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Non-invasive prenatal diagnosis: Public and patient perceptions

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Summary The possibility of the application of reliable non-invasive prenatal diagnosis to clinical practice, and its likely availability as a tool for routine antenatal screening, is an important and exciting development that will be of interest to women and couples – especially where there is a known risk arising from family history, or some other source, of having a baby with a serious, disabling or life-limiting condition. Managing the introduction of this new technology will require attention to the understanding and perceptions of women, couples and the wider society, as well as to the clinical, scientific, technical and logistic issues that will inevitably arise.

Introduction

For women and couples expecting a wanted baby, the desire to have a healthy child often outweighs all other considerations. Until relatively recently, obstetrics could offer little or nothing in response to the question 'Will my baby be all right?' beyond general lifestyle and dietary advice, and statistical risk prediction based on Mendelian inheritance in families with a known history of genetic disease.

Scientific and technological developments have opened up novel ways of generating information. Ultrasound pictures (of ever improving quality) and invasive procedures such as amniocentesis and chorionic villus sampling (CVS; neither of which is risk free), coupled with advances in DNA testing that can identify mutations in genes responsible for an ever growing number of serious and potentially life-limiting diseases in the developing fetus, have provided parents with information of increasing complexity and accuracy. However, until now, confirmation of the presence or absence of a given genetic disease in the fetus tended to require an invasive procedure, which carried a degree of risk to the pregnancy that was, in itself, a source of stress to women and couples. It also served to deter some women and couples who might otherwise have wanted to know the health status of their unborn baby (in order to have reproductive choice) but who were unwilling to accept the risk of losing a wanted pregnancy.

The possibility of the application of reliable non-invasive prenatal diagnosis (NIPD) to clinical practice, and its likely availability as a tool for routine antenatal screening, is an important and exciting development that will be of interest to women and couples, especially those at known risk (arising from family history) of having a baby with a serious, disabling or life-limiting condition. Managing the introduction of this new technology will require attention to the understanding and perceptions of women, couples and the wider society, as well as to the clinical, scientific, technical and logistic issues that will inevitably arise.

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Patient and family issues

As and when NIPD is brought into routine clinical use, a critical prerequisite will be to have a well-developed communication strategy in place. It will be important to be clear about the limits and possibilities of this novel technology. Issues that will need to be laid out include (in no particular order):

- The conditions for which NIPD can be used to provide a diagnosis.
- When it can be done (i.e. how early in the pregnancy).
- How accurate and reliable it is.
- What is involved in the procedure.
- Whether it will hurt.
- Information about previous pregnancies (if any).
- How long will it take to get the results.
- How these will be explained (and by whom).
- Whether the service provided is for everybody, or whether is there something about a particular individual/couple that makes the procedure appropriate/necessary/a good idea. If so, what are these reasons?
- If a result is 'bad', what happens next — what are the options?
- How long the individual/couple will have to make up her/their mind as to what to do.
- Whether anyone offers additional information, advice and support.
- What the condition being tested for means for a child. What effect will it have on him/her?

Not all parents will need to know the answers to all the above. Some will ask other questions, which might appear idiosyncratic to the members of the clinical team but which are pertinent to the couple in their particular situation. The use to which couples put the information they receive in response to their questions might also not fit with preconceived ideas. However, properly supported, women and couples are more likely to reach conclusions with which they are comfortable and with which they can live in the long term.

Many or all of the above questions are not unique to NIPD. They apply equally in situations in which invasive procedures are currently the intervention of choice, but in these cases the number of women affected by such decisions is a small proportion of all pregnant women. NIPD has the potential to become a routine population-screening tool, involving many more healthcare professionals in discussions with families. One group likely to be particularly affected will be community midwives who, to date, are not often actively involved in the introduction of the notion of genetic risk to the fetus, although they do discuss the risks of Down syndrome. This will have significant training implications if it is to be managed successfully.

Further issues need to be considered if communication regarding the nature and purpose of NIPD is to be managed successfully, and patients and families understand the nature and context of the 'offer'. The first of these relates to the fact that, although many of us are capable of assimilating complex, high-level concepts in contexts in which we feel comfortable and when we are operating in a familiar milieu, when things are unfamiliar and/or we feel under stress, our ability to process and retain information can be seriously compromised. NIPD can be a difficult idea to grasp; antenatal screening is not without stress for women and couples. Both of these factors can conspire to make effective clinical communication more difficult and, in the event of an adverse result, leave those on the receiving end of this unwelcome information feeling bewildered and less prepared for the potentially traumatic decisions that they have to face.

Second, the UK is an increasingly multicultural society. Significant fractions of the population come from communities and cultures with languages, attitudes and beliefs that differ significantly from those of the indigenous population (see Chapter 7). Again, whereas many or all members of a particular ethnic minority might be able to function perfectly well in English in everyday life and normal work contexts, understanding sensitive medical information is often easier and more effective if it is delivered in the recipients' mother tongue.

Translating information from one language to another is more than a matter of substituting words. Similes and metaphors that convey meaning in English might be utterly incomprehensible to speakers of other languages. Cultural sensitivities need to be respected, as a failure to do so will reduce the likelihood of messages being conveyed accurately. Above all, translations and spoken interpretation must use terms that reflect the same intended concept accurately. A survey by the Genetic Interest Group (Pritti Mehta)[1] found a number of examples of significant errors in translation, probably arising from the fact that the translator did not adequately understand the material he/she was asked to translate and resorted to guesswork when faced with technical terms in English. One particularly glaring example arose in a leaflet in which a 'chromosomal abnormality' became a 'pigmentation disorder' in translation. The possible consequences of such an error in the context of NIPD can all too easily be imagined.

Reaching ethnic minority communities

Awareness of the above problems, and in the light of long-standing inequalities of access to NHS services and support by some minority communities, led the Genetic Interest Group (GIG) and the London IDEAS Genetic Knowledge Park to undertake a project to provide high-quality genetic information to some, at least, of the groups whose mother tongue was not English. This ultimately resulted in 38 leaflets about various aspects of genetic disease being translated into eight languages: Arabic, Gujarati, Punjabi, Somali, Sylheti, Turkish, Urdu and Farsi. A robust protocol for translation was used, involving texts in English that were checked for their understandability by bilingual speakers of the relevant language, and then translations that were tested by monolingual (non-English) speakers of the same language. Of particular significance was the construction of a glossary of relevant technical terms in each language. These were checked to ensure scientific accuracy and appropriate understanding of their meaning by the non-English speakers. This information will be freely