The ROYAL COLLEGE of OPHTHALMOLOGISTS

Ophthalmic Services Guidance

Genomics Services

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1 Summary

The NHS in England is undergoing a radical change in the structure and breadth of genomic services. The drive towards developing a national ***Genomic Medicine Service*** will facilitate the delivery of personalised approaches to diagnosis and treatment for patients across the UK. It is anticipated that similar changes will apply to the devolved nations of Scotland, Wales and Northern Ireland.

Overall, these innovative changes represent an exciting opportunity to embed personalised approaches to diagnosis and treatment of ophthalmic disease, at the heart of clinical practice. Ophthalmology is well placed to take advantage of this given the preponderance of genetic advances in our specialty

Ensuring clinicians, researchers and those who influence funding streams within ophthalmology embrace genomic medicine is key to capitalising on all the benefits from this new branch of medicine.

This document is to keep members informed of developments with the UK genomics services. However, this is a very fast moving area with rapid developments driven at the national level. The information in this document is correct as at February 2020.

2 Introduction

The NHS in England is undergoing a radical change in the structure and breadth of genetic services. The drive towards developing a national ***Genomic Medicine Service*** will in turn facilitate the delivery of personalised approaches to diagnosis and treatment for patients across the UK. The evolving infrastructure developed to support this encompasses seven regional Genomic Laboratory Hubs (GLHs) and Genomic Medicine Centres (GMCs), with a comprehensive genomic test directory and concurrent development of education and training for clinical and scientific staff.

These innovative changes represent an exciting opportunity to embed personalised approaches to diagnosis and treatment, which lie at the heart of clinical medicine. Together, with the NHS Five Year Forward View embracing a technologically advanced and data driven approach to healthcare, the opportunity to improve health, reduced healthcare costs and accelerate research will be realised.

The implementation of genomic medicine is possible due to the development of powerful new DNA sequencing technologies. It is widely acknowledged that genomic medicine has the potential to bring enormous potential benefits to clinical care delivering personalised medicine for patients. This was underlined by the 2016-17 Chief Medical Officer's report Generation genome (published July 2017), which aligns to the Life Sciences sector report. Following the success of the 100,000 Genomes Programme, the development of a national Genomic Medicine Service by NHS England will allow a wider delivery of genomic medicine making it available to all patients throughout the UK benefitting the entire population.

As with any medical advance, it is important that the implementation process is carefully managed, as part of a planned programme so that patient safety and clinical benefit are ensured. This document aims to describe the opportunities and challenges for ophthalmology posed by this innovation in healthcare delivery.

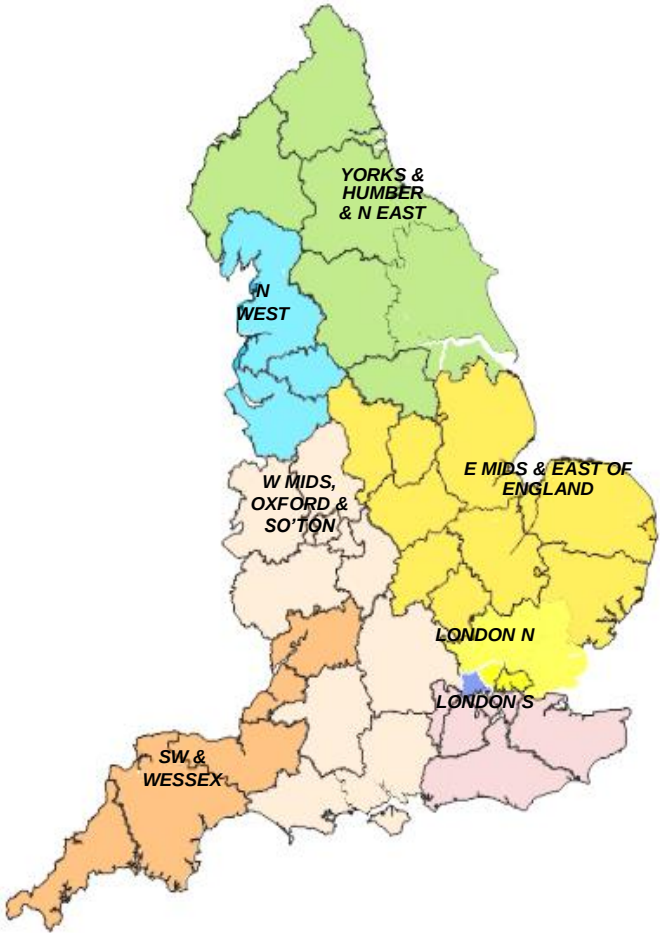
3 What's New?

3.1 Changes to the Genomic Medicine Service in England

There are three key changes to the system providing genomic testing.

- Genomic Laboratory Hubs
- A Genomic Test Directory
- Centralised funding for genomic tests

Genomic Laboratory Hubs mapped to NHS STPs
(Sustainability & Transformation Partnerships)



Driven by technological change, the NHS has moved towards a smaller number of larger genomic technology centres, termed ***Genomic Laboratory Hubs (GLHs)***, designated in the second half of 2018. It is intended these will provide standardised approaches to genomic testing and that all data derived from testing will be shared amongst the centres to maximise clinical benefit.

This marks a shift from a regionally based system (with a degree of inter-laboratory competition that had developed because of the internal NHS market) to an integrated service based upon data sharing with a more cost-effective and collaborative way of working.

Each of the seven GLHs (see map) is responsible for the provision of genomic medicine testing services across a defined geographical region. All core testing for rare disease and cancer services will be provided within this region. Cancer genomic testing, in particular delivering testing of tumours for somatic genetic changes that determine chemotherapeutic prescribing choices, represents a very significant proportion of the work undertaken within the genomic laboratories. Rare disease testing is broadly divided into core and specialist testing. Core tests include those provided for developmental and paediatric disorders and will be provided by all centres.

3.2 Specialist testing including ophthalmology

Specialist testing is designed to meet the needs of mainstream clinical specialties (see table 1) including cardiology, immunology, endocrinology and ophthalmology.

Table 1

RD Specialist Test Groups
Cardiology
Deafness
Endocrinology
Eyes
Gastrohepatology
Haematology
Immunology
Inherited cancer
Metabolic
Musculoskeletal
Neurology
Renal
Respiratory
Skin

Three laboratories (in the North East West London, Wessex/West Midlands & North West regions) will provide all ophthalmic tests across England. All three laboratories will provide genomic testing for the specialty and each hospital in England will order testing via one of these three laboratories.

In order to clarify the nature of the testing within each specialty a Genomic Test Directory has been defined and will be updated on an annual basis.

The Genomic Test Directory defines a series of clinical indications for which genomic tests are available within the NHS genomic testing system (i.e. free at the point of access). These are broad and include:

- Developmental disorders (anophthalmia/microphthalmia/coloboma; anterior segment dysgenesis and developmental glaucoma)
- Congenital cataract and childhood lens disorders
- Corneal disorders including corneal dystrophies
- Inherited retinal disorders including retinal dystrophies and vitreoretinal disorders

A range of highly specialist tests previously provided on a national basis including national services for Stickler & Bardet-Biedl syndromes and Retinoblastoma are also included in this testing schedule.

3.3 Changes to the funding structure.

There has previously been considerable variability and difficulty in obtaining molecular testing for genetic diseases across the UK. Prior to 2018, UK genetic testing budgets were not held in ophthalmology, making it challenging for eye departments to order genetic testing. The result was significant geographical variation in obtaining testing, and a failure to seek testing in some areas, within the context of an NHS system.

From 2020, the presence of centralised funding means any patient, for whom a test is indicated as per the genomic test registry, will be eligible for testing. The clinician requesting the testing will not need approval from a local budget, as the unit will ***not be asked to pay*** for the test. This will be paid centrally by NHSE with subsequent reimbursement to the testing laboratory upon delivery of a test report. As a result, molecular genetic testing should be both possible and affordable. This creates a genomic medicine system that offers equity of provision for genetic testing regarding conditions listed in the Genomic Test Directory. However, it is important ophthalmologists with an interest in inherited ocular disorders along with local geneticists are gatekeepers for the testing otherwise the number of unnecessary tests will go up.

3.4 Opportunities that result from implementation of genomic medicine approaches.

Prevention of disease

One of the largest workloads of the new genomic medicine service will be testing and identification of patients with genomic variants that predispose to cancer susceptibility in order to offer them primary prevention strategies e.g. chemoprevention or risk reducing surgery. Outside of retinoblastoma and ocular melanoma, this is likely to have limited impact upon the speciality of ophthalmology in the short term.

Early diagnosis

Rare diseases: Defined as those that affect fewer than 1 in 2,000 individuals in the general population (https://www.raredisease.org.uk/what-is-a-rare-disease/). Collectively common, patients with rare diseases are seen by clinicians in all areas of medicine. Diagnosis is often based on ascertaining accurate phenotype /clinical findings and assessing family history. Obtaining a diagnosis for these conditions has traditionally resulted in multiple sequential diagnostic investigations and referrals, often with delays in arriving at a definitive diagnosis, sometimes by many years. The addition of genomic tests has proved beneficial in accelerating the provision of a clear diagnosis for many patients in this category.

Common diseases, including cancers: Genotyping and genome sequencing can provide access to genomic risk profiles that are of value in common disorders. Outside of ophthalmology, this will include non-malignant diseases such as hereditary cardiomyopathy, and cancer. Within ophthalmology, it is plausible to suggest that this might – in the future - be relevant to age-related macular degeneration and glaucoma, particularly when positive clinical utility can be demonstrated. In general, polygenic risk scores, when added to other risk factors, can help to provide information regarding possible surveillance or treatment options. In the future, it is highly likely that genomics will be applicable to a number of common ophthalmic diseases and a wide range of clinical staff will need to understand the relevance of such tests. In addition, there are dominant and rare genetics variants in common diseases such as glaucoma and age related macular degeneration where genetic

testing may be relevant to families and cascade genetic testing would be warranted for earlier diagnosis in related family members.

Accurate Diagnosis

Providing an accurate diagnosis is the cornerstone of safe medical practice. In the context of modern clinical practice, diagnostic efficacy may be enhanced by discerning a precise molecular signature (genotype) that explains a given constellation of clinical features (phenotype). This principle applies to all diseases, both rare and common, including cancer phenotypes.

Over the past five years, since genomic sequencing has matured and been positioned for large-scale roll-out, a ***genomic model of clinical diagnosis*** is possible. With the advent of a broader genomic medical service, this will widen access of genomic testing to general clinicians (i.e. those who do not have training in genomics). Where such genomic investigations have already been introduced, e.g. for many rare conditions and for some tumour types, they have altered diagnostic algorithms and added considerably to the portfolio of clinical and investigative approaches. The result is that genomic testing is now possible ***early in the patient pathway*** – which can address uncertainty, improve management decision-making, permit early treatment and improve prognosis.

Pharmocogenomics

Due to different genetic variants, drug metabolism enzyme activity shows considerable individual variation. Furthermore, idiosyncratic drug reactions may also have a genetic predisposition. Knowledge of which drugs, many of which are in common usage, may be subject to such variation (e.g. warfarin, azathioprine etc) and which genomic tests are required might prevent inappropriate and unnecessary prescribing and enable tailoring of appropriate personalised dosage.

All those who prescribe medications should know which drugs are susceptible to genetic variation and which tests should be applied.

Novel Therapies

Within ophthalmology, genomic testing will have the greatest initial impact upon those with rare diseases, in which a diagnosis will identify those who will benefit most from gene-directed management and treatment strategies.

This includes identification of patients:

- At risk of systemic complications (Case study #1)
- With a rare disease for which a conventional therapy or dietary modification can alter disease course (Case study #2).
- Who may benefit from novel gene-directed therapeutics such as gene therapy and small molecule drug therapy? Although novel, these therapies are moving into clinical practice. Patients eligible for such treatments will be treated both within the NHS and within research-based clinical trials.

Case Study #1 Leber congenital amaurosis

Jim was born with poor vision and was diagnosed with LCA (See right for fundus picture). Genomic testing became available on the NHS when he was 8 years old. This demonstrated that he had a mutation in *IQCB1*, a gene that when faulty, causes a form of LCA that is often complicated by renal disease and progressive renal failure.

At that stage, he was referred to the renal physicians who found that his kidneys were scarred; their function was severely compromised. He was going to require renal transplantation, which has a much better long-term outcome when undertaken prior to onset of acute renal failure.

Within 12 months, he had received a renal transplant from his mother. Here we can see that Ben received appropriate and timely care enabled by the disease stratification provided by his genomic testing. Importantly Ben's sister also has LCA and has poorly functioning kidneys - she is being closely monitored by the paediatric renal service.

Case Study #2 Cerebrotendinous xanthomatosis

Lisa is a 9-year-old girl who is well, has no previous medical history and takes no medications. She reported blurring of her vision, which had progressed over the previous year (no previous ophthalmic examinations) and was found to have mixed layer bilateral cataracts, visual acuities of 0.6 logMAR in either eye and an otherwise normal ocular examination.

At the time of cataract surgery, a congenital cataract gene sequencing panel was sent, which identified a homozygous, known disease-causing variant in the *CYP27A1* gene suggesting a diagnosis of autosomal recessive Cerebrotendinous Xanthomatosis (CTX).

Subsequent segregation in her parents and cholestanol studies confirm the diagnosis, which triggered a prompt referral to the regional metabolic service.

CTX is a multisystem disorder and oral bile acid replacement therapy can halt disease progression and prevent symptoms entirely in asymptomatic individuals. Therefore, early diagnosis and treatment is extremely important in order to prevent the significant systemic complications of this condition including seizures, ataxia, dementia, cholestatic liver disease and cardiovascular disease. In this case, ophthalmic panel testing, in a girl with a seemingly isolated ophthalmic disorder, was key to this patient's life-long medical care and provided targeted screening for family members.

4 The need for change

Genomic - as opposed to genetic - testing (i.e. testing multiple genes at the same time) has rapidly improved diagnostic rates over recent years in ophthalmology. For example, diagnostic rates for inherited retinal diseases have jumped from 13% to 60% and for congenital cataract from 3% to over 70%. The use of genomic testing early in the diagnostic pathway is becoming increasingly utilised.

Patients with rare diseases are becoming more aware of the power of genomics to help them find a diagnosis. As demonstrated by the case studies above and in line with the drive towards a national Genomic Medicine Service, it is important that testing is offered to anyone who is eligible and would benefit.

5 Priorities for gene testing

It is likely that genomic testing will affect certain ophthalmic subspecialties earlier than others. These include paediatric ophthalmology, medical retina and neuro-ophthalmology. Furthermore, not everyone with a genetic disease will benefit from diagnostic testing to the same degree and, at least initially, there may be limitations on testing resources. Consequently, it is important to understand the drivers for testing and to prioritise those who would most benefit from testing – i.e. tests with the highest clinical utility.

For example:

Condition	Reasons for Testing
Retinoblastoma	Identification of familial forms Support for tumour surveillance
Bilateral congenital cataract	Exclusion of syndromic subforms Identification of metabolic forms that are treatable
Anophthalmia, Microphthalmia and Coloboma	Over 60% are associated with syndromic features, key for assembling multidisciplinary team
Aniridia	In children to rule out WAGR and risk of Wilm's tumour
Inherited retinal dystrophy	Identification of syndromic subforms
Early onset retinal dystrophy / Leber congenital amaurosis Bardet-Biedl syndrome,	Identification of mutations in cilopathy genes for renal screening

Stargardt disease	Define prognosis, eligibility for treatment trials
X-linked retinitis pigmentosa, Choroideremia	Family counselling Identify patients for treatment trials
Lens subluxation	Identify Marfan syndrome patients
Stickler syndrome	Family screening and prophylactic treatment
Early onset Nystagmus	Early diagnosis
Albinism	Early diagnosis Identify patients with Hemansky Pudlak 7 Chediak Higashi syndromes for treatment
X-linked Retinoschisis	Testing RS1 in family members to exclude need to screen peripheral retina in unaffected
Von Hippel Lindau	Family screening to avoid need for intensive monitoring /retinal screening

6 Implication of genomic medicine for ophthalmologists, ophthalmology services and generalists

6.1 Ophthalmologists

General

For ophthalmology, it is likely in the short term that the greatest impact of the changes in genomic medicine will be for patients with rare disease. While all ophthalmologists see patients with rare disorders from time to time, these are generally managed within subspecialist clinical services and this situation is likely to continue. However, knowledge of the new systems, to ensure that there is delay in referral and access will be critical.

In the longer term, it is likely that genomic medicine approaches will impact upon common disease and pharmacogenetics. For this reason, it will become increasingly important for clinicians to understand the capabilities and limitations of genomic approaches.

Paediatric Ophthalmology and Medical Retina and Neuro-Ophthalmology

A wide range of the disorders seen within paediatric ophthalmology, such as developmental eye disorders, congenital cataract, paediatric glaucoma, inherited retinal disease, nystagmus and albinism, have a genetic basis or represent part of a differential diagnosis list that encompasses inherited disorders. The same is true for medical retina services, where a wide a range of conditions are managed including inherited retinal disease such as macular

dystrophies, early onset macular disorders, rod-cone and cone rod dystrophies and neuro-ophthalmology for example inherited optic neuropathies, chronic progressive external ophthalmoplegia (CPEO), optic nerve hypoplasia.

Therefore, these ophthalmic subspecialties will have a greater need to develop models of working with the new Genomic Medicine Service. This may include ordering of genomic tests; working within multidisciplinary teams (MDTs) to facilitate genomic variant interpretation; as well as working with genomic counsellors and nurses to deliver genomic results to patients. With time this will lead to changes in care pathways for many conditions

It remains unusual for ophthalmologists currently working within these existing subspecialties to have the full range of expertise across these rare genetic disorders. Developing a workforce for these clinical areas with adequate expertise will need a comprehensive training programme across the UK.

Genetics and genomics specialists

There are a number of different models of care provision at present (http://www.phgfoundation.org/report/genetic-ophthalmology-in-focus). These include ophthalmologists who have specialised in inherited retina/optic nerve disease providing these clinics with input from either a genetics counsellor or a clinical geneticist; or genetics clinics where a clinical diagnosis has been made already by an ophthalmologist and the patient has been referred to a clinical genetics clinic; or a mixed model. These models vary throughout the UK, depending on the ocular condition, the breadth of expertise in the centre and whether more complex syndromic or dysmorphic features are present. Patients with specific conditions may be referred to centres specialising in that condition for example, Bardet Biedl, or mitochondrial disorders.

In time, with appropriate education and training interventions for both ophthalmology trainees and established ophthalmologists, delivery of genomic medicine providing clear benefits for patients will be realisable for all patients, wherever they live in the UK. However, training and education will need to be expedited, with appropriate resources to ensure this happens, as currently there is a limited number of specialists in this rapidly developing field. Genomic ophthalmology specialists will be required in all regions as not all cases will be straightforward. Here, a network will continue to exist both to take referrals and to provide advice to more mainstream clinicians.

6.2 Implications for service delivery models and quality standards.

Although all ophthalmologists will need to be upskilled in understanding genetics and genomics, how patients are identified, investigated and managed, there will be a hierarchy of levels of expertise, as discussed above. Here, the establishment of infrastructure with associated teaching and training will improve engagement and delivery of the NHS genomics strategy.

In developing a new service model, it will be critical to identify and mitigate potential risks and to develop safe networks of care that are adequately overseen by those with high genomics expertise and strong links to research and testing centres whilst ensuring wide access through many more general units. This may be similar to what has already been established in other areas for delivering specialised ophthalmology services. This will involve developing care pathways, protocols for local delivery of appropriate aspects and clear guidance on when to pass on up the network chain, teaching and training and open, clear channels of communication.

In addition, local services will need to consider how they upskill and utilise their non-medical healthcare professional staff such as nurses, orthoptists and optometrists. This is especially important now as the national initiative is to expand their roles into advanced practice (delivering care traditionally delivered by doctors). Units may also need to consider whether they need access to genetic counsellors or can develop their own staff to deliver some aspects of a genomic service as more care and testing is delivered locally.

At noted above there are a small number of highly specialised services delivered in specific centres that focus on the treatment and management of rare, highly complex genetic disorders. These include neurofibromatosis, Stickler and Bardet-Biedl syndromes, retinoblastoma, and lysosomal storage disorders. While all such services evolve with time, there are no current plans for the changes in genomic medicine to alter these services.

Finally, there will be a need to develop quality standards and agreement on audit and metrics for activity and success and safety for these services both locally and across the network. Staff compliance with training and competencies, CPD, appropriate diagnostic equipment and expertise, appropriate access to more specialist centres will need to be addressed. Quality assurance regarding patient and testing volumes, appropriateness of testing, patient experience, accuracy of interpretation will need to be assessed to ensure that futures services are fit for purpose and delivers on the objectives of the National Genomics Medicine Service.

7 Consent issues

Delivery of genomic data has broad implications, and testing requires a discussion that examines the risks/benefits and allows patients to make informed decisions. To support the implementation of genomic testing within the Genomic Medicine Service, a new patient choice (consent) model is being developed. Key aspects of this model are to set clear and informed choice about use of NHS GMS and Genomic Tests to patients. Information resources will be provided to help patients understand the choice and consequences. Consent will cover both usual use of patient data within healthcare services (e.g. clinical and administrative standards), as well as innovative aspect of genomic care and data use to support/develop services. Importantly this will mean that many patients offered whole genome sequencing will have an opportunity to participate in research. Thus, within the consent a clear and distinct choice will be offered to the patient to be part of a research programme, without impact on their standard clinical care

A detailed series of workshops and discussion, involving the Medical Royal Colleges (including RCOphth), The British Society for Human Genetics, Patient Groups and NHS England has developed a series of consent documents as well as a ‘Record of Discussion’ that can be used as the basis for recording such patient choice.

Moving forwards there will need to be a process of update and education in the processes required for ordering of genomic tests including consent issues. Initially this will need to include all those involved in the management of patients with rare disease. As the indications for genomic testing broaden, this will encompass increasing numbers of clinicians.

8 Data and IT

It is increasingly recognised a data-driven approach to medicine will help deliver a personalised approach to care, enhance efficiency and improve outcomes for patients. One major advantage of this will be the integration of large datasets such as patient information, including phenotypic, clinical and imaging together with genomic data.

Enabling access to integrated patient data is expected to drive forward a systematic adoption of Electronic Patient Records.

It is likely that the large datasets such as personal genomic data will be an important testing ground for such initiatives. It will be important for ophthalmology to ensure that ophthalmic-specific EPR packages are fit for purpose with appropriate safeguards to embrace genomic medicine.

9 Training and education

Training in genomic medicine will need to encompass all levels of training within medicine /ophthalmology. How this is best approached has been extensively discussed by stakeholders, HEE and by the Academy of Medical Sciences (see table 2).

Table 2

	Undergraduate	Postgraduate	Workforce
Stage/ responsibility	MBBS GMC University	Colleges JC Deanery	Colleges NHS Trust
Objectives	Embed in curriculum Futureproof	General and specialist training	Skills essential for service delivery
Barriers	Long lead-in	Competing with other key areas Different Specialty needs	Competing service needs
Existing initiatives	Intercalated degrees	Genomics champions/SACs HEE MOOC MSc Oncology	Genomics Champions College president's meeting Update study days/seminars/online learning including relevant examples HEE MSc
Plans	Consistency Learning outcomes Liaison postgraduate providers	Learning outcomes Dual accreditation Expert input into curriculum Assessment	Prioritise frontline On-line signposting Certification Educational library RCP Fellow

9.1 Medical school curricula

The genetics framework of six learning objectives (below) was created for UK medical students and endorsed by the Joint Committee of Medical Genetics in October 2006. The framework was reviewed for genomics by the National Genome Education Centre (now Genomics Education) in August 2013.

Learning objectives

- Understand and describe the structure and function of the human genome, and how it is transmitted
- Understanding of how genome variation arises and its role in health and disease
- Be aware of how information from the genome is obtained and how it can be used to identify the genetic variation associated with disease and to inform clinical management
- Be able to identify patients and families with, or at increased risk of, a condition with a strong genetic component
- Be able to communicate genetic information in an understandable, non-directive manner, being aware of its potential impact on an individual, family and society
- Know how to obtain up-to-date information about scientific and clinical applications of genomics

9.2 Professional curricula for the RCOphth

A long-term objective will be to work with the Training Committee, in conjunction with other Royal Colleges, to develop an appropriate curriculum to reflect on-going changes to clinical practice that result from the development of mainstream genomic medicine.

9.3 CPD and up-skilling of the current work force

While the move towards adoption of genomic medicine is imminent, it is likely that the speed of development of skills will differ between subspecialty groups, and objectives will be different for subgroups of clinical ophthalmology.

Potential ophthalmic geneticists. It will be important to develop a small number of fellowships in this area to ensure ongoing training of clinicians following a path towards delivery of genomic medicine. It will be important to have clarity on where such training should be provided, and how to set up accreditation of training centres. These could form part of medical retina, medical ophthalmology, or paediatric ophthalmology TSC.

Sub-specialities where genomic medicine already plays an important role including paediatric ophthalmology and medical retina. Ensuring that individuals training in these areas can develop sufficient skills to work in genomic ophthalmology, alongside clinical genetics will be an important priority.

General ophthalmology There are likely to be different levels of skill required in different circumstances but a general understanding of clinical pathways, consent as well as applications of genomic medicine including pharmacogenetics will be increasingly important to all clinicians. Courses will be made available to all ophthalmologists at all stages of their career to gain knowledge and CPD in this area, extensive free courses already exist: https://ukegg.com/education/

10 Research

Ophthalmology has been at the forefront of the discovery era of genetics, as is reflected in the numerous single genes available for genetic testing. Such discoveries have included rare and common disorders, and clinical trials for aniridia, retinal and optic nerve disorders

The move towards genomic medicine has been catalysed, in part, by the research agendas of the NIHR as well as Genomics England through the 100,000 genomes programme. This is seen as a strong demonstration of the success of the biomedical research agenda as well as promoting the health and wealth agenda that is reflected in the Government's Life Sciences Sector Report.

12 Impact of changes in Devolved Nations.

Northern Ireland

Miss Giuliana Silvestri and Dr Shane McKee have commented that it is planned that genomic and genetic tests that are currently sent to English laboratories (apart from Whole Genome Sequencing) will be repatriated into a NGS platform based in Belfast, although some tests (e.g. mitochondrial) will continue to be accessed via the specialised labs.

It is expected that the system in Northern Ireland will closely mirror the National Genomic Medicine Service for England.

This is intended to ensure that NI patients have equity of access to genetic and genomic tests.

On the informatics side, one key learning point from the 100,000 Genomes is that the collation of detailed phenotypic information is critical for the analysis and prioritisation of variants detected in genomic sequencing. Geneticists in Northern Ireland are working with colleagues in North Thames and West Midlands to put in place a phenotyping platform based on GEMS and OpenEHR, and this will be made available to ophthalmologists to allow ordering of genetic tests and subsequent analysis of variants. A key element (via the OpenEHR) is to interface with hospital systems to automatically gather as much data as possible/relevant in an open-standards format that allows for many types of downstream analysis (apps, portals, other specialties etc).

Scotland

Dr Carol Gardiner has kindly reviewed and made comments on this document.

Wales

Miss Marcela Votruba has communicated that Wales will have access through a combination of local testing and referral as required. The All Wales Genetic Testing Service is leading this.

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