

## Delivering the benefits of genetics across mainstream medical practice



Traditionally, genetics has been the preserve of specialised clinical genetics services. However, this situation is changing rapidly as genetics offers increasing opportunities to deliver improved care and management for patients in many different areas of medicine, from cancer to cardiovascular disease, making it an essential component of mainstream health services.

Current UK policy is based on knowledge and expertise spreading out from clinical genetics into different areas of mainstream medicine. However, research from the PHG Foundation challenges this approach, with increasing evidence that it is not functioning effectively. Although where geneticists work with other specialists, the joint services they provide are typically highly regarded, access is very variable, and too many patients miss out.

*Genetics and mainstream medicine* presents an alternative paradigm whereby mainstream clinical areas develop and expand to integrate new genetic expertise and genomic technologies into their own clinical pathways, with regional genetics services playing a leading role. This will deliver the immediate benefits of genetics for patients, whilst building capacity to expand and improve services in the future as new tools emerge.

*“The strategies identified in this report will be key to ensuring health services are ready to take advantage of the new genomic technologies”*

## Why genetics matters

Patients with genetic disorders form a significant sub-group in many clinical areas, with screening programmes identifying growing numbers of those who may be at risk. Fortunately, there is also increasing scope to help them.

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 in some cases by the use of cochlear implants.

Similarly, patients with Long QT Syndrome, a heart rhythm abnormality, may respond to quite different treatments or be at risk of sudden cardiac death from different triggers (such as exercise or loud noises) depending on the presence of specific genetic mutations.

## The current picture

The 2003 Genetics White Paper *Our inheritance, our future* set out UK health service plans to diffuse genetics services from newly modernised laboratories into GP surgeries, health centres and local hospitals.

Between 2006 and 2009, the PHG Foundation reviewed two clinical areas in which the diagnosis and management of people with inherited disorders is already very important - ophthalmology and cardiovascular disease.

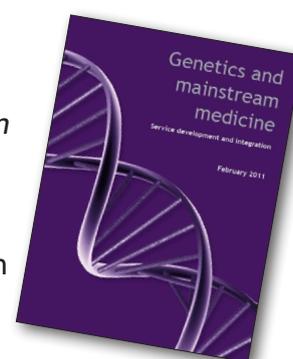
We found that joint clinics with geneticists and other clinical experts provide excellent care and are highly regarded by patients and families. However, our findings also showed that access to such services is highly variable across the UK, and current inequity is likely to worsen as the demand for input from clinical genetics to other specialities increases.

## The way forward

Leadership and close co-operation between key stakeholders, including commissioners and providers of both specialised mainstream clinical and regional genetics services, will be essential. Key recommendations in the report include:

1. **Commissioning guidance and clinical pathway development for clinical areas with significant inherited disease elements**
2. **A programme to ensure access to high quality specialised services for inherited disease within these clinical specialities**
3. **Review of genetic test provision, family history taking and cascade testing**
4. **Sub-specialisation in inherited disorders by health professionals**
5. **Provision of effective and ongoing specialist genetics support for other clinical services**

*Genetics and mainstream medicine: service development and integration* is available from the PHG Foundation



The PHG Foundation is an independent, non-profit organisation working to ensure that biomedical innovations are put to the best use in health services.

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