Screening Committee.

Systematic reviews of research literature have been portrayed as accessible sources for informing policy development. Therefore, a systematic review of recent reviews about communication for newborn screening was seen as an essential first step towards new evidence-informed policies.

### **1.2 Methods**

Readily accessible systematic reviews (published on The Cochrane Library and by the NHS Health Technology Assessment programme) were searched for evidence addressing communication for newborn screening. Reviews were included if they considered either newborn bloodspot screening or communication for screening. The methods for each review were examined and judged as likely to:

- comprehensively capture studies relevant to bloodspot screening communication;
- capture focused selections of relevant literature; or
- capture limited or no relevant studies, possibly unsystematically.

Each review was read and all references to parent or patient communication, information or choice potentially applicable to newborn screening were extracted and summarised. Each piece of data referring to evidence of effectiveness of communication or studies of parents' and health professionals' experiences and views was related to:

- the stages in the screening pathway (pre-screening information, antenatal screening information, parental choice / consent to test, parental choice / consent to test the baby's DNA, parental choice / consent to receive results, heel prick / screening test, subsequent tests, information with results, information with carrier results, and follow-up / post-test information); and
- characteristics of communication interventions (the provider of information; the timing of information; the written format of information; the non-written format of information; and the setting in which communication takes place).

Conclusions about interventions for implementation were drawn from reviews that comprehensively sought and appraised evidence of effectiveness of communication for screening. Other issues to be considered in developing policies were drawn from evidence of parents' and health professionals' experiences and views about newborn screening. Inferences were also drawn from evidence from other screening programmes.

The need for primary research was identified from authors' conclusions of comprehensive reviews and from gaps in the evidence that fell within or between the scope(s) of comprehensive reviews.

The need for secondary research (systematic reviews) was identified from comparing incomplete coverage of communication by systematic reviews with systematic reviews currently in preparation.

### **1.3 Results**

There is limited research reported about parents' and professionals' views and experiences of: pre-screening information (and none of antenatal information); consent for screening; the heel-prick itself and subsequent tests; the information provided with screening results; and in particular communication about carrier testing. This evidence is largely relevant to screening for cystic

fibrosis (CF), and little refers to screening for phenylketonuria (PKU), congenital hypothyroidism (CHT) or sickle cell disorders (SCD).

A single trial found that, despite counselling, receiving a false-positive screen result for cystic fibrosis can be difficult to understand and lead to anxiety, confusion and depression. Even after a normal sweat test some parents still worry about the health of their child, and this concern may be greater following DNA testing. Few parents appeared to change their reproductive plans. However, numbers of false-positives in this trial were very small, and more research is needed.

Little or no research addressed the effectiveness of pre-screening information or informed choice, communication of test results or follow-up screening or diagnostic tests. Limited research is reviewed about the effectiveness of communication about the heel-prick itself, before or at the time of the screening test.

At present, parents are offered little information and less choice. Anxiety may result from waiting for test results, poor communication of test results, false-positive results or carrier results. Refusal rates are negligible.

Research from other programmes confirms the need for education about the role and limitations of screening and the meaning of test results. Research specifically about uptake has little relevance while newborn screening is fully integrated with routine maternity care and refusal rates are negligible. Research about decision aids and informed consent may become more relevant with the offer of more screening programmes and the need to seek informed consent, whether this is for screening itself, the reporting of results, or the storage of bloodspots for clinical reasons or research.

#### **1.4 Implications for policy and practice**

There is a general lack of both procedures, and research to inform the development of such procedures, for:

- providing parents with information about the newborn bloodspot screening
- inviting informed consent for newborn bloodspot screening
- routinely informing parents of the results (positive or negative)
- explaining to parents the need for further tests
- addressing the potential for misunderstanding by parents of the test results; and
- understanding and addressing the particular difficulties raised by revealing carrier status.

#### **1.5 Recommendations for future research**

There is a need for primary research about parents' and professionals' experiences and views of screening and about the details of communication practice: who should provide any information, what, how, when, or where. Specifically, more research is needed about: consent to screen using DNA; consent to receive results; and parental response to false-negative results, false-positive results and carrier results.

There is a need to survey newborn screening services, in the UK and elsewhere, for their resources and policies and to compare these with the challenges to communication noted in this review in order to identify good practice.

## 2.0 Background

Newborn bloodspot screening was introduced on a national level in 1969 following the development of the Guthrie card, which allowed for the collection of full blood samples on filter paper. This technique for collecting blood samples enabled the introduction of national screening on bloodspots for phenylketonuria (PKU), replacing earlier urine tests. Since 1969, newborn screening for congenital hypothyroidism, cystic fibrosis and haemoglobinopathies have been introduced using the same Guthrie blood sample. More recently, innovations in DNA technologies and tandem mass spectrometry (which substantially increases the number of metabolic disorders that can be detected from dried bloodspot specimens) have introduced the capability to screen for a large number of additional metabolic disorders.

In the UK, the National Screening Committee (NSC) has adapted the 1968 WHO criteria (Wilson and Junger, 1968) to inform its screening policies. These revised criteria specify that screening should only be available under the following conditions:(National Screening Committee, 1998)

### Screening criteria:

1. clinically and bio-chemically well-defined disorder
2. known incidence in relevant populations
3. disorder associated with significant morbidity or mortality
4. effective treatment available
5. period before onset during which intervention improves outcome
6. ethical, safe, simple and robust screening test
7. cost-effectiveness of screening

Although the original phenylketonuria (PKU) screening programme conforms to the WHO criteria for a screening programme as formulated by Wilson and Junger (1968), other programmes have not fitted so closely (Seymour *et al.*, 1997). The advantages of screening for some conditions are not completely clear, and the detection of other conditions may result in unintended information, for example, the identification of non-affected carriers of recessively inherited conditions as a result of testing using DNA. This may cause concern about the baby's health and about non-paternity.

Current systems for screening are further challenged by raised expectations for informed consent for treatment, screening and diagnosis and research. (Medical Research Council, 2001);(The House of Commons, 2001);(Human Genetics Commission, 2002) The NSC has identified the need for "interventions that will make the nature of screening, with all its strengths and weaknesses, more immediately apparent to the person being screened", and calls for the introduction of two measures, to ensure that 1) "screening is offered and that the individual to whom it is offered is helped to make an informed choice", and 2) "screening is seen for what it is, a programme to reduce the risk of diseases and not a guarantee of diagnosis and cure."(National Screening Committee, 2002)

The second report of the National Screening Committee describes this shift of emphasis clearly to include promoting informed choice:(National Screening Committee, 2002)

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\* Some areas of the UK had already been screening for phenylketonuria since 1964.

## **2.1 Promoting informed choice**

The NSC's first report defined screening as:

*The systematic application of a test or inquiry, to identify individuals at sufficient risk of a specific disorder to warrant further investigation or direct preventative action, amongst persons who have not sought medical attention on account of symptoms of that disorder. (National Screening Committee, 1998)*

A new definition is proposed to take account of the importance of informed choice and risk reduction and this is set out below.

*A public health service in which members of a defined population, who do not necessarily perceive they are at risk of, or affected by, a disease or its complications, are asked a question or offered a test to identify those individuals who are more likely to be helped than harmed by further tests or treatment to reduce the risk of disease or its complications. (National Screening Committee, 2002)*

## **2.2 Defining informed choice**

Informed choice has been defined in a number of ways by a range of different stakeholders. Some refer to 'choice', others to 'consent' or 'dissent'. The NSC has adopted the principle that:

*Screening programmes should offer choice to individuals and that each individual should appreciate the risks and benefits of the screening programme for them as an individual. (National Screening Committee, 2002)*

The Department of Health Guide to Consent for Examination or Treatment provides a definition for wider settings:

*For consent to be valid, it must be given voluntarily by an appropriately informed person who has the capacity to consent to the intervention in question. Acquiescence where the person does not know what the intervention entails is not 'consent'. (Department of Health, 2001)*

Marteau provides a researcher's perspective:

*An informed decision is one where all the available information about the health alternatives is weighed up and used to inform the final decision: the resulting choice should be consistent with the individual's values. (Marteau et al., 2001)*

The National Childbirth Trust's policy statement on the importance of evaluation in maternity care provides a consumer perspective on informed choice, emphasising that although individuals may be provided with research evidence to inform their decisions, they can make their own choices:

*While parents may be guided by research evidence in making decisions, individuals making decisions will be influenced by their own beliefs, wishes and priorities. (Oliver, 1995)*

Informed choice in newborn screening is further complicated by the fact that it is the parent and not the patient who is informed and offered a choice, acting as a proxy for their child.

We propose that the tension, between the aim of detecting disease and a desire to promote informed choice, can be addressed by working with parents and health professionals to seek ways of maximising the acceptability of screening processes.

This has increased awareness of the need to communicate clearly with parents about their child's

health, about available tests and their potential consequences, and about the choices they face. This is reflected in a growing research literature about communication for screening (see special issue of *Health Expectations* (2001)). In addition there is an acknowledgement that patients and parents need to be included in policy-level decisions about health-care provision. (Consumers in NHS Research, 2002)

The UK Newborn Screening Programme Centre aims to address these issues by working in partnership with health professionals, laboratory scientists, parents and their children towards a common goal of assuring quality in newborn screening. The perspectives of parents and babies relate to the Programme Centre's plans for developing:

- evidence-informed resources for parents
- procedures for obtaining informed consent
- training for health professionals
- standards for screening processes
- standards for clinical follow-up
- standards for disease registers; and
- ethical standards for informatics.

The perspectives of parents will be sought through the recruitment of parent members to the Programme Centre's working groups. As fully integrated and supported members of these groups their perspectives will be actively drawn upon and utilised to shape products of working groups.

All these working groups, as part of their remit, will contribute to developing a comprehensive strategy for communication with parents about newborn bloodspot screening. Communication includes the provision of information on the aims and processes of the screening programme, potential outcomes and the offer of informed choice. For those parents who choose to have their baby screened, test results and their implications must be communicated, and support provided.

Developing guidelines for communication and support is comparable to methodologies for clinical guideline development. The starting point for each is assessing the relevant evidence, drawing, wherever possible, from systematic reviews as these are likely to provide a short-cut to a comprehensive and appraised synthesis of the evidence.

Research evidence is required to inform the nature of communication at different stages throughout the screening pathway, from pre-screening information, through informed choice and the screening test, to results and subsequent follow-up. This includes evidence of parents' and health professionals' views on different communication strategies, and evidence of the effectiveness of different strategies. The most relevant evidence would relate directly to newborn screening, but evidence drawn from broader scope of screening and communication research could also be informative.

As well as research into the effectiveness of screening in decreasing mortality and morbidity, there is a need to consider parent and patient-centred outcomes in order to understand how to communicate information in ways that achieve their needs. The tendency to address health professionals' priorities rather than parents' needs is illustrated in the quick reporting of positive results to parents, and complete lack of reporting negative results in many screening programmes in the UK.

There was thus a need to systematically review the research evidence on newborn bloodspot screening and communication with parents. A number of relevant systematic reviews had already been carried out in the area of newborn bloodspot screening, as well as in relation to communication about screening more generally. Systematically reviewing this evidence was seen as a first step towards developing evidence-informed policy for communication about newborn screening.

### **3.0 Aims and Objectives**

#### **3.1 Aim**

The aim of this review was to find out what is known about communication with parents about newborn bloodspot screening. This included exploring parents' and health professionals' views, as well as the effectiveness of different methods of communication.

#### **3.2 Objectives**

The objectives were to:

1. Identify reviews addressing methods of communication along the screening pathway in terms of the following characteristics of communication:
  - a. communicator (eg midwife, paediatrician, GP, specialist counsellor)
  - b. timing of communication (eg before the baby is born, when the blood sample is taken, when the results are given)
  - c. format of communication (eg verbal, written, audiotape, video )
  - d. setting of communication (eg clinic, home, GP surgery)
2. Appraise the strength of evidence provided by these reviews
3. Synthesise the evidence provided by these reviews
4. Identify gaps in the evidence in both primary and secondary research
5. Prepare summaries to inform working groups of the Newborn Screening Programme Centre.

## 4.0 Methods

### 4.1 Search strategy

The **Cochrane Library** was searched using the following search strategy:

newborn AND screening  
OR  
neonatal AND screening  
OR  
screening AND communication

to identify reviews in the Cochrane Database of Systematic Reviews and the Database of Abstracts of Reviews of Effectiveness (DARE).

The list of reviews prepared by the Cochrane Consumers and Communication Review Group was also searched for relevant reviews.

The Health Technology Assessment (HTA) publications website was searched electronically using the search term 'screening'. Titles were then screened, applying the selection criteria below:

### 4.2 Selection criteria

Reviews were included if they considered either:

1. newborn bloodspot screening; or
2. communication about screening generally.

### 4.3 Quality appraisal (see appendix 1)

Reviews were appraised by asking:

1. Was the aim of the review specified?
2. Was the search strategy reported?
3. Were the methods for assessing the quality of included studies reported?
4. Was the search strategy likely to capture:

- a. Studies of effectiveness of communication?

Search strategies were judged on two criteria: whether they applied specific terms relevant to communication about screening; and whether the selection criteria allowed for the inclusion of trial evidence.

- b. Studies addressing parents' and professionals' views?

Search strategies were judged on two criteria: whether they applied specific search terms relevant to views such as 'acceptability' or 'psychological impact'; and whether the search and selection criteria allowed for the inclusion of non-trial and trial evidence.

5. Were the methods for assessing the quality of included studies appropriate for:

- a. Studies of effectiveness of communication?

Methods for assessment were judged to be appropriate if they applied a hierarchy of evidence of effectiveness, weighting studies according to their study type, with most weight being given to randomised controlled trials, and less weight to non-randomised trials etc.

- b. Studies addressing parents' and professionals' views?

Methods were judged to be appropriate if they included a formal assessment of the quality of studies which specifically excluded any hierarchy of evidence of effectiveness. (Oakley, 2000) The EPPI-Centre

criteria for assessing the quality of qualitative studies are an example of such methods.(Peersman *et al.*, 1997)

Reviews were then categorised into those that were likely to either:

- capture comprehensively studies relevant to newborn bloodspot screening
- capture only focused selections of the relevant literature; or
- capture, possibly unsystematically, only very limited, or no relevant studies

for both:

- evidence of effectiveness of communication; or
- studies of parents' and health professionals' experiences and views.

Reviews were deemed to capture relevant studies **comprehensively** if they had:

- appropriate search terms covering communication or screening *broadly*
- selection criteria which focused on communication interventions rather than only screening interventions; and
- selection criteria which allowed for the inclusion of appropriate study designs (trials for addressing the effectiveness of communication, and non-trials for addressing parents' and health professionals' views).

Reviews were deemed to capture **only focused selections** of the relevant literature if they had:

- appropriate search terms focusing on *one area* of communication or screening
- selection criteria which focused on communication interventions rather than screening interventions; and
- selection criteria which allowed for the inclusion of appropriate study designs (trials for addressing the effectiveness of communication, and non-trials for addressing parents' and health professionals' views).

Reviews were deemed to capture **only very limited, or no** relevant studies if they had:

- no search terms focusing on *any area* of communication
- selection criteria which focused on screening interventions rather than communication interventions; or
- selection criteria which did not allow for the inclusion of appropriate study designs (trials for addressing the effectiveness of communication, or non-trials for addressing parents' and health professionals' views),

OR

- had no search strategy or selection criteria specified.

#### **4.4 Data extraction**

Each review was then read and all references to parent or patient communication, information or choice applicable to newborn bloodspot screening extracted and summarised.

#### **4.5 Synthesis**

The evidence was synthesised using matrices to collate the research required to support evidence-based policy and practice for communication. The coverage of research addressing the experience and communication of screening was mapped with a matrix that considers both the stages of the parents' journey and different characteristics of the communication process (see appendices 2 and 3).

The following stages in the parent journey were considered:

1. pre-screening information in general (ie information provided to parents prior to neonatal screening)
2. screening information given antenatally (as a source of pre-neonatal screening information)
3. parental choice / consent to test
4. parental choice / consent to test the baby's DNA
5. parental choice / consent to receive results
6. heel prick / screening test
7. subsequent tests (following the initial heel-prick)
8. information with results
9. information with carrier results
10. follow-up / post-test information

Within each of these stages in the screening pathway, findings were grouped into themes arising from the data.

The following five characteristics of communication interventions were specified in advance:

1. the provider of information
2. the timing of information
3. the written format of information
4. the non-written format of information; and
5. the setting in which communication takes place.

Further themes were identified from within these categories, arising from the data.

The relevance and strength of the evidence that is readily available from reviews was illustrated in a further matrix that distinguished evidence from reviews likely to capture studies of newborn screening most relevant to questions about the **effectiveness of communication**, from evidence from reviews likely to capture studies of newborn screening most relevant to parents' and health professionals' **experiences and views of screening**, and from evidence from reviews that may be transferable from reviews **other than newborn screening** (see appendix 3).

In this way, research evidence from reviews with search strategies most likely to capture relevant studies for addressing our question were given the most weight in our review. Research evidence drawn from reviews with search strategies which were incomplete for addressing our question, were considered less relevant, and were given less weight in our conclusions. In addition, it was noted which reviews applied quality appraisal methods appropriate for the research addressing our question.

Conclusions about interventions for implementation were drawn from reviews that comprehensively sought and appraised evidence of effectiveness. Other issues to be considered in developing policies were drawn from evidence of parents' and health professionals' experiences and views about newborn screening. Inferences were also drawn from evidence from other screening programmes. Research evidence from studies of newborn bloodspot screening was considered more pertinent than evidence from other newborn screening programmes, and more pertinent than other child and adult screening programmes.

The need for primary research was identified from authors' conclusions of comprehensive reviews and from gaps in the evidence that fell within the scope(s) of comprehensive reviews.

The need for secondary research (systematic reviews) was identified from comparing incomplete coverage of communication by systematic reviews with systematic reviews currently in preparation.

Finally, summaries were prepared to inform working groups of the Newborn Screening Programme Centre (see appendix 4).

## 5.0 Results

### 5.1 Included reviews

Thirteen systematic reviews were identified from the Cochrane Library, the Database of Abstracts of Reviews of Effectiveness (DARE), and the Health Technology Assessment (HTA) Programme publications website.\*

#### *Inclusion 1*

Of the 13 reviews identified, eight were reviews of newborn screening: for haemoglobinopathies (3), cystic fibrosis (3), and inborn errors of metabolism (2) [which includes screening for phenylketonuria or congenital hypothyroidism].

#### *Inclusion 2*

A further five reviews were identified which addressed communication in relation to screening programmes in general.

### 5.2 Quality appraisal of included reviews (see appendix 1)

Appraisal of the reviews' search strategies and criteria for inclusion distinguished reviews which captured relevant studies comprehensively from reviews that captured focused selection of the relevant literature and from reviews that did not capture relevant studies systematically (see tables 1 and 2).

**Table 1 - Studies of parents' and health professionals' views**

<i>reviews captured relevant studies comprehensively</i>	<i>reviews captured focused selections of the relevant literature</i>	<i>reviews captured only very limited, or no relevant studies</i>
Pollitt <i>et al.</i> 1997 (neonatal screening for inborn errors of metabolism)	Petticrew <i>et al.</i> 2000 (false-negative results)	Bastian <i>et al.</i> 2002 (people's experiences of screening)
		Davies <i>et al.</i> 2002 (sickle cell and thalassaemia)
		Edwards <i>et al.</i> 2002 (risk communication in screening)
		Jepson <i>et al.</i> 2000 (uptake of screening)
		Lees <i>et al.</i> 2002 (neonatal screening for sickle cell)
		Merelle <i>et al.</i> 2002 (newborn screening for cystic fibrosis)
		Murray <i>et al.</i> 1999 (screening for cystic fibrosis)
		O'Connor <i>et al.</i> 2002 (decision-aids)
		Serra-Prat <i>et al.</i> 2000 (neonatal screening for cystic fibrosis)
		Seymour <i>et al.</i> 1997 (newborn screening for inborn errors of metabolism)
		Zeuner <i>et al.</i> 1999 (antenatal and neonatal haemoglobinopathy screening)

\* The thirteen included systematic reviews are referenced in section 7.1.

**Table 2 - Evidence of effectiveness of communication**

<i>reviews captured relevant studies comprehensively</i>	<i>reviews captured focused selections of the relevant literature</i>	<i>reviews captured only very limited, or no relevant studies</i>
	Jepson <i>et al.</i> 2002 <sup>+</sup> (uptake of screening)	Bastian <i>et al.</i> 2002 (people's experiences of screening)
	O'Connor <i>et al.</i> 2002 <sup>+</sup> (decision aids)	Davies <i>et al.</i> 2000 (sickle cell and thalassaemia)
	Petticrew <i>et al.</i> 2000 <sup>+</sup> (false-negative results)	Edwards <i>et al.</i> 2002 (risk communication in screening)
		Lees <i>et al.</i> 2002 (neonatal screening for sickle cell)
		Merelle <i>et al.</i> 2002 (newborn screening for cystic fibrosis)
		Murray <i>et al.</i> 1999 (screening for cystic fibrosis)
		Pollitt <i>et al.</i> 1997 (neonatal screening for inborn errors of metabolism)
		Serra-Prat <i>et al.</i> 2000 (neonatal screening for cystic fibrosis)
		Seymour <i>et al.</i> 1997 (newborn screening for inborn errors of metabolism)
		Zeuner <i>et al.</i> 1999 (antenatal and neonatal haemoglobinopathy screening)

From these tables we can see that there is no comprehensive source of literature addressing communication with parents about newborn bloodspot screening. Literature about parents' and health professionals' experiences and views was addressed comprehensively in a review about neonatal screening for inborn errors of metabolism (ie pertinent to the current UK screening programmes for phenylketonuria (PKU), congenital hypothyroidism (CHT) and cystic fibrosis (CF)) (Pollitt *et al.* 1997).

Most reports could not be relied upon as comprehensive sources of evidence about communication for screening, although some were protocols and would be expected to lead to reliable systematic reviews in due course. However some focused aspects of communication, have been systematically reviewed, namely the effectiveness of communication for screening in the areas of uptake, decision-aids and false-negative results (although little of the primary research in these reviews related to newborn screening).

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<sup>+</sup> Only these reviews captured relevant studies and applied appropriate quality appraisal to these studies.

### 5.3 Research data from reviews

The research evidence identified from the systematic reviews is described in appendix 2\* (and summarised further in a matrix in appendix 3) under the following 10 headings matching stages in the parent journey.

1. pre-screening information in general (ie information provided to parents prior to neonatal screening)
2. antenatal screening information (as a source of pre-neonatal screening information)
3. parental choice / consent to test
4. parental choice / consent to test the baby's DNA
5. parental choice / consent to receive results
6. heel prick / screening test
7. subsequent tests (following the initial heel-prick)
8. information with results
9. information with carrier results
10. Follow-up / post-test information

As well as general information, for each heading, research evidence relating to the following five characteristics is specified: evidence which relates to the provider of information; the timing of information; about written information specifically; about non-written information specifically; and the place at which the exchange of information takes place.

The research evidence identified was grouped into:

- evidence of parents' and professionals' experiences and views
- evidence of effectiveness of communication; and
- evidence that may be applicable from other screening programmes.

The research evidence which came from systematic reviews with search strategies very unlikely to identify relevant literature comprehensively was separated from research evidence which came from systematic reviews with search strategies likely to identify relevant literature comprehensively. The results are summarised in appendix 3.

Research evidence from newborn bloodspot screening was considered before information from other newborn screening programmes, which in turn was considered before wider screening programmes.

### 5.4 Evidence of parents' and professionals' experiences and views

There is some evidence of parents' and health professionals' experiences and views of newborn screening drawn from the one review which had a search strategy likely to cover the relevant literature comprehensively (see Table 1).

- ➔ There is a general consensus for the need for public knowledge about the aims of screening and antenatal and neonatal screening provide an opportunity for this (Pollitt *et al.* 1997 and supported by Petticrew *et al.* 2000 and Davies *et al.* 2000). It is suggested that better understanding by parents of the limitations of screening prior to newborn bloodspot screening would reduce false expectations and confusion over results (Pollitt *et al.* 1997). The need for

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\* Within this report the systematic reviews are referenced, but not the primary studies reported within them. The full references of the primary studies reported within the systematic reviews are included in Appendix 2.

the sensitive provision of accurate information prior to screening is emphasised (Pollitt *et al.* 1997).

- ➔ Research about offering choice in newborn screening suggests that informed consent is not currently always obtained (Pollitt *et al.* 1997). One study of consent for newborn bloodspot screening in America implies that mothers do not want to be given a choice (Pollitt *et al.* 1997). There is an emphasis that detailed information is needed for parents if they are expected to make an informed choice (Pollitt *et al.* 1997)
- ➔ Although research suggesting that the initial heel-prick causes parents little concern is cited (Pollitt *et al.* 1997), there is also evidence to suggest that parents receive very little information and that it does affect their experience of screening. Research reviewed of communication about further tests following the initial heel-prick suggests that parents often don't receive accurate information about the need for further blood tests (Pollitt *et al.* 1997). Those parents who were given accurate information about the need for a second sample in newborn cystic fibrosis screening, were more satisfied as a whole with the screening process and less likely to need further information about the repeat test (Pollitt *et al.* 1997).
- ➔ Research reviewing the psychological impact of diagnosis following screening compared to traditional symptom-based diagnosis suggests that parents experience scepticism from health professionals over a symptom-based diagnosis which increases parental anxiety (Pollitt *et al.* 1997). Although some research found no differences in anxiety or depression levels amongst parents whose children were diagnosed following newborn screening and those who had a symptom-based diagnosis, parents' experiences of diagnostic delay and misdiagnosis have led to widespread support of newborn screening for cystic fibrosis (CF) and Duchenne (Pollitt *et al.* 1997).
- ➔ Research reviewed of parental response to false-positive results following newborn screening suggest that the psychological effects of false-positive screening results may be linked to the provision of information about the testing process (Pollitt *et al.* 1997). The evidence suggests that parents who were well informed about the screening process are not as anxious as less informed parents, when they discover that their positive screening result is false. Longer periods of waiting before obtaining the results of repeat tests are also associated with increased anxiety (Pollitt *et al.* 1997). Other research suggests that the time waiting for newborn screening results may contribute to parental anxiety (Pollitt *et al.* 1997, and Bastian *et al.* 2002). One newborn study showed that parents who were informed about a negative sweat test for cystic fibrosis on the phone were more likely to misunderstand the implications than those told face-to-face (Pollitt *et al.* 1997). Another study reviewed by Pollitt *et al.* (1997) suggested that when a second sample needs to be taken for newborn cystic fibrosis screening, a letter should be sent out prior to the visit by midwife, alerting parents to the need for a second sample.

Other research from within the reviews addresses parents' and professionals' experiences and views of screening, but because the search strategies of the systematic reviews do not cover this area comprehensively it is possible that they do not report the full picture.

- ➔ Consent for haemoglobinopathy screening is discussed briefly within the research. Zeuner *et al.* report negligible refusal rates for newborn metabolic screening. They note that there is also limited research of newborn screening on parents' reasons for acceptance or refusal of screening and call for more research into how much information is required for choice to be considered informed for antenatal and newborn screening (Zeuner *et al.* 1999). It is noted that obtaining consent around delivery is difficult and unsatisfactory (Zeuner *et al.* 1999).

- ➔ Zeuner *et al.*'s review (1999) of antenatal and newborn haemoglobinopathy screening suggests that uptake of genetic tests is associated with the manner in which the test is offered, pre-screening information and the availability of treatment (Zeuner *et al.* 1999).
- ➔ Published guidelines and current trends in newborn bloodspot screening support the active notification of all test results, although currently many areas do not notify parents of negative results (Zeuner *et al.* 1999). Research into parents' response to false-positive results suggests that some parents who received a positive screening result for cystic fibrosis remain anxious following a negative sweat test (Merelle *et al.* 2002 and Murray *et al.* 1999). The limited research reviewed of parental response to true-positive results is inconclusive (Murray *et al.* 1999).
- ➔ Research into the way in which results are reported reviewed in Davies *et al.* (2000) queries whether notification of haemoglobinopathy traits by post (following newborn bloodspot screening) increases parental anxiety. The evidence reviewed suggests the potential for excessive anxiety (Davies *et al.* 2000). Bastian *et al.* (2002) support this idea, commenting that research of parental understanding of carrier results suggests that being told your baby is a carrier following newborn screening may have adverse effects.
- ➔ The identification of carrier results for cystic fibrosis (CF) following DNA screening arises as a potential challenge for communication. Firstly, although there is no discussion within the reviews of consent to screen using DNA, Murray *et al.* note that carrier identification in newborn screening breaks confidentiality. Research of parents' understanding of carrier results suggests that 3 months after testing 17% of parents believed that they were at no risk of having a child with CF despite written and verbal information about the meaning of their baby's carrier result (Petticrew *et al.* 2000).
- ➔ The rates of attendance for cystic fibrosis (CF) counselling following newborn screening carrier results are reported as being over 50% (Murray *et al.* 1999).

### **5.5 Gaps in the evidence of parents' and professionals' experiences and views**

There is no reported research addressing views and experiences of receiving antenatal screening information and its influence on newborn screening information. There is limited research reported about: pre-screening information; consent for screening; the heel-prick itself and subsequent tests; the information provided with screening results; and in particular communication about carrier testing. With the exception of one review addressing inborn errors of metabolism (including cystic fibrosis (CF), congenital hypothyroidism (CHT) and phenylketonuria (PKU), the research evidence presented is likely to be incomplete.

### **5.6 Evidence of effectiveness of communication**

There is some research from reviews likely to capture evidence of the effectiveness of communication specifically addressing parents' understanding of false-positive results. However, this research is taken from reviews with search strategies that were likely to identify only focused selections of the relevant literature, or very limited studies (see Table 2). There were no reviews presenting evidence of effectiveness of communication comprehensively, so the research presented below is likely to represent an incomplete picture of the research evidence in this area.

- ➔ Research of parental awareness, understanding and knowledge from the Wisconsin Trial (Merelle *et al.* 2002) on newborn cystic fibrosis screening shows that despite intensive counselling of parents whose children had a false-positive IRT result, five percent of parents in the Trial still believed their children might have cystic fibrosis when questioned a year later. Parents who received their child's false-positive screening result in the newborn period had

better understanding and knowledge than parents who received their child's false-positive screening result at the un-blinding of the trial at four years of age.

- ➔ Research from the same study (Merelle *et al.* 2002) of parental emotional response to false-positive screening results, showed that false-positive results caused greater anxiety, depression and shock amongst those parents who received their baby's results following newborn screening, than amongst those who received the results at four years of age. Research reviewed in Murray *et al.* showed that two weeks after receiving a normal sweat test, 36% of parents had concerns about their child's health. It is also suggested that this reaction may be greater when DNA testing is involved (Murray *et al.* 1999), and that receiving false-positive newborn screening results may adversely affect parents' relationships with their baby (Merelle *et al.* 2002).
- ➔ There is limited research reviewed of the implications of false-positive results on future reproductive decision-making following newborn screening. The Wisconsin Trial found that 69% of parents who had received a false-positive screening result said they had not changed their reproductive plans, 8% had, and 22% were uncertain (Murray *et al.* 1999).

### **5.7 Gaps in the evidence of effectiveness of communication**

Very limited research is reported, none of which is drawn from comprehensive systematic reviews of the evidence of effectiveness of communication about newborn bloodspot screening. Of the research which is reported, several gaps can be identified. None of the research reported addressed the effectiveness of pre-screening information in general or, more specifically, the impact of antenatal information on communication about newborn screening. Research on the effectiveness of communication about parental consent or informed choice is not reported. Limited research is reviewed about the effectiveness of communication about the heel prick itself, before or at the time of the screening test. There is no research reported about communication about follow-up screening or diagnostic tests and only limited research about the effectiveness of communicating different results in different ways. There is no research reported addressing the effectiveness of communication about carrier results in particular.

### **5.8 Evidence that may be applicable from other research programmes**

Research from other screening programmes other than newborn bloodspot screening may be applicable to newborn bloodspot screening and is reported below. (No distinction has been made as to how comprehensively the available literature has been reviewed.)

- ➔ The limited research reviewed into communication and choice suggests that offering screening can itself suggest the person is at risk and have a negative impact on well-being (Bastian *et al.* 2002). It is reported that individuals' values affect their choices (Edwards *et al.* 2002 and O'Connor *et al.* 2002). Research on HIV screening suggests that the uptake of screening depends on the person offering the test (Zeuner *et al.* 1999). The need for education and non-directive counselling is acknowledged in antenatal screening research (Zeuner *et al.* 1999). Evidence as to the nature of this is not discussed.
- ➔ Research reviewed from other screening programmes (Jepson *et al.* 2000) about the uptake of screening suggests that educational home-visits and opportunistic screening may be effective in increasing uptake. Audio-visual educational materials, educational sessions, risk-factor questionnaires, face-to-face counselling and the use of incentives have been shown to be ineffective in related programmes. Attending for a previous screen and recommendations from GPs were both shown to increase uptake of mammography. Reminder interventions for health professionals were found to be effective. Office and audit systems for health professionals may also have increased uptake.

- There is limited evidence of parents' poor understanding of the purpose of antenatal screening (Zeuner *et al.* 1999 and Pollitt *et al.* 1997). There is also some evidence that parents with false-negative results from antenatal screening experience difficulties accepting positive neonatal screening results (Petticrew *et al.* 2000)
- Some reluctance amongst ultrasonographers to offer fully-informed choice is noted (Petticrew *et al.* 2000). There is also a suggestion (Petticrew *et al.* 2000) that screening programmes linked to target payments, such as cervical screening, as well as health professionals views on the efficacy and cost-effectiveness of screening, impacts on the offer of informed choice.
- Murray *et al.* cite the most commonly elicited reasons for the acceptance and refusal of cystic fibrosis antenatal screening which suggest that pregnant women: accept screening for reassurance that their child does not have cystic fibrosis (CF) and to prepare if they do have CF; and refuse screening because they perceive themselves to be at low risk of a not very serious condition. Jepson *et al.*'s review of screening programmes in general found that there was not sufficient evidence to examine uptake rates in relation to informed choice.
- O'Connor *et al.* (2002) discuss how decision-aids play a role in encouraging people to make an informed choice. No evidence is presented from newborn screening. Two trials of the impact of decision-aids in antenatal screening suggest that decision-aids lead to greater parental satisfaction about their choice (O'Connor *et al.* 2002)
- There is no newborn screening research reviewed addressing the specific effects of different ways of presenting information about screening to parents. Research of other screening programmes suggests that how information is provided can influence decisions (Edwards *et al.* 2002 and Petticrew *et al.* 2000). Research reviewed of the content of written information sent to women undergoing cervical screening, suggests that a 'normal result' letter should explain that this means low risk rather than no risk of developing cervical cancer (Petticrew *et al.* 2000). Leaflets and videos were shown to improve general knowledge of cystic fibrosis and carrier testing (Murray *et al.* 1999). Different methods to help people cope with screening results more generally are noted (Bastian *et al.* 2002) Research suggests that receiving false-negative screening results may cause people to later ignore symptoms putting them at risk (Bastian *et al.* 2002).
- Research also suggests there is a need for training for health professionals in the 'language of risk' (Petticrew *et al.* 2000). One study of obstetricians found that health professionals felt inadequately resourced to provide counselling for screening programmes (Petticrew *et al.* 2000)
- It is acknowledged that people often leave screening programmes unsure of their result (Bastian *et al.* 2002) There is very little discussion within the reviews of parental understanding of genetic information in relation to newborn screening programmes although research on antenatal screening programmes suggests mothers' understanding and recall of their own carrier results is poor, and that non-disclosure of individual results in antenatal couple carrier testing may reduce anxiety and the need for counselling (Murray *et al.* 1999).
- As well as identifying carriers of haemoglobinopathy traits and cystic fibrosis in the newborn period, antenatal and population screening programmes identify adult carriers. The benefits of detecting carriers for haemoglobinopathies and for cystic fibrosis for the individual and/or the community are unclear (Zeuner *et al.* 1999, and Murray *et al.* 1999) The potential resource implications and psychological effects of detecting carriers are noted by Zeuner *et al.* (1999) The potential for stigmatisation is also highlighted (in Murray *et al.*) There is limited research reviewed about antenatal and general population cystic fibrosis carrier testing which suggests initial individual carrier results cause anxiety, but that this returns to normal relatively quickly (Murray *et al.* 1999).

- ➔ UK research reviewed suggests adult carriers of cystic fibrosis do not report feeling stigmatised in their personal relationships, although there are examples from the USA of discrimination by private health insurance companies (Murray *et al.* 1999). The impact of disclosing non-paternity following carrier testing is only discussed in the research reviewed in relation to the impact of non-paternity on the estimation of at-risk couples in antenatal haemoglobinopathy screening (Zeuner *et al.* 1999).
- ➔ Research on follow-up after antenatal screening reviewed by Davies *et al.* (2000) suggests that lack of knowledge, accessibility of services, and time since the initial test, influence attendance. Zeuner *et al.* and Davies *et al.* reviewed the labour-intensive antenatal and newborn screening follow-up programme implemented by Brent Sickle Cell Centre, and found that it had high rates of coverage and acceptance. The cost-effectiveness of such a labour-intensive programme is unknown.

## **6.0 Discussion**

### **6.1 *Main findings***

Although recent systematic reviews have collated the evidence about the effects of the heel-prick and subsequent bloodspot screening tests, we found little evidence about how to share this information with parents.

At present parents are offered little information and less choice for newborn bloodspot screening. Anxiety may result from waiting for test results, poor communication of test results, false-positive results or carrier results. Refusal rates are negligible.

There is limited research reported about parents' and professionals' views and experiences of: pre-screening information (and none of antenatal information); consent for screening; the heel-prick itself and subsequent tests; the information provided with screening results; and in particular communication about carrier testing.

A single trial found that, despite counselling, receiving a false-positive screen result for cystic fibrosis can be difficult to understand and lead to anxiety, confusion and depression. Even after a normal sweat test some parents still worry about the health of their child, and this concern may be greater following DNA testing. Few parents appeared to change their reproductive plans. However, numbers of false-positives in this trial were very small, and more research is needed.

Little or no research addressed the effectiveness of pre-screening information or informed choice, communication of test results or follow-up screening or diagnostic tests. Limited research is reviewed about the effectiveness of communication about the heel-prick itself, before or at the time of the screening test.

Research from other programmes confirms the need for education about the role and limitations of screening and the meaning of test results. Research specifically about uptake has little relevance while newborn screening is fully integrated with routine maternity care and refusal rates are negligible. Research about decision aids and informed consent may become more relevant with the offer of more screening programmes and the need to seek informed consent, whether this is for screening itself, the reporting of results, or the storage of bloodspots for clinical reasons or research.

## **6.2 Strengths and weaknesses of the study**

This review of reviews has synthesised the knowledge base most readily available to screening programmes: systematic reviews prepared by reliable and readily accessible sources.

These sources have both strengths and weaknesses. The majority of published systematic reviews address questions of effectiveness, and these sources provide vigorously conducted reviews. More published Cochrane and Health Technology Assessment (HTA) reviews on newborn screening have focused on randomised control trial data addressing the effectiveness of screening in reducing mortality and morbidity, than on issues of communication and informed choice. Indeed social interventions and outcomes within the screening processes, such as the provision of information and the offer of choice have been neglected within both the primary and secondary research literature.

Limiting searches to the most readily available sources failed to capture some potentially relevant systematic reviews which need to be considered (Bekker *et al.*, 1999);(Broadstock *et al.*, 2000), one of which is not yet published (Green, 2000).

In addition we are aware of primary research which has not been picked up by the included systematic reviews and which might be relevant (Laird *et al.*, 1996). The relevance of literature from wider screening programmes is debatable, and feedback about the incorporation of wider research following early drafts of this review have been contradictory. Additional, potentially relevant research includes research on antenatal ultrasound (Garcia *et al.*, 2002) and newborn Duchenne screening.(Parsons *et al.*, 2002)

Tabulating the findings from the studies reviewed in a matrix that accommodated the screening pathway and the coverage of the reviews, allowed both a summary of the existing evidence and the identification of gaps in the evidence needed for the production of evidence-informed parent information for the UK Newborn Screening Programme Centre.

Much of the evidence in the reviews was from primary research not addressing questions of effectiveness of communication strategies. In view of the current lack of consensus about quality criteria for these study designs, the authors of the reviews did not formally assess the quality of these studies when including them. Thus conclusions of this report must rely on research that is largely unappraised.

This review has not sought to distinguish between primary studies of screening in the UK and elsewhere. This is due to the constraint of reviewing existing systematic reviews rather than primary studies. Reviews do not report this information in a consistent manner. This may limit the applicability of any findings to specific screening programmes.

## **6.3 Other relevant literature**

Our findings can be compared to those of related research in newborn and other screening programmes.

### *Carrier screening*

The lack of clarity found as to how to communicate effectively with parents about carrier status in newborn bloodspot screening is reflected in both cystic fibrosis and haemoglobinopathy literature.(Agency for Health Care Policy and Research, 1993);(Parsons *et al.*, 2003)

Research into the psychosocial implications of carrier identification in newborn cystic fibrosis screening found that some families experienced unnecessary anxiety because of the way they had been told there was a query on the newborn screening test (Parsons *et al.*, 2003). This research also highlighted the burden placed upon families of notifying their relatives about the result.

Communicating the difference between a carrier and affected result in haemoglobinopathy screening is highlighted as a priority.(Agency for Health Care Policy and Research, 1993) The AHCPR review reflects our finding that communication and parent choice for the heel-prick test has not commonly been incorporated into screening policies. Instead priorities for communication in haemoglobinopathy screening have focused on counselling for at risk couples following antenatal screening and education for parents of affected children to identify early signs of disease.

The NIH consensus statement on population genetic screening for cystic fibrosis (CF) cites evidence of high public support for such a programme, whilst also voicing the concern that “disclosure of genetic test results might affect one’s family relationships, employment, educational or other opportunities, or ability to maintain or obtain health insurance”. Whilst listing what information should be provided about the tests, no research is presented about how this information should be communicated.(National Institutes of Health, 1997)

### *Ultrasound in pregnancy*

Our finding that parents are provided with very little information is common to research about ultrasound screening. Garcia *et al.* report that women often lack information about the purposes for which an ultrasound scan is being done and the technical limitations of the screening test (Garcia *et al.*, 2002). Some women also remain worried about the scan even after, they are told that nothing bad has been found. As with newborn screening research into Down Screening suggests that better information on the limitations of screening programmes would reduce the adverse effects of false-negative results.

### *Pain relief during the heel prick*

Although not directly linked to communication about the newborn bloodspot screen, the baby’s experience of the heel-prick procedure is likely to influence parents’ views. Evidence of effectiveness of ways of ameliorating the experiences of the screened newborn have been well reviewed by Pollitt *et al.* (1997). The effects of the heel-prick on the baby are discussed, and suggestions made of methods for reducing the pain and pacifying the baby.

## 6.4 Implications for policy and practice

There is a general lack of both procedures, and research to inform the development of such procedures, for:

- providing parents with information about the newborn bloodspot screening
- inviting informed consent for newborn bloodspot screening
- routinely informing parents of the results (positive or negative)
- explaining to parents the need for further tests
- addressing the potential for misunderstanding by parents of the test results; and
- understanding and addressing the particular difficulties raised by revealing carrier status.

This development should be informed by the findings of this review and the findings of the relevant primary research included in systematic reviews.

## 6.5 Implications for future research

There are no systematic reviews of the effectiveness of communication for newborn screening. Neither has the literature on parents' and health professionals' experiences and views been systematically collated for newborn sickle cell screening. However, gaps in systematic reviews are likely to be filled by reviews currently in progress addressing:

- 'Personalised risk communication in health screening programmes' which includes consideration of both antenatal and newborn screening (Edwards *et al.* 2002);
- 'Psychosocial aspects of genetic screening of pregnant women and newborns'; (Green, 2000) and
- 'Psychological impact and understanding of screening'. (Doust *et al.*, 2003)

Newborn bloodspot screening was introduced with phenylketonuria (PKU), and then congenital hypothyroidism (CHT), at a time when less attention was paid to communication with parents. This probably explains why there is almost no specific research contained in these reviews about PKU or CHT.

The literature about parents' and professionals' experiences and views of screening available within these systematic reviews, rarely addressed the details of communication practice: who should provide any information, what, how, when, or where. Specifically, there was no discussion of consent to screen using DNA, nor consent to receive results. Neither is there research of parental response to false-negative results following newborn screening; nor newborn screening carrier results, communication and anxiety or non-paternity. Except for responses to false-negative results, these other specific gaps in the evidence will be addressed in an on-going study of telling parents their child's carrier status following newborn screening. (Oliver *et al.*, 2002)

What literature there is about parents' and professionals' experiences and views of newborn screening has been well reviewed for phenylketonuria (PKU), congenital hypothyroidism (CHT) and cystic fibrosis (CF) (Pollitt *et al.* 1997). We recommend that the parallel literature for sickle cell disorders (SCD) be systematically reviewed to complete the current knowledge base about challenges to communication for newborn screening. Such a review should include an attempt to appraise the quality of included research.

Very little research was found relating to developing and evaluating communication strategies for newborn screening. There is a need to survey newborn screening services, in the UK and elsewhere, for their resources and policies and to compare these with the challenges to communication noted in this review in order to identify good practice.

Although assumptions about parents not wanting the offer of informed choice, and fears that they might make the 'wrong' decision, have contributed to the lack of research around informed consent for newborn screening, recent debates around informed choice in newborn screening may lead to changes in policy and practice. There is therefore a real need for high-quality primary research exploring parents attitudes to and understanding of the choices offered to them, as well as better understanding of the rates of consent and dissent. Furthermore, research into health professionals' attitudes to and understanding of the process is also needed.

We also noted that although the literature addressed false-negative results comprehensively, there was little literature exploring the impact of, or communication about, false-positive results and we recommend more research is done in this area.

When on-going reviews of communication about screening are published, they should be examined for primary studies that report: evaluations of any type of intervention aimed at communicating with parents about newborn bloodspot screening; the processes involved in delivering such interventions; or conclusions from other screening programmes that could be applicable to newborn screening.

Authors of reviews of decision-aids (O'Connor *et al.* 2002), screening uptake (Jepson *et al.* 2000), and false-negative results (Petticrew *et al.* 2000), recommended the following research:

- O'Connor *et al.* (2002) note that little is known about the practitioner's perspective on decision-aids, or the impact of decision-aids on communication between the practitioner and patient. They stress the need for better understanding of what type of decision support works with which types of people.
- Jepson *et al.* (2000) recommend that all future studies should measure informed uptake as well as actual uptake and might include a measure of the decision-making process. They specifically recommend that a systematic review of informed uptake be undertaken which includes studies which have measured informed uptake, and / or decision-making processes.
- Petticrew *et al.* (2000) underline the need for research into the long-term medical, psychological and other consequences of false-negative results, as well as on the most effective means of presenting information on residual risks to those individuals undergoing screening.

## **6.6 Implications for the Programme Centre**

The Programme Centre is convening a number of working groups to set standards for newborn bloodspot screening in the UK. Issues of informed consent for screening, and for the storage of residual bloodspots will be considered by these groups. Structured summaries arising from this review will be made available to those groups (see appendix 4).

The Programme Centre will draw on the findings of this review to develop information for parents and health professionals about newborn bloodspot screening (see appendix 4).

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