ABSTRACT

Background Scientific advances in the understanding of the molecular biology of inherited eye conditions now allow more effective diagnosis and management for patients and families. For translation into clinical practice, it is vital that specialist services are developed with the necessary multi-disciplinary expertise, investigatory resources and organizational arrangements. We investigate the equity of specialist provision in the UK and make recommendations for service development.

Methods A questionnaire survey was carried out of all providers of specialist genetic services in the UK. Results were analysed by provider, catchment population and Strategic Health Authority population.

Results Nineteen specialist services were identified. Provision of annual out-patient clinics and medical consultant sessions varied widely with many small services lacking full multi-disciplinary teams. There was an 8-fold regional variation in patient activity. Across the UK, we estimated an annual shortfall of 1000 new patient referrals.

Conclusions There should be a national programme of strategic planning of specialist genetic ophthalmology services. Necessary elements will include service specifications and standards, overall number and configuration of services, models which maximize the efficiency of use of specialist genetics elements and education of specialist and general ophthalmologists in genetics elements of their speciality.

Keywords eye disorders, genetics, public health

Introduction

During the last two decades, rapid scientific advances have been made in understanding genetic causes of rare Mendelian disorders affecting the eye. With the identification of genes associated with the hundreds of inherited eye conditions, there are improved possibilities for diagnosis and management of patients and their families and for prenatal diagnosis. The challenge is now to translate these new capabilities, particularly those resulting from new genetic tests, into services to meet the needs of this patient group. Throughout the UK, the service response has largely been through the development of various joint and specialist services but so far this has been without formal planning or commissioning. In 2006, an expert working group was set up by the UK Genetic Testing Network to undertake a needs assessment and service review for genetic ophthalmology and to make recommendations for key issues in service development. This paper focuses on the comparative review of services and recommendations for action.

Inherited conditions affecting the eye form a large and heterogeneous group. Although each is rare, together hereditary disorders are thought to contribute to one-third of the severe visual impairment or blindness diagnosed in children (about 150 each year in the UK) and about 16% of adults of working age who receive blindness certification (about 250 adults each year in the UK). Using available epidemiological birth prevalence estimates wherever possible, it is estimated that each year in the UK, there would be about 1300–1800 new cases.1

Despite their clinical diversity, inherited eye disorders need to be viewed as a specialist care group because of the particular range of clinical, investigative and laboratory expertise required in their management. The working group

Hilary Burton, Consultant in Public Health Medicine
Corinna Alberg, Project Manager
Anthony T. Moore, Professor of Ophthalmology
considered that patients need to be managed by a specialized multi-disciplinary team that integrates the assessment of the experienced ophthalmologist with that of the geneticist.

**Methods**

The working group was led by Professor Anthony Moore of the Institute of Ophthalmology in London and included ophthalmologists, geneticists and laboratory scientists from some of the main UK inherited eye disorders services as well as a genetic counsellor and a representative of patient voluntary groups. Project management and expertise in public health and epidemiology were provided by the Foundation for Genomics and Population Health (PHG Foundation) in Cambridge.

The main services in the UK were identified through an initial letter sent to heads of regional genetics services requesting information on services available for patients with inherited ophthalmic disorders, which might take the form of a joint clinic, combined genetic ophthalmology services or other special arrangements. A final list was agreed by the working group. Questionnaires were emailed to all services in November 2006 and followed up by further emails and telephone calls until a completed questionnaire was received.

Services were asked to provide a general outline of their structure, including the population served and target age groups; sessional commitment of consultant genetic and ophthalmology medical staff and genetic counsellors; and the numbers of specialist out-patient sessions provided. Services were asked to provide information on numbers of patients and/or families attending the service annually; if unavailable from routine data sets, this was estimated based on average numbers attending each out-patient session. Where activity was reported as family attendance, to allow for comparison, an average family was counted as two persons on the advice of members of the working group. Regional rates for the numbers of patients attending specialist centres and the provision of clinic time were calculated using Government Office regions population statistics for England and Wales and total population statistics for Scotland and Northern Ireland.

**Results**

Replies about the general availability of services were received from all 23 regional genetics services. Four regional genetic services reported that they did not operate a special service for this group of patients and in one service the specialist service was provided by outreach from the specialist Eye Hospital (Moorfields). Full questionnaire responses were received from all 18 regional genetics services providing services and from Moorfields Eye Hospital, which provides a specialist service.

**Services and populations served**

Specialist services were provided in 18 cities throughout the UK, with 17 services outside London and 2 in London, including a specialist service at Moorfields Eye Hospital. Only two genetic services outside London (one in England and one in Scotland) were not involved in special service provision for this patient group. The specialist Eye Hospital at Moorfields in London has an established Genetic Eye service with an extensive service serving London and Southeast and, to a lesser extent, the whole of the UK.

**Overview of services provided in responding centres**

Services mostly took the form of a joint clinic between genetic and ophthalmology services. The core of the service was usually a consultant clinical geneticist with a special interest in eye disorders working alongside an ophthalmologist with a special interest in inherited conditions. The clinics were mostly for diagnostic purposes although some also offered ongoing surveillance and management. One service was aimed primarily at children but all other services included children and adults, or were explicitly focused on the care of families.

While the majority of services offered a general ophthalmic genetic service, a number of specialist clinics existed, notably Von Hippel Lindau (VHL) clinics (4 centres) and retinal clinics (3 centres). Other specialities offered by a small number of services included services for specific disorders, notably, Marfan clinics, Stickler clinics and a neurofibromatosis clinic.

The Moorfields Eye Hospital model differed from all other services in the UK. Here most diagnosis and genetic counselling was carried out in specialized clinics by ophthalmologists with a special interest in inherited disorders. Some patients, for example, those with undiagnosed multi-system disease, complex developmental abnormalities or those wishing to consider prenatal diagnosis, were referred from this service to their local clinical genetic services. This service received referrals from across the UK but with the majority coming from London and the South East.

The data below refer to the 18 ‘joint’ regional services except where otherwise stated.

**Out-patient clinics**

The annual average number of out-patient clinics provided by the regional services varied from 5 to 100 with a median value of 17 (Fig. 1). This was not simply related to the
catchment population as the figure also shows that the number of out-patient clinics provided in relation to catchment population also varied greatly ranging from 1.7 to 90.0 per million with an average of 17.1 and a median of 10.5.

**Specialist medical, genetic counsellor and nursing staff supporting the clinics**

Seventeen services were able to provide details of average monthly contracted sessions of medical consultant (including genetic and ophthalmological) and genetic counsellor time assigned to specialist genetic ophthalmology work (Fig. 2). The formal commitment of medical sessions to the services was extremely variable from an average of one session per month in three services to over 20 sessions per month in one service. Three services had additional research or specialist registrar time.

Ten services (56%) had specific genetic counsellor sessions assigned, though the time commitment was usually

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**Fig. 1** Ranked average annual number of out-patient clinics and number of out-patient clinics per million catchment population by each service centre.

**Fig. 2** Average number of sessions provided by medical consultants (genetic and ophthalmology) and genetic counsellors per month in each service centre.
very small (less than one session per month). Three services had one to two sessions per week. Two services had an almost full-time commitment of genetic counsellor time: in one, this had been funded as part of the DH Genetics White Paper funding for service development in mainstream genetics. Three of the 10 services did not have specific genetic counsellors attached to the clinics, but counselling time was provided on a rotating basis.

Activity information
The total annual numbers of patients attending each service varied from 40 to 500 with a median of 97. Figure 3 shows that there were a large number of services seeing relatively few patients, with 50% seeing <100 patients per year and only three services seeing more than 300. For the two more established services seeing large numbers of patients, this was related to a large regional catchment population and did not translate into high rates per million. Overall, in relation to catchment population, the average number of patients seen by individual services varied from 15 to 314 patients per million with a median rate of 61.

Regional provision of specialist ophthalmology services
The data for out-patient session provision and out-patient visits were examined on a regional basis, so that comparisons could be made about equity of access to specialist ophthalmology services across the UK. These data include Moorfields Eye Hospital, which is the major provider of services for London and Southeast Coast. In view of the established referral practices around the southeast into London, the populations of the Strategic Health Authorities (SHAs) have been adjusted for the eastern regions to include only the old East Anglia primary care trusts (Norfolk, Suffolk and Cambridgeshire) and for South Central to exclude Berkshire. Excluded populations from both the SHAs have been added to the London and Southeast populations.

Some level of specialist service was available in each of the SHA areas and in Scotland, Wales and Northern Ireland, but this was very variable. Average annual provision of out-patient sessions per million population varied 11-fold between the best and the worst provided region (Fig. 4a).

Regional calculations of patient activity per million population included only new patients seen at Moorfields Eye Hospital, estimated at 25% of the 4777 total patients seen in 2005/06. Over the UK, there was an 8-fold variation between the worst and the best provided region in terms of numbers of patients seen per million population (Fig. 4b).

Estimates of new patient shortfall in the UK
A total of 4015 new patients were reported as accessing the services across the UK on an average annual basis. This includes 1194 patients seen in London and 2821 patients elsewhere in the UK. The new patient rate per million for the region that the expert group thought had the most comprehensive service in relation to its population (the Northwest) was 81.7. If this rate of 81.7 were applied to the whole population of the UK (60.6 million), we estimated that a total of 4951 patients should be in contact with

![Fig. 3](https://academic.oup.com/pubhealth/article-abstract/32/2/259/1609942/35a722f4ad01625669942) Ranked total number of patients seen annually and number of patients per million catchment population by each service centre.
services, representing a shortfall of about 1000 new patients not getting access to specialist services.

Discussion

Main findings of this study

In this study, we identified all 19 providers of specialist services for genetic eye disorders in the UK and obtained both quantitative and qualitative information from each one. All services were based in regional genetics services except one specialist provider in a major London eye hospital. There was huge variability in what was provided, with a large number of small services, many with very infrequent and limited consultant sessional input and out-patient provision, lacking access to the full multi-disciplinary team and with very low levels of patient activity. Examination of provision
What is already known on this topic

The rapid advances in understanding of the basic science underlying genetic causes of rare Mendelian disorders affecting the eye have arisen largely through the identification and mapping of the many genes associated with disease and characterization of mutations associated with the various phenotypes. These have turned out to be very complex; for example, there are 130 genes associated with retinal disease and many different mutations associated with disease in each of the genes. This has allowed disease to be diagnosed at a molecular level and, in many cases, management fine-tuned accordingly.

Genetic testing has many uses, including providing more information about likely prognosis; enabling further preventive care (for example, by distinguishing between somatic and germ-line mutations in retinoblastoma and hence determining whether further close surveillance is needed); enabling those identified in the early stages of the disease to plan for the future in light of deteriorating vision; providing the basis for cascade-testing of other family members and enabling parents to consider prenatal testing for further offspring; finally, new treatments, such as gene therapy, are currently being trialled which are gene-specific and will require prior genetic testing.

The availability of more effective ways of diagnosing and managing patients and their families with inherited eye disorders thus creates a new health ‘need’ among this patient group (where need is defined as ‘an ability to benefit’ from a health service). In this clinical area, this need broadly translates into the need to be referred to, and managed by a specialist service where consultant geneticists and ophthalmologists with special expertise in inherited conditions work together. The working group considered that the service must also include high-quality ocular electro-physiological services, genetic testing and interpretation, genetic counsellors and specialist nurses, and systems of family records and follow-up to ensure effective and ethical follow-up of at-risk family members.

In the UK, services have responded, usually with the lead taken by clinical genetics services, and have created ‘joint clinics’ in which genetic and ophthalmology expertise is brought together. However, this growth has been largely piecemeal and opportunistic, for example, using service development money provided by the DH Genetics White paper initiative on service development in one region. With scarce resources, services have usually been limited in their outreach to referring consultants. Lack of awareness of inherited disease among clinicians in mainstream services, as catalogued, for example, in the recent House of Lords Science and Technology Committee on Genomic Medicine, has compounded this variation, which is thus a product both of limited provision and of limited demand.

No evaluation has been undertaken of the effectiveness of specialized services as a whole, and there are no published data on the level or organization of such service provision. Likewise, the international literature contains no examples of description or evaluations of specialized services for inherited eye disorders.

What this study adds

This study and associated Report provide the first consideration nationally and internationally of the need for specialized services for genetic eye disorders, bringing together expert opinion from a wide stakeholder group. With a 100% response from services identified in the UK, it provides a graphic description of the variation of specialist services throughout the UK. Lack of strategic planning has meant that many small services have developed that individually lack critical mass.

Limitations of this study

The survey was dependent on self-reports by service providers. Although inconsistencies and incomplete returns were followed up, no attempts were made to provide independent validation. We think that reporting biases would probably exaggerate the coherence of various elements of the service resulting in higher reported rates for ‘specialist’ activity—for example, services counting ophthalmology patients who were seen in general genetics clinics. There may have been some underestimates of activity by services not including the work of highly specialist clinics such as those for Stickler disease, or VHL disease.

Owing to lack of available data, new patient activity was related to region of care provision rather than of residence. The London service might provide some compensation for low levels of activity in the regions by providing a national service. However, we think that this is relatively minor, as the service noted that ‘the majority of patients come from London and the South East’. Similarly, some patient shortfall in regions may be made up by patients travelling to more distant services.
Conclusions

We conclude that there is a real need in the UK to develop the necessary capacity and quality of inherited eye disease services to ensure that all patients who need it can get access to specialist advice. This will require sustained strategic planning and leadership. Services for inherited eye disorders are ‘specialized’, in that they are high cost, low volume services, provided in relatively few centres and to a population >1 million. The obvious mechanism for development would be NHS processes for commissioning of specialized services, which are the responsibility of the 10 specialized commissioning groups set up following the Carter review. However, the current work of Specialised Commissioning Groups is focused largely in ‘national specialized definitions sets’, which do not cover services for inherited eye disorders and there is little capacity for ventures outside these national priorities.

Similarly, costing and pricing mechanisms can influence whether and how Trusts develop specialized services. Under current policy, they are nationally derived, but the grouping of conditions into Healthcare Resource Groups often means that complex services, such as those with multiple specialists, tend to lose out. Such mechanisms, together with the particular issue of funding expensive genetic tests, may deter the development of specialized services within ophthalmology.

The working group made a number of recommendations for strategic development on a national basis. This should preferably be led from the two specialties (ophthalmology and genetics) and could be taken forward by professional groups from the relevant Royal Colleges, including particularly the Royal Colleges of Ophthalmologists, Physicians and Pathologists. Close involvement of specialized commissioners and voluntary organisations will be vital.

(i) Standards of care should be agreed that include structure, composition and function of multi-disciplinary teams, access to specialist investigations and appropriate organizational arrangements such as referral protocols.

(ii) Consideration should be given to the optimum configuration of specialist ophthalmology genetics services in the UK, particularly the number and distribution of services that will lead to the best balance of high quality and reasonable accessibility.

(iii) The different models of service provision and the relative leadership and roles of genetics and ophthalmology elements should be evaluated. They should recognize that genetics capacity will be limited as genetics becomes important in many areas of mainstream medicine and that competence in genetic aspects of ophthalmology will increase within the specialty.

(iv) Capacity in inherited eye disorders should be increased by developing specialist education and training in inherited eye disorders for ophthalmologists wishing to specialize and for general ophthalmologists needing to recognize and refer possible cases.

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