

Health service approach to genetics in medicine needs to change

A new paradigm for using genetics in mainstream medicine is needed for acceptable patient care

A new report from the PHG Foundation challenges the existing NHS approach to the use of genetics in general clinical services, and encourages mainstream medical specialities to adopt genomic knowledge and technologies as part of their standard care pathways and professional training.

Patients with inherited disorders and disease predispositions represent a significant proportion of those seen in 'mainstream' medical services - for example, cardiology, ophthalmology and renal clinics. There is increasing potential to improve their care by taking into account emerging understanding of genetics and new forms of genetic testing.

For example, genetic testing of a newborn baby diagnosed with congenital hearing loss could diagnose Usher Syndrome; if this is the cause then sadly progressive loss of sight is also likely, making it vital to maximise hearing as soon as possible, perhaps by the use of cochlear implants. Similarly, a patient with Long QT Syndrome, a heart rhythm abnormality, might respond to different treatments or be at risk of sudden cardiac death from different triggers, depending on the presence of specific genetic mutations.

The UK Government's 2003 Genetics White Paper (*Addressing genetics, delivering health*) set out a vision of diffusion from specialised clinical genetics into other areas of medicine. However, research from the PHG Foundation has revealed that it is not working effectively. Although where geneticists work with other specialists, the joint services they provide are typically very good, access is very variable, and too many patients miss out.

The new report presents an alternative strategy where, rather than genetics moving into mainstream medicine, these clinical areas develop and expand to integrate new genetic expertise and genomic technologies firmly within their own care pathways. Regional genetics services should play a key leadership role. This will deliver the immediate benefits of genetics for their patients, whilst building capacity to expand and improve services in the future as new tools become available.

In the twenty-first century, genetics is no longer an optional extra, and must become a key element of mainstream services.

The full report, *Genetics and mainstream medicine*, including recommendations, is available from the PHG Foundation website: www.phgfoundation.org

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Notes to Editors

- The PHG Foundation is a genetics policy think-tank and health service development NGO based in Cambridge, UK
- Our mission is *making science work for health* - identifying the best opportunities for 21st century genomic and biomedical science to improve global health, and to promote the effective and equitable translation of scientific innovation into medical and public health policy and practice.
- We generate knowledge, evidence and ideas to inform, educate, and stimulate debate
- We also provide expert research, analysis, health services planning and consultancy services for governments, health systems, and other non-profit organisations

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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