Commissioning Guide: Ophthalmology services for patients with inherited eye conditions

Context
Recent innovations in the diagnosis and management of individuals with inherited eye disorders have led to improved outcomes related to visual disability and general health for patients and family members. Such benefits can be best achieved by specialist services. However, there is no specific NICE, HTA or other evidence based guidance related to this group of disorders. This Guide is based on the report of a working group of the UKGTN entitled *Genetic ophthalmology in focus: a needs assessment and review of specialist services for genetic eye disorders* and on evidence from clinical and laboratory experts active in this specialist field, with input from patient and voluntary organisations. Although conditions are variable, the unifying theme for this patient group is the need for a precise genetic diagnosis if possible and to provide management and counselling and advice for patients and families around inheritance issues. This requires the integration of two specialist disciplines (ophthalmology and genetics).

The Commissioning Guide is produced in the context of the revision in 2009 of the Specialised Services National Definition Set: 20 Medical genetic services (all ages) and the development of a new definition for specialised ophthalmology services.

Inherited eye conditions cover a wide range of conditions that affect the:
- Development (for example aniridia, anophthalmia or microphthalmia)
- Structures (for example disorders of cornea, lens, retina, or optic nerve) and
- Function (for example disorders with multi-system involvement such as lysosomal storage disorders)

Annex 1 provides a list of the main conditions to be included in the servic
A service for patients with inherited eye conditions will include:

- Identification of patients with suspected inherited eye disorders
- Appropriate referral
- Diagnosis (including specialist ophthalmological investigation and clinical and laboratory genetic elements)
- Communication and information giving about the condition and its implications
- Advice and provision of treatment
- Long term support and management
- Consideration and provision of counselling about risk of recurrence and risk to other family members and
- Follow up work with family members

**Current UK provision**

There are 19 services throughout the UK including one very well established national service at Moorfields Eye Hospital in London. In the regions, many services are very small and operate on an infrequent basis without the full establishment of health professional staff. For example, only five services see more than 200 patients per year and 5 see fewer than 50. Similarly, 8 services had no genetic counsellor. It is probably that lack of clinical capacity currently limits the amount of patient activity in smaller services and that in many SHAs the population as a whole has restricted access.

It is unlikely that small services can develop the necessary critical mass to gain sufficient experience of these rare conditions and to justify the investment in professional training (medical, genetic counsellor, specialist nurse, laboratory, and electrophysiology) and organisational development to develop into a fully comprehensive service. Commissioners will need to work together to decide where specialist services will be provided. All SHAs should not expect to develop their own service.

**Factors that are important in commissioning for service development include:**

- Collaboration of commissioners
- Involvement of clinical and laboratory service providers
- Involvement of patient groups
- The development of information systems to audit activity with respect to geographic area of residence
- The development of a local service specification including local service standards

**National priorities and initiatives include:**

1. World Class Commissioning
2. Delivering the 18 week patient treatment pathway
3. Expert patients programme
4. Transition: getting it right for young people
5. NHS Operating Framework
6. Genetics White Paper
Service models

Two main models currently exist:

The provision of joint genetic / ophthalmic clinics. This is the model currently provided in most centres in the UK and is recognised as being successful. However, to address the current under provision of service and anticipated future demands using this model throughout the UK would require a significant increase in resources, particularly from clinical geneticists, genetic counsellors and ophthalmologists. In the light of similar increases in demand as genetic aspects of other services develop (e.g. cardiovascular services, renal services etc.) this model might not be robust in the longer term.

An ophthalmology led service, in which ophthalmologists with special interest and training in genetics manage most of the patients with the support of a genetic counsellor and consult with geneticists regarding those with particularly complex diagnostic, ethical, counselling or interpretive issues. Currently this model is only available at Moorfields Hospital in London and, if expanded to other centres would require increased education and training of ophthalmologists, with possible issues of accreditation in inherited disease aspects and further provision of genetic counsellors. This model requires close working relationships with a clinical genetics department for supervision and provision of genetic elements of care. The service should also have access to genetics provided familial aspects of care including family record keeping and family follow-up or else provide this itself to the same standards.

Key clinical issues

Key clinical issues in providing an effective ophthalmology service for inherited eye conditions include:

Recognition of possible inherited eye disease by paediatricians, general practitioners, adult and paediatric ophthalmologists with appropriate referral for specialist advice

Ensuring that appropriate referral pathways are in place and well known and that the specialist genetic ophthalmology service is integrated with other services including primary and secondary care

Providing effective and efficient clinical care of these conditions by provision of multidisciplinary team experienced in diagnosis and management of patients and families with inherited eye disorders

Close involvement of patient organisations in the planning and delivery of care in order to optimise long term patient care and satisfaction

Audit and research to develop the evidence base for effective services and ensure innovations are translated into clinical practice
Specifying an ophthalmology service for patients with inherited eye conditions

Service components include:
- Ensuring access to specialist services for people with inherited eye disorders
- Ensuring timely and accurate diagnosis of people with inherited eye disorders
- Ensuring access to a high quality prevention, treatment and follow up service
- Meeting the needs of the family.

Ensuring access to specialist services for people with inherited eye disorders
Local clinicians in primary care and relevant areas of secondary care currently lack knowledge about inherited eye disorders and the existence of specialised services for diagnosis and management.

Commissioners should specify that specialist genetic ophthalmology services should:
- Work with district services and primary care to develop and implement protocols for referral
- Devise means of bringing referral protocols and information about services to the attention of relevant professionals
- Audit referrals by geographic area of residence and patient characteristics such as ethnicity
- Work in conjunction with voluntary organisations to raise awareness of services with patients and families.

Ensuring timely and accurate diagnosis of people with inherited eye disorders
To make an accurate diagnosis the service should have access to the following:
- Consultant ophthalmologist with special interest in inherited conditions
- Consultant paediatric ophthalmologist with special interest in inherited conditions
- Consultant clinical geneticist
- Genetic counsellor
- Laboratory scientist with expertise in molecular genetic testing specialising in eye conditions
- Electrophysiologist with experience of inherited conditions

These individuals should function as a multi-disciplinary team with joint input into diagnosis.

The following investigations should be available to support the diagnostic process:
- Specialised electrophysiology including high intensity flash; intermediate flash intensity; S cone electro-retinogram; On and Off responses; multifocal electro-retinograms; electro-oculogram; multichannel visual evoked potential; dark adaptometry; colour vision testing
- Molecular genetic testing, including conditions relevant to eye disorders listed by the UKGTN

Genetic testing aims to identify the particular mutation in the proband (first individual to contact services) with testing of further family members for that particular mutation only. The identification of a mutation is usually necessary if prenatal testing is to be undertaken.
Commissioners should require services to specify:

- How they will ensure services are aware of and can access UK Genetic Testing Network (UKGTN) listed tests
- The adoption of UKGTN testing criteria or other approved referral criteria for genetic testing
- How services will decide who can request a genetic test and relevant education or other support for those individuals
- The methods and personnel involved in gate-keeping
- How commissioners and services will monitor equity of access to testing

Ensuring access to a high quality prevention, treatment and follow up service for patients

Once diagnosis of an inherited eye disorder is made patients are likely to require long-term follow up. Although some routine aspects of care might take place nearer to their homes, in general patients value long term management by a consultant with expertise in the particular condition, who they trust will have pertinent information about the best methods of treatment and keep up to date with advances in clinical management. They also need to have continuing contact with services as their own circumstances evolve - for example, other family members, including their own children, might need testing or reproductive advice.

The specific preventive, treatment and follow up services are particular to the disease in question and are many and varied. Examples of particular interventions include:

- Diagnosis of the child with Usher syndrome (which presents as congenital hearing loss) allows the child, parents and school to be prepared for the onset of visual loss; currently this might involve early cochlear implantation
- Diagnosis of Alström disease (a multisystem condition including retinitis pigmentosa, deafness, cardiomyopathy and diabetes) enables surveillance and management to be set up for accompanying cardiomyopathy, diabetes or obesity
- Genetic testing in Norrie disease (a severe disorder in which retinal disease can be accompanied by developmental delay, behavioural difficulties and sometimes hearing loss) may allow carrier testing of mother and other female relatives in order to offer reproductive choice

Commissioners should require services to specify:

- How wider multi-disciplinary input into management plans is provided and coordinated
- How patients are enabled to contribute to and take responsibility for aspects of their own management plans
- How patients are followed up by the specialist service long-term
- How paediatric patients are supported in the transition to adult services
- How arrangements are made for routine care closer to home
- How patients and families are made aware of support provided by the voluntary sector
Meeting the needs of the family

The family needs of an ophthalmology service for people with inherited eye disorders involve identification and follow up of family members who might also be affected or who might be at risk of an affected child.

The ability to link patients and individuals in families through family records is also useful in achieving the most efficient means of testing where multiple family members have contacted different services at different times from different geographic locations.

Many services are currently run by joint genetic/ophthalmology services and the family record keeping is undertaken within the genetic department. At present this is a regional rather than a national service.

Commissioners should ensure that services set out:

- How family records will be kept and which service is responsible (genetics or ophthalmology). This should include necessary elements of consent and confidentiality and what links will be made between the genetic and ophthalmology records.
- How the decision to initiate family cascade testing is undertaken and who takes responsibility for it. How at risk relatives are approached and how any complex ethical issues in contacting relatives (such as estranged or adopted relatives) are resolved.
- How services are alerted to testing that might have taken place for another family member.
Commissioning a service for the management of inherited eye conditions

Determining Local Service Levels

Benchmarking data is based on epidemiological data and information derived from the survey of UK genetic ophthalmology services ¹.

Information on the incidence of blindness and severe visual disability due to inherited eye disorders is about 2.5 per million in children under 16 and about 4 per million in adults of working age. Information on total estimated birth prevalence of single gene disorders affecting the eye suggests that, in the UK, we would expect about 1500 new diagnoses per year (25 per million). As well as dealing with new cases, services are involved with patients in whom genetic eye disease is suspected but not eventually confirmed, including at risk family members.

Available data suggest that the standard benchmark rate for annual numbers of patients seen by the service is around 80 per million. This includes some patients seen individually and as families and is based on the rates in the Northwest of England which had one of the most comprehensive and well established services providing care for a well defined population. Rates were established by questionnaires to the services and the two services reported estimated average number of patients seen as 500 (Manchester) and 60 (Liverpool). With an SHA population in the Northwest of 6.8 million, the rate per million population is 81.7.

For a standard primary care trust population of around 250,000, or an average practice (list size 10,000) it is thus clear that such activity would not justify provision of a specialised service.

For an average Strategic Health Authority with a population of around 5 million this would amount to about 400 patients per year (including new and follow-up patients).

For the UK as a whole it is estimated that about 4,500 patients would be seen each year. This relates quite well to the possible incidence estimated from the literature, as all patients seen in the service would not eventually turn out to have inherited eye disorders.

Based on our survey, a service seeing 400 patients per year would require provision of between 50 and 60 outpatient sessions per year; between 16 and 20 medical consultant (ophthalmology and genetic) sessions per month; and between 20 and 30 genetic counsellor sessions per month (WTE).

*Figures for our benchmarking were derived from the service survey* ¹.
Data on average annual outpatient sessions, consultant and genetic counsellor provision from the more established Manchester service (serving 500 patients per year) were used to estimate the establishment required for an estimated 400 patients (the estimated rate for an SHA of 5 million). In the Manchester service the relevant data from the survey was as follows:

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<tr>
<td><strong>Average annual outpatient sessions</strong>*</td>
<td>70</td>
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<tr>
<td><strong>Average monthly consultant sessions</strong> (approximately half genetic and half ophthalmology) (note that this includes clinic time as well as administrative time)</td>
<td>21</td>
</tr>
<tr>
<td><strong>Average monthly genetic counsellor sessions</strong></td>
<td>30</td>
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* 1 session equals one half day

Currently there is no data on the number of genetic tests that would be required in the diagnosis of this number of patients, or the level of other specialist investigations.

It should be noted that this is likely to be a minimum level of service at present. Services noted upward trends in patient referrals and it is likely that this trend will continue because of increasing awareness, further development of diagnostic and treatment options and systematic identification of family members also at risk.

**World Class Commissioning competences**

Commissioners should work towards the following World Class Commissioning competences to provide key benefits for patients:

**Competency 2:** Work with community partners by:

- Ensuring good communication and liaison with other support services including social services, disability support services, employment services, housing, careers advice and the voluntary sector.

To provide **enhanced long term management** of patients and families with good patient follow-up and robust links with relevant health and other services.

**Competency 3:** Engage with public and patients by:

- Involving patients in service planning
- Developing effective methods for two-way communication between professionals and patients
- Developing close working arrangements with supporting voluntary organisations to provide support for patients and families

To provide **improved overall quality of life** for patient and family by the provision of accurate and trusted information on prognosis in order to support life decisions that maintain social and psychological well being.
Competency 4: Collaborate with clinicians by:

- Commissioning multidisciplinary teams including geneticist, ophthalmologist, genetic counsellor, laboratory scientist, electrophysiologist and specialist nurse
- Developing services which are integrated with other components of patient care including other specialist services (particularly important where the disease is multi-system) and with primary and secondary care elements
- Providing training for staff to achieve relevant competencies
- Developing effective referral protocols and shared care arrangements.

To provide:

- Increased patient satisfaction and trust that they will be managed by a team with experience in their particular condition
- Improved clinical outcomes by provision of the most effective preventive and treatment options to slow disease progression in the eye and (through knowledge and appropriate liaison with other specialties) in other organ systems involved
- More efficient and effective care by the formalised coordination of expertise from the two main specialities involved (ophthalmology and genetics)

Enhancing reproductive choice for parents by providing information on recurrence risk of conditions for future children and providing counselling on reproductive options

Competency 5: Manage knowledge and assess needs by:

- Recognising the need to balance specialisation with reasonable geographic accessibility
- Audit and research of service activity.

To provide reduced inequalities by the development of services that are accessible to all geographic areas and outreach, through general ophthalmology services, to all patient groups.

Competency 6: Prioritise investment by:

- Developing systems of decision making for the use, interpretation and prioritisation of genetic testing.

To ensure efficient and effective use of genetic testing.

Competency 8: Promote improvement and innovation by:

- Provision of professional education and training including awareness-raising in primary and secondary care.

To provide efficient referral of patients to the most appropriate specialty in a timely manner.
Competency 10: Manage the local health system by:

- Ensuring good communication and transitional arrangements between paediatric and adult care
- Developing systems for long term follow up including systems for initiating and undertaking family cascade testing and family record-keeping.

To provide continuity of care for all family members at risk or affected by an inherited eye condition.

Further information


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Annex 1

Main inherited conditions with significant ocular manifestations
(derived from Genetic Ophthalmology in Focus Table 2.4)

Anterior segment disorders
- Aniridia
- Axenfeld-Rieger
- Anterior segment dysgenesis
- Juvenile open angle glaucoma
- Nail-Patella Syndrome
- Peters anomaly
- Primary congenital Glaucoma
- WAGR syndrome

Corneal Diseases
- Granular lattice, Bowman corneal dystrophies
- Cystinosis
- Fabry's Disease
- Mucopolysaccharidosis

Disorders of the lens
- Alport syndrome
- Chondrodysplasia Punctata
- Congenital Cataract
- Galactosaemia
- Homocystinuria
- Lowe Syndrome
- Marfan Syndrome
- Myotonic Dystrophy

Vitreoretinal disorders including:
- Incontinentia Pigmenti
- Kniest Syndrome
- Norrie Disease
- Retinoschisis, X-linked, Juvenile
- Stickler Syndrome
- Familial exudative retinopathy

Retinal dystrophies and degenerations
- Achromatopsia
- Best disease
- Choroideremia
- Doyne honeycomb dystrophy
- Leber congenital amaurosis
- Retinitis Pigmentosa
- AR- retinitis pigmentosa
- AD- retinitis pigmentosa
- X-linked-retinitis pigmentosa
- Cone and cone-rod dystrophies
- Stargardt disease
- Sorsby fundus dystrophy
- Gyrate Atrophy of Choroid and Retina
- Alagille Syndrome
- Alstrom Syndrome
- Bardet-Biedl Syndrome
- Ceroid Lipofuscinosis (Batten disease)
- Cohen Syndrome
- Cockayne Syndrome
- Gangliosidosis
- Gaucher disease
- Kearns- Sayre
- NARP (neuropathy, ataxia, RP)
- Niemann- Pick disease
- Pseudoxanthoma Elasticum
- Refsum Disease
- Usher Syndrome

Optic nerve
- Leber Hereditary Optic Neuropathy
- Optic Atrophy, type 1
- Optic Atrophy, type 3
- Renal-coloboma syndrome
- Wolfram Syndrome (DIDMOAD)

Defects of pigmentation
- Oculocutaneous Albinism
- Ocular albinism
- Chediak Higashi
- Hermansky-Pudlak

Conditions associated with increased risk of malignancy
- Retinoblastoma
- Neurofibromatosis1
- Neurofibromatosis2
- Tuberous Sclerosis
- Von Hippel Lindau Disease

Miscellaneous
- Microphthalmia/anophthalmia, coloboma