

Communication of carrier status information following universal newborn screening for sickle cell disorders and cystic fibrosis: qualitative study of experience and practice

J Kai*, F Ulph, T Cullinan and N Qureshi

Division of Primary Care, School of Graduate Medicine & Health, University of Nottingham, Nottingham, UK

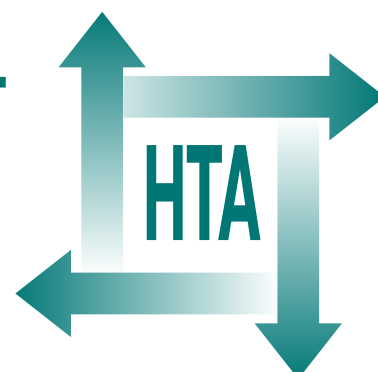
*Corresponding author



Executive summary

Health Technology Assessment 2009; Vol. 13: No. 57
DOI: 10.3310/hta13570

Health Technology Assessment
NIHR HTA programme
www.hta.ac.uk





Executive summary

Background

Universal newborn screening for sickle cell (SC) disorders and cystic fibrosis (CF) has recently been implemented across England as part of the NHS newborn blood spot (heel prick) programme. The aim is early identification and treatment of babies affected by these disorders, but screening can also identify infants who are healthy carriers of the conditions.

Differences between newborn screening for SC and CF are that identification of newborn SC carriers is relatively common while identification of newborn CF carriers is rare. Also, antenatal carrier screening for SC means parents may be more prepared for the possibility of their newborn being a carrier, and this clear result is available following the newborn blood spot alone. A two-stage screening process for CF means parents experience the newborn blood spot, an initial result suggesting increased risk of CF, with need for a second blood spot sample, before being later informed their child is a carrier. Apparent carriers of CF also have a small residual risk of being affected with CF.

Knowledge of a child's carrier status and its implications may be helpful as this can have reproductive implications for the child, parents, and their wider family. Parents of infants identified as carriers must be informed of their baby's result. However there is a lack of knowledge of current practice nationally, and lack of evidence internationally to inform the most effective ways of doing so, in particular from parents' experiences.

Objectives

The study aimed to describe and explore current practice, methods and experience of communicating carrier status information following newborn screening for CF and SC disorders, to inform practice and further research. The study sought to address the following questions:

- What is current practice for communicating carrier status information following newborn screening for SC disorders and CF in England?

- What are the views of health professionals communicating carrier status information on acceptability, feasibility, and effectiveness of methods for informing parents?
- What are parents' experiences and views of how they are informed and the support they are offered?
- How well is carrier status information understood by parents?
- What is the impact on a family of being informed of newborn carrier status?
- What can we learn from existing evidence and current practice and experience about effectiveness and feasibility of methods for communicating carrier status information, and what further research is required?

Methods

A qualitative study across England using (1) a preliminary phase of semi-structured telephone interviews with child health screening co-ordinators in all nine English health regions, and thematic analysis of data; (2) semi-structured face-to-face interviews with purposeful samples of 67 family members (49 mothers, 16 fathers, 2 grandparents) of 51 infants identified by universal newborn screening as carriers of CF ($n = 27$) and SC ($n = 24$), with experience of carrier status information communicated by a range of different methods in localities across England, with data analysis by constant comparison, and subsequent respondent validation; and (3) semi-structured telephone interviews, and focus groups, with a key informant sample of 16 differing health professionals currently tasked with communicating results to parents in a range of ways, with thematic analysis of data. In parallel, existing evidence was reviewed, focusing on methods of communicating newborn carrier information.

Results

Methods for and respondents' experiences of communication of carrier results varied considerably within and between regions, and within and between SC and CF contexts. Approaches ranged from letter or telephone

call alone, to in-person communication in the clinic or at home, with health professionals from haemoglobinopathy, CF, screening and genetics backgrounds, or from community and primary care, such as health visitors with SC carrier results. Health professionals identified pros and cons of different methods, preferring opportunity for face-to-face communication with parents where possible, particularly for CF carrier results. They were concerned by regional variations in protocols, the lack of availability of translated information on SC carrier results, and the feasibility of sustaining more 'specialist' involvement at current levels, particularly for SC carriers. They were positive about involvement of primary care based generalists if appropriately supported, but felt this may be less feasible for rarer and potentially more complex CF results.

Parents were often poorly prepared for the possibility of a newborn carrier result. Some had felt overloaded by screening information received during pregnancy or prior to newborn screening, or found this information failed to meet their needs. They sought timely and specific information at each successive stage of the screening and communication pathway.

Opportunity for face-to-face communication of results was valued by parents of SC carriers and appeared particularly necessary for those without prior knowledge of SC carrier status or where English was not their first language. Indirect communication of results by letter appeared effective and feasible for parents more aware of SC carrier status from antenatal or earlier experience, and where this communication contained an unambiguous opening statement emphasising 'your child is not ill'. Face-to-face communication of CF carrier results by professionals with screening, CF or genetics backgrounds worked well for parents, but communication and information was crucially lacking at the earlier stage of repeat blood spot testing, which involved midwives or health visitors who could be uncertain of the CF screening process, creating considerable distress among half of respondents.

Rather than learning of their newborn's carrier status in itself, untoward anxiety or distress among parents appeared influenced firstly by *how* information and communication was offered to them during the screening process, and secondly if they had less prior awareness of carrier status or the possibility of a carrier result. Parents could fear their child had a serious problem, particularly

while awaiting results or before seeing a professional, and be left in an information vacuum. Parental distress and anxiety appeared mostly transient, subsiding with understanding of carrier status and communication with a professional. Only a minority of parents appeared to have continued concerns about their child.

Respondents had no particular preference for the type of health professional who communicated results to them, as long as they were well informed and could answer their queries. Parents who had received written information about carrier results found this useful for reference and for discussion with their families. However, this information could be insufficiently detailed for some, and poorly accessible in content and language for others.

Parents regarded carrier results as valuable information gained fortuitously. They sought to share this with their extended families and to inform their children in the future. Respondents felt community awareness and information about SC and CF could be improved. Although there was some evidence of misconceptions about SC, most parents understood the benign implications of carrier status and that it may impact on future reproductive decisions. However, parents needed greater support after communication of results in considering and accessing cascade testing, and negotiating further communication within their families. Extended families' reception of carrier information ranged from being supportive to negative reactions or avoidance of the news.

Conclusions

Methods of communication of newborn carrier results vary considerably across England. Parents' needs for timely and appropriate information may not be met consistently or adequately. Respondents' experiences suggest a need for greater recognition of communication with individuals occurring across a screening pathway, rather than as a discrete event.

Implications for health care

Current practice could be enhanced by improving pre-screening information to include the prevalence of SC and CF carrier status, the common possibility of a newborn SC carrier result, and what to expect in relation to a repeat blood spot; recognition that the effectiveness and

acceptability of communication of results indirectly by letter or in person may vary according to individuals' prior awareness or language needs; and provision of translated forms of SC carrier result information. In communication of CF screening, clear specification of information for provision to parents at the time of repeat blood spot testing is needed, with explicit guidance for communication by professionals undertaking this test; and in-person communication of carrier results by a well-informed professional.

Growth in carrier identification following expansion of newborn screening programmes may increase demand on those with condition-specific or genetics expertise. According to local contexts, such as prevalence of SC, the potential for greater involvement of primary care based professionals within mixed models of communicating carrier information could be explored; and a locality-based screening practitioner role operating across programmes to provide support for parents, and liaison with other professionals, during screening and following screening results, could be further developed.

Recommendations for research

Further research is needed to: (a) design and evaluate specific information for parents approached for a repeat blood spot in CF screening; (b) explore the value of refining current pre-screening information to better prepare

parents for the possibility of carrier identification; (c) develop and evaluate the accessibility and acceptability of translated forms of standardised SC carrier result information; (d) prospectively study or audit practice with the further establishment of screening programmes; (e) investigate how health professionals use and present information across the screening pathway; (f) develop and evaluate support and training for health professionals involved in screening to be able to communicate relevant information; (g) examine the use of differing mixed service models according to local contexts; (h) investigate parents' attitudes towards, access to and experience of further carrier testing for themselves or their other children, and its impact on later reproductive decisions; (i) develop and evaluate methods to support cascade testing and communication of carrier information with children and families; (j) explore the uptake of information and counselling, community awareness and its influence on the screening experience; and (k) further experience of families over time to enable greater understanding of longer term benefits or harm of newborn carrier identification.

Publication

Kai J, Ulph F, Cullinan T, Qureshi N. Communication of carrier status information following universal newborn screening for sickle cell disorders and cystic fibrosis: qualitative study of experience and practice. *Health Technol Assess* 2009;**13**(57).



How to obtain copies of this and other HTA programme reports

An electronic version of this publication, in Adobe Acrobat format, is available for downloading free of charge for personal use from the HTA website (www.hta.ac.uk). A fully searchable CD-ROM is also available (see below).

Printed copies of HTA monographs cost £20 each (post and packing free in the UK) to both public **and** private sector purchasers from our Despatch Agents.

Non-UK purchasers will have to pay a small fee for post and packing. For European countries the cost is £2 per monograph and for the rest of the world £3 per monograph.

You can order HTA monographs from our Despatch Agents:

- fax (with **credit card** or **official purchase order**)
- post (with **credit card** or **official purchase order** or **cheque**)
- phone during office hours (**credit card** only).

Additionally the HTA website allows you **either** to pay securely by credit card **or** to print out your order and then post or fax it.

Contact details are as follows:

HTA Despatch
Magellan
Concept House, Bell Road
Basingstoke, Hants RG24 8FB, UK

Email: orders@hta.ac.uk
Tel: 02392 492 000
Fax: 02392 478 555
Fax from outside the UK: +44 2392 478 555

NHS libraries can subscribe free of charge. Public libraries can subscribe at a very reduced cost of £100 for each volume (normally comprising 30–40 titles). The commercial subscription rate is £300 per volume. Please see our website for details. Subscriptions can be purchased only for the current or forthcoming volume.

Payment methods

Paying by cheque

If you pay by cheque, the cheque must be in **pounds sterling**, made payable to *Direct Mail Works Ltd* and drawn on a bank with a UK address.

Paying by credit card

The following cards are accepted by phone, fax, post or via the website ordering pages: Delta, Eurocard, Mastercard, Solo, Switch and Visa. We advise against sending credit card details in a plain email.

Paying by official purchase order

You can post or fax these, but they must be from public bodies (i.e. NHS or universities) within the UK. We cannot at present accept purchase orders from commercial companies or from outside the UK.

How do I get a copy of HTA on CD?

Please use the form on the HTA website (www.hta.ac.uk/htacd.htm). Or contact Direct Mail Works (see contact details above) by email, post, fax or phone. *HTA on CD* is currently free of charge worldwide.

The website also provides information about the HTA programme and lists the membership of the various committees.

NIHR Health Technology Assessment programme

The Health Technology Assessment (HTA) programme, part of the National Institute for Health Research (NIHR), was set up in 1993. It produces high-quality research information on the effectiveness, costs and broader impact of health technologies for those who use, manage and provide care in the NHS. 'Health technologies' are broadly defined as all interventions used to promote health, prevent and treat disease, and improve rehabilitation and long-term care.

The research findings from the HTA programme directly influence decision-making bodies such as the National Institute for Health and Clinical Excellence (NICE) and the National Screening Committee (NSC). HTA findings also help to improve the quality of clinical practice in the NHS indirectly in that they form a key component of the 'National Knowledge Service'.

The HTA programme is needs led in that it fills gaps in the evidence needed by the NHS. There are three routes to the start of projects.

First is the commissioned route. Suggestions for research are actively sought from people working in the NHS, from the public and consumer groups and from professional bodies such as royal colleges and NHS trusts. These suggestions are carefully prioritised by panels of independent experts (including NHS service users). The HTA programme then commissions the research by competitive tender.

Second, the HTA programme provides grants for clinical trials for researchers who identify research questions. These are assessed for importance to patients and the NHS, and scientific rigour.

Third, through its Technology Assessment Report (TAR) call-off contract, the HTA programme commissions bespoke reports, principally for NICE, but also for other policy-makers. TARs bring together evidence on the value of specific technologies.

Some HTA research projects, including TARs, may take only months, others need several years. They can cost from as little as £40,000 to over £1 million, and may involve synthesising existing evidence, undertaking a trial, or other research collecting new data to answer a research problem.

The final reports from HTA projects are peer reviewed by a number of independent expert referees before publication in the widely read journal series *Health Technology Assessment*.

Criteria for inclusion in the HTA journal series

Reports are published in the HTA journal series if (1) they have resulted from work for the HTA programme, and (2) they are of a sufficiently high scientific quality as assessed by the referees and editors.

Reviews in *Health Technology Assessment* are termed 'systematic' when the account of the search, appraisal and synthesis methods (to minimise biases and random errors) would, in theory, permit the replication of the review by others.

The research reported in this issue of the journal was commissioned by the HTA programme as project number 04/10/01. The contractual start date was in May 2006. The draft report began editorial review in February 2009 and was accepted for publication in July 2009. As the funder, by devising a commissioning brief, the HTA programme specified the research question and study design. The authors have been wholly responsible for all data collection, analysis and interpretation, and for writing up their work. The HTA editors and publisher have tried to ensure the accuracy of the authors' report and would like to thank the referees for their constructive comments on the draft document. However, they do not accept liability for damages or losses arising from material published in this report.

The views expressed in this publication are those of the authors and not necessarily those of the HTA programme or the Department of Health.

Editor-in-Chief: Professor Tom Walley CBE
Series Editors: Dr Aileen Clarke, Professor Chris Hyde, Dr John Powell,
Dr Rob Riemsma and Professor Ken Stein

ISSN 1366-5278

© 2009 Queen's Printer and Controller of HMSO

This monograph may be freely reproduced for the purposes of private research and study and may be included in professional journals provided that suitable acknowledgement is made and the reproduction is not associated with any form of advertising.

Applications for commercial reproduction should be addressed to: NETSCC, Health Technology Assessment, Alpha House, University of Southampton Science Park, Southampton SO16 7NS, UK.

Published by Prepress Projects Ltd, Perth, Scotland (www.prepress-projects.co.uk), on behalf of NETSCC, HTA.

Printed on acid-free paper in the UK by Henry Ling Ltd, The Dorset Press, Dorchester.