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Case study: Genetic Ophthalmology in Focus

Introduction: the size of demand

Eye disease and visual impairment are very common, and severe visual impairment or blindness is a significant form of disability. In children, it affects their development and education; in adults, it has many implications for their independence and ability to work. It affects social and psychological well-being, and places a burden of care on families and health and social services.

Each year in the UK, about 450 children and 2,500 people of working age are diagnosed as blind or severely visually impaired. Of these, 150 children and 250 adults will have an inherited disorder. Although individual single gene diseases are rare, taken together they are significant, with consequent demands on health services. In younger age groups, and in the UK's South Asian population, a higher proportion of sufferers have conditions that are inherited. Genetic testing is becoming more important, and our ophthalmology services need to keep pace with new knowledge and technologies.

Latest research also suggests that genetic factors play a complex role in the common eye diseases of later life, for example age-related macular degeneration ('AMD'), glaucoma and cataract, which result in many thousands of older people losing vision and requiring care. And as people live longer, demand for testing, diagnosis and treatment will increase.

In 2006, PHG Foundation was asked by the UK Genetic Testing Network[1] to convene an expert working group, chaired by Professor Tony Moore of the Institute of Opthalmology, to look at how NHS ophthalmology services need to change in response to the opportunities offered by genetic science. The results are published in our report *Genetic Ophthalmology in Focus* (April 2008) and are summarised here.

Current clinical and laboratory services

Genetic eye disease is a specialist area within ophthalmology, and requires an experienced and multidisciplinary team of professionals, with specialist facilities for electrophysiological testing, genetic testing, counselling and care. We found some specialist provision in each area of the UK, though outside London many services lacked access to the full multidisciplinary team. Some of this deficit reflects a lack of suitably trained and experienced staff.

Although there are some weaknesses in the data available, we are confident in concluding that there is significant variability in the scope and spread of services across the population. The best-served regions provide seven times as many clinic sessions and see seven times as many patients than the worst-served. Specialist services are highly focused on London and a few other regional centres, elsewhere there are small services that lack critical mass. We estimate that around 1,000 patients across the UK are not having their needs adequately met.

We found a lack of formal arrangements for referring patients from local ophthalmology clinics and children's services. Most genetics services do not attempt to reach out to their potential users in a systematic way and it is likely that many eye clinics and other services are not aware of the existence of specialist genetics services for ophthalmology.

1 Established by the Department of Health in 2003, the UKGTN is a network of NHS laboratories, health care providers and commissioners that exists to promote the quality of, and equity of access to, genetic services in the UK National Health Service

Patients' views

We invited patients and voluntary organisations (see box 1) to tell us what they thought about the value of genetic testing and their experiences of using ophthalmology genetics services.

Patients want access to a specialist service that is up-to-date, and experienced in diagnosing and managing their particular condition. They want care to be integrated – between specialist services and eye clinics; and between their local health services and, where relevant, social services, education or employment services and voluntary organisations.

The quality of service is more important to patients than the distance they have to travel to access it. By their nature, genetic disorders may affect other members of the patient's family, and they want services to be able to encompass other family members, even where they may live outside the normal catchment area.

Good communication and the availability of counselling were felt to be important attributes of a quality service. Patients want to be kept fully informed of advances in testing and treatment. They want counselling to help them to understand the diagnosis and its implications, for emotional support, and to help make decisions about the effects on family, education, employment and mobility.

Patients are keen to be part of the research effort and they want to be kept up to speed with research developments. Those who had taken part in research wanted to be informed of the outcome and, where appropriate, be referred for treatment when the research trial had finished.

Asked about their experience of current services, patients highlighted a number of concerns pointing to the lack of a consistent, high quality service across the UK. Many reported that their GP, optometrist and even hospital eye clinics didn't know what specialist genetics services were available. Patients placed a high value on genetic counselling but this was often not available to them in eye clinics.

Genetic testing

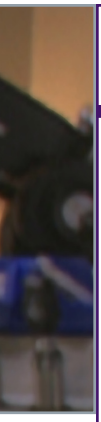
Developments in molecular genetic testing mean that it is increasingly possible to make a precise diagnosis of genetic eye disease, something valued by both patients and doctors. A genetic test might allow clinicians to avoid other complex, expensive or time-consuming diagnostic procedures and lead directly to a definitive diagnosis, information about prognosis, possible treatment or surveillance programmes. It might provide other family members with testing options to assess their own risk of disease; and it might provide parents with options for prenatal testing for subsequent children.

However, patients' experiences of current genetic testing services were inconsistent and they reported that the absence of a proper, simple and integrated management process often left them confused. Importantly they were not as concerned about the speed or location of laboratory services as they were about service quality.

On the horizon

The need for genetic testing is set to expand. Research is already yielding new treatments that are gene-specific and so require prior genotyping to be carried out. This is driving patient demand for molecular testing to enable them to take part in clinical trials. As these treatments become proven and mainstream, there will probably be an increasing need for genetic testing within NHS services. New technologies are also making diagnosis possible for a wider range of disorders.

Finally, there is the real expectation that, in the future, knowledge of genetic factors will play a significant role in the prevention, early diagnosis and management of some of the common eye diseases that are the main cause of visual loss in the general population. Genetic testing will therefore become important within the general ophthalmology service as well as services dealing specifically with genetic eye disease – again meaning that a wider range of professionals in ophthalmology will need expertise in genetics.



Conclusions and recommendations

Commissioning

All regions in the UK have some specialist service provision but small-scale services often lack specialist elements, leading to inequality of access for patients.

- Primary Care Trusts need to be supported in commissioning specialist ophthalmology services with coverage of the entire population
- A service specification with defined quality standards should be developed
- Commissioners in areas where currently there is little or no specialist service for genetic eye disease should urgently review how their patients with these conditions are managed

Provision of specialist genetic ophthalmology services

The need for accurate diagnosis, prognosis and counselling, and the advent of new therapies for ophthalmic genetic conditions make it imperative that ophthalmology genetics is configured to adequately serve regional populations, with multidisciplinary specialists and effective national laboratory support.

- Existing services should develop integrated systems for providing multidisciplinary care now and to respond effectively to future needs
- Explicit care pathways are needed to facilitate access to the specialist service and new technologies, and their performance should be monitored and evaluated
- Specialist ophthalmology genetics services should publish agreed standards of care
- Services should work together to better coordinate their management of patients with complex disorders whose sight problems are only one of a range of symptoms
- A service network should be established to share best practice
- Overall the NHS should increase capacity in medical, surgical, nursing, genetic counselling, electrophysiology and other services that support ophthalmology genetics
- Services should embrace the support offered by voluntary organisations, particularly in providing assistance and information for patients and their families

Laboratory services

In the UK, tests for 18 conditions are available from 9 different laboratories. Not all of these tests have been evaluated or are 'listed' by the UKGTN. There is lack of clarity about what tests can be requested and who can request them. Difficulties with funding are experienced, especially where funding for genetic testing comes from a different local budget.

- The UKGTN should continue its role in monitoring, evaluating and prioritising available genetic tests
- The UKGTN and NHS laboratories should work together to develop the best model for providing genetic testing for ophthalmology
- Laboratory services should be prepared to increase the volume of testing
- Further work should be done now on access to testing and future demand, to prepare for increases in the number of tests available and demand for testing for common eye diseases
- An urgent review is needed of the way in which genetic testing in ophthalmology is funded
- Consideration should be given to the use of commercial laboratory services and the ways in which they might be integrated with NHS systems

Ensuring access to specialist services

Locally, health professionals often lack knowledge of rare eye conditions and do not know that specialist services exist. Most specialist centres do not have any formal outreach activity to raise awareness of their service and how to use it.

- Specialist genetic ophthalmology services should work with hospital ophthalmology departments to develop and implement systems for referral and shared care
- Providers and commissioners should devise a way of ensuring that information about specialist services is available to health professionals when they need to refer patients
- Specialist centres should work with voluntary organisations to raise awareness of their services amongst patients and their families

Education and training for health professionals

Specialist education and training is needed to increase capacity and manpower. Within ophthalmology all should have a basic knowledge of the relevant genetics.

- Professional organisations, Royal Colleges and training providers should be encouraged to provide specialist training for geneticists, ophthalmologists, genetic counsellors, ophthalmology nurses and electrophysiologists in the management of genetic eye disease
- The National Centre for Genetics Education and Training should be involved in education about genetic eye disease for these groups
- Opportunities should be sought to raise awareness through professional publications, conferences and meetings

Box 1: Focus group participants

RP Society (Retinitis Pigmentosa)
RNIB (Royal National Institute for the Blind)
Contact-A-Family
Macular Disease Society
International Glaucoma Association
Action for Blind People
Childhood Eye Cancer Trust
Usher Research
Sense (the National Deafblind and Rubella Association)
Patients receiving care from the Oxford Regional Genetics Service

Box 2: Members of the Project Working Group

Professor Tony Moore (Chairman), *Institute of Ophthalmology, UCL*
Dr Hilary Burton (Project Manager), Programme Director, *PHG Foundation*
Ms Corinna Alberg (Coordinator), *PHG Foundation*
Professor Graeme Black, Clinical Geneticist, *University of Manchester*
Ms Sue Carless, Genetic Nurse Counsellor, *Birmingham Hospital*
Mr Clive Fisher, Board Member, *British Retinitis Pigmentosa Society*
Dr Rajalakshmi Lakshman, Specialist Registrar, *PHG Foundation*
Ms Sue Lydeard, Research Manager, *Moorfields Eye Hospital*
Dr Simon Ramsden, Molecular Geneticist, *St Mary's Hospital, Manchester*
Mr Ananth Viswanathan, Consultant Ophthalmologist, *Moorfield's Eye Hospital*
Professor John Yates, Medical Genetics, *University of Cambridge*

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For a copy of the full report or a large print version

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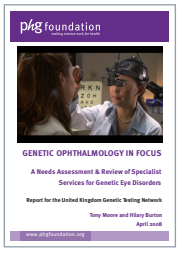

Challenges in 'mainstreaming' genetics

In 2003, developing genetics in mainstream health services was one of the main themes of the Government's Genetics White Paper. Five years on, our experience has highlighted that, irrespective of the specialities involved, there are some common challenges that inhibit the translation of genetics research and biomedical innovations into mainstream medical care:

- Lack of service capacity and capability
- Complexity of the underlying genetics
- Technological barriers
- Cost and funding issues
- Lack of data for evaluation and funding decisions

Genetics services will be facing ever increasing demands from mainstream medicine as the number of diagnostic tests increases and requests for genetic testing become routine. Increasingly knowledgeable and mobile patients will demand referral to specialist centres. But genetics is a small specialty with dispersed service providers who lack the resources to raise their local profile or to exert the necessary pressure for change.

There is rarely any strategic planning in specialist areas. Developments in genetics have tended to be led by enthusiastic providers, resulting in uneven services that are vulnerable to commissioners' funding decisions. It is usually difficult to engage commissioners in planning for these specialist areas because of the inherent nature of the work: the diseases individually are rare; data and evidence are not routinely collected to show that services and tests are beneficial; the benefits are not immediate, often concerned with prevention rather than treatment; and beneficiaries (family members) may be outside the commissioner's area.

PHG Foundation believes that health services can and should be shaped now in ways that can capitalise on genomics advances. We believe that detailed examination of services, such as this latest study of ophthalmology genetics, and involving stakeholders and service users, are necessary steps in translating science into health services. We have developed a model that can be used to identify the opportunities offered by genomics and the barriers that have to be overcome, and to recommend to policy makers and service providers the ways in which health services need to adapt. Public funding concentrates on basic research and translational research, and this interface between research and practice is relatively neglected. This is the gap in translation that PHG Foundation seeks to fill for the benefit of population health.

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