Psychosocial aspects of genetic screening of pregnant women and newborns

Psychosocial aspects of genetic screening of pregnant women and newborns: a systematic review

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Executive summary

Health Technology Assessment 2004; Vol. 8: No. 33

Health Technology Assessment
NHS R&D HTA Programme

Executive summary

Background

More genetic screening takes place during pregnancy and the newborn period than at any other time. These are key points in the life course where people are accessible to the health services. However, these are also periods when parents are at their most vulnerable. With developments in technology, such tests are multiplying. It is therefore considered important to understand the psychosocial aspects of screening in order that screening programmes can be designed in ways which minimise harm. Our plan of investigation had two guiding principles:

- Screening programmes need to be considered according to how they are likely to be experienced by the recipients, rather than from the perspective of the service provider.
- The ultimate aim of the review is to learn lessons from psychosocial aspects of past screening programmes which can be used to inform genetic screening in the future. This does not preclude learning from the examples of non-genetic screening programmes, particularly where the evidence suggests that the genetic/non-genetic distinction is not highly salient from the recipients' point of view.

Objectives

The review aimed to address five broad questions concerned with knowledge, anxiety, other emotional aspects of screening, factors associated with participation/non-participation in screening programmes and the long-term sequelae of false-positive, false-negative, true-positive and true-negative results.

Three revisions were made. The literature on other emotional aspects of screening and on false-negatives was too fragmented for useful conclusions to be drawn, and discussion of true-positives is confined to newborn screening, for the same reason.

Methods

This review started from a substantial literature base that provided the basis for (a) scoping the literature, (b) informing search strategy terms and (c) identifying preliminary article inclusion and exclusion criteria. The main eligibility criteria were:

- Any screening programme aimed at pregnant women or newborn babies that included a 'genetic' target condition. 'Genetic' includes chromosomal anomalies.
- Any study that reported psychosocial data collected directly from parents.

There were no geographical or methodological limits except that studies asking only hypothetical questions and case reviews/single experiences were excluded.

Five electronic databases were searched, two journals were hand-searched and attempts were made to locate unpublished work. The data elicited from articles using the data extraction form developed for this study were entered into an SPSS database (version 10.1).

Results

A total of 288 candidate publications were identified, 106 of which were eligible: 78 concerned with antenatal screening and 28 with newborn screening. The main findings were as follows.

Knowledge

- Levels of knowledge adequate for decision-making are not being achieved.
- Information leaflets and videos have some effect but large gaps in knowledge usually remain.
- Procedural aspects of testing are better understood than material related to the meaning of risk calculations.
- Substantial social and cultural inequalities exist in knowledge about testing.
- The above findings almost certainly underestimate the extent of the problem, because only limited aspects of knowledge have been studied to date.

In addition

- Knowledge is not the same as understanding.
- Public understanding of the basic concepts associated with screening is poor.
- Knowledge that is only superficially acquired may not be retained.
- Informed consent for neonatal screening has been little studied.

Anxiety

- Studies that have succeeded in increasing knowledge have not observed a corresponding increase in anxiety.
- Anxiety is clearly raised in women receiving positive screening results but evidence is lacking of a beneficial (i.e. reassuring) effect of receiving a screen-negative result.
- Anxiety in screen-positive women falls on receipt of subsequent reassuring results but some residual anxiety may remain.
- The way in which carrier screening is offered may affect anxiety in screen-negative women.

In addition

- Knowledge that improves decision-making may not be the same as that which reduces anxiety.
- Some anxiety might be an appropriate response and might aid coping and decision-making.
- Young women may be more vulnerable to anxiety arising from positive screening test results.
- Knowledge and anxiety in men whose partners are undergoing screening have been little studied.

Attitudes and test uptake

- Most women hold positive attitudes towards prenatal screening.
- Women having screening tend to hold more negative attitudes to abnormality, to perceive their likelihood of having an affected child (or themselves being a carrier) as greater, to perceive the risks of subsequent procedures as lower, to perceive others as thinking they should have the test and are more likely to intend to have a termination if an abnormality is detected.
- Women who were more satisfied with their choices were also more falsely reassured, and made their choices less systematically, than women with lower satisfaction scores.
- A minority (perhaps up to 30%) of women receiving a screen-positive result in pregnancy expressed regret about their screening decision.
- Uptake of neonatal screening has been treated as a 'given' and not a research topic.

Policy implications and recommendations for future research

The results of the review have many implications for the work of the National Screening Committee. The most pressing of these, in order of priority, relate to:

- the inadequacy of current procedures for achieving informed consent
- the cost of providing a satisfactory service
- the unmet needs of 'false-positives'
- the unmet needs of women's partners, particularly in carrier screening.

We suggest that research is conducted on the above four topics in order to fill gaps in the evidence base that relate to screening technologies which have been available for many years. In addition, future screening programmes will create a new list of research questions, based on the same main agenda but applied to new areas, for example, to

- new conditions such as haemoglobinopathies and fragile X syndrome
- new client groups such as young women and minority ethnic groups
- new testing modalities such as ultrasound.

Research is needed which incorporates these topics into the mainstream of work, including that on informed consent, on the resource requirements of providing a satisfactory service, on people with false-positive results and on partners.

Publication

Green JM, Hewison J, Bekker HL, Bryant LD, Cuckle HS. Psychosocial aspects of genetic screening of pregnant women and newborns: a systematic review. *Health Technol Assess* 2004;**8**(33).

NHS R&D HTA Programme

The research findings from the NHS R&D Health Technology Assessment (HTA) Programme directly influence key decision-making bodies such as the National Institute for Clinical Excellence (NICE) and the National Screening Committee (NSC) who rely on HTA outputs to help raise standards of care. HTA findings also help to improve the quality of the service in the NHS indirectly in that they form a key component of the 'National Knowledge Service' that is being developed to improve the evidence of clinical practice throughout the NHS.

The HTA Programme was set up in 1993. Its role is to ensure that high-quality research information on the costs, effectiveness and broader impact of health technologies is produced in the most efficient way for those who use, manage and provide care in the NHS. 'Health technologies' are broadly defined to include all interventions used to promote health, prevent and treat disease, and improve rehabilitation and long-term care, rather than settings of care.

The HTA programme commissions research only on topics where it has identified key gaps in the evidence needed by the NHS. Suggestions for topics are actively sought from people working in the NHS, the public, consumer groups and professional bodies such as Royal Colleges and NHS Trusts.

Research suggestions are carefully considered by panels of independent experts (including consumers) whose advice results in a ranked list of recommended research priorities. The HTA Programme then commissions the research team best suited to undertake the work, in the manner most appropriate to find the relevant answers. Some projects may take only months, others need several years to answer the research questions adequately. They may involve synthesising existing evidence or designing a trial to produce new evidence where none currently exists.

Additionally, through its Technology Assessment Report (TAR) call-off contract, the HTA Programme is able to commission bespoke reports, principally for NICE, but also for other policy customers, such as a National Clinical Director. TARs bring together evidence on key aspects of the use of specific technologies and usually have to be completed within a limited time period.

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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 monograph was commissioned by the HTA Programme as project number 93/56/99. As 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.

ISSN 1366-5278

Published by Gray Publishing, Tunbridge Wells, Kent, on behalf of NCCHTA.
Printed on acid-free paper in the UK by St Edmundsbury Press Ltd, Bury St Edmunds, Suffolk.