

Non-invasive prenatal diagnosis of Down Syndrome and other conditions in the UK

The use of cell-free nucleic acids for non-invasive prenatal diagnosis is relatively new technique that involves the extraction and testing of fetal DNA (or RNA) from a sample of maternal blood. This technology has significant potential to improve various forms of antenatal testing for specific genetic conditions and warrants close scrutiny over coming months as the technology develops. Further evaluation will be needed to determine just how effective it is for different applications before it can be offered responsibly by the NHS.

It is important to note that the approach is not currently available as a routine NHS service. At present it is only being used on a research basis in selected centres for women who are at high risk of passing on certain specific serious inherited diseases, or of developing serious complications of pregnancy related to blood-group incompatibility. Although in the longer term, there is potential for the technology to be used within the NHS for a variety of other applications including the diagnosis of Down Syndrome, this is not yet possible in the UK.

A working group of technical experts, NHS service providers and wider stakeholders has recently been convened to assess the prospects for this technique in the UK for the [Joint Committee on Medical Genetics of the British Society of Human Genetics](#), the [Royal College of Physicians](#) and the [Royal College of Pathologists](#). Led by the PHG Foundation in Cambridge, the work will result in a report detailing their conclusions and recommendations, including proposals for the NHS to ensure that it is able to respond promptly and effectively to scientific developments in this area. **The report will be released by the PHG Foundation early in 2009.**

Bodies represented in the expert group include the British Maternal & Fetal Medicine Society, the Royal College of Obstetricians and Gynaecologists, the Royal College of Midwives, the UK National Screening Committee, the Human Genetics Commission and the Genetic Interest Group and Antenatal Results and Choices charities, as well as expert scientists and clinicians, NHS managers and policy makers, and experts in law and ethics.

The group's preliminary conclusion is that before any of these techniques can be used in routine care a number of things need to happen:

- Techniques need to be validated in large numbers to determine accuracy and will not replace invasive testing for some time
- Technological development is required to produce machines that can cope with a high throughput of samples
- Some methods generate a huge amount of data that requires careful analysis and this is an area that will require development.
- Laboratory standards will need to be developed
- The limits of gestation for testing will need to be determined
- Non-invasive techniques have the potential to replace current Down's syndrome screening tests with a test that would be diagnostic and we must be sure that women and healthcare professionals understand the changes and women fully understand the implications of these tests.

- Ends -

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Notes to Editors

- The PHG Foundation is a not-for-profit, multidisciplinary health policy research and development organisation based in Cambridge, UK, but working internationally.
- We focus on 'bridging the gap' in clinical translation to move new genomic and biomedical knowledge and applications into health service practice and better health for people and populations.
- We are the leading UK centre for public health genomics, and have considerable experience in policy research and development, as well as evidence-based evaluation of clinical and scientific tools.
- Our staff have broad-ranging expertise in areas including the scientific, clinical, public health, ethical, legal, social and regulatory aspects of genomics and biomedicine.

The PHG Foundation is the working name of the Foundation for Genomics and Population Health, a charitable company registered in England and Wales, charity no. 1118664 / company no. 5823194.

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