

## Expert recommendations for non-invasive prenatal diagnosis (NIPD) in the UK

The detection of fetal DNA (and RNA) in the mother's blood from early in pregnancy is an exciting new technique with potential to improve various forms of antenatal testing for specific genetic traits, by allowing earlier diagnosis without the risk associated with current invasive techniques.

A new report from the PHG Foundation in Cambridge, *Cell-free nucleic acids for non-invasive prenatal diagnosis*, sets out the findings of a working group of technical experts, NHS service providers and wider stakeholders for the Joint Committee on Medical Genetics (JCMG) of the Royal College of Physicians.

Funded and led by the PHG Foundation, this group was convened to assess the prospects for this technique in the UK and implications for the NHS. Bodies represented include the Royal College of Obstetricians and Gynaecologists, the Royal College of Midwives, the British Maternal & Fetal Medicine Society, 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.

Conclusions and recommendations include:

- Reliable non-invasive prenatal diagnosis using fetal DNA is already possible from the first trimester of pregnancy for some applications and likely to become available for others within the next 3-5 years.
- The NHS should take steps now to ensure that it is able to respond in a timely and appropriate manner as the technology develops. This includes formal evaluation for different purposes, development of specified care pathways and national best practice guidelines, and oversight from the appropriate authorities.
- Both public engagement efforts and health professional education about the potential and limitations of the technique are urgently needed.
- The technique should only be used according to standardized protocols within agreed clinical pathways, with formal audit and monitoring processes, and quality assurance frameworks. This will ensure appropriate use and accurate, reliable results.
- Some applications of non-invasive prenatal diagnosis are already available privately on a direct-to-consumer basis, and may have an increasing impact on NHS primary care and antenatal services. A voluntary code of conduct should be supported to help ensure the quality of private services.
- Ethical and social issues associated with some of the broader implications of non-invasive prenatal diagnosis using fetal DNA warrant further consideration and research.

Responding to the report, chair of the Joint Committee on Medical Genetics Professor Trevor Cole said: *"The PHG Foundation has led a large multidisciplinary expert group to produce this timely appraisal of non-invasive prenatal diagnosis. The report provides an authoritative account of the technologies and their current and possible future uses in clinical care and sets out clearly the many issues that will need to be addressed. Organisations and individuals with an interest in this area should use it as a valuable source of information about the technology and its wider implications, and as a starting point for further work. The Joint Committee is very enthusiastic about the opportunities NIPD will provide for patient benefit; however, it endorses the cautious approach set out in the report, in which the excitement and opportunities of technology advances need to be balanced against the necessity of properly validated techniques, effective and patient centred clinical services and responsible actions of society"*.

Jane Fisher, Director of the UK Charity Antenatal Results and Choices (ARC) and a member of the expert working group, said that she was pleased that the NHS had the opportunity to respond in a

timely manner to technological developments, adding: *"We take calls every day on our national helpline from women agonizing over whether to have an invasive test and desperate to know when a safe alternative will be available"*.

PHG Foundation Programme Director Dr Hilary Burton commented: *"Technologies for safer, non-invasive prenatal diagnosis using cell-free fetal nucleic acids are advancing rapidly and there will inevitably be enormous pressure from patients and clinicians for translation into clinical practice. This Report assembles the best evidence available combined with expert opinion to ensure that the NHS responds in a timely, effective and responsible way to these exciting new opportunities."*

The full report, including separate executive summary and appendix relating to ethical, legal and social issues, and a condensed overview, are available from the PHG Foundation website, [www.phgfoundation.org](http://www.phgfoundation.org)

- Ends -

For more information please contact:

Dr Philippa Brice

[philippa.brice@phgfoundation.org](mailto:philippa.brice@phgfoundation.org)

07505092081 or 01223 741506

[www.phgfoundation.org](http://www.phgfoundation.org)

#### Notes to Editors

- The PHG Foundation is an independent, 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 tools and health services.
- Our 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.*

*Registered address: Strangeways Research Laboratory, Worts Causeway, Cambridge, CB1 8RN, UK.*

*Telephone: +44 (0) 1223 740200*

*Fax: +44 (0) 1223 740 892*