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Genomics in Medicine

Delivering genomics through clinical practice

Report of the Joint Committee on Medical Genetics

June 2012

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Foreword

Sir Richard Thompson

President, Royal College of Physicians

The physician's role at the forefront of medical diagnosis requires the highest standards of medical professionalism. The Royal College of Physicians, with its commitment to improving the quality of care for patients and to educating doctors, plays a key role in ensuring that the best evidence-based medicine is practised in the UK and beyond.

As this report, and the recent report of the Human Genomics Strategy Group (HGSG), make abundantly clear, the processes of diagnosis and decisions about clinical management are increasingly aided by genomic tests – whether this is in the context of diseases with a heritable component, the DNA profile of tumours in patients with cancer, or a host of other applications.

The UK is a world leader in genomic science and clinical research, but too often the translation and application of such knowledge into routine clinical practice does not keep pace. Centres of research excellence forge ahead, but there is a risk of mismatch between the services provided in those centres and the needs of the population at large.

The meeting held at the RCP in June 2011 involving some fifteen specialties examined the challenges of integrating genomic medicine across a wide range of clinical practice. It confirmed the expectation that, although genomics is poised to become a vital constituent of many areas of clinical medicine, medical professionals in the services are ill-prepared to take advantage of this to improve the precision and quality of care for their patients. The education of physicians and other health professionals to fully use genomics in their practice, whether as a generalist or a sub-specialist, is of prime importance, together with the development of standards for high quality care that optimise the use of genomics in patient pathways. I do not underestimate the difficulties for practising physicians to keep up with the rapid advances.

Genomic medicine is here to stay, and the UK medical system has a real opportunity to lead the world in the provision of modern medicine. But action must be taken now. The Royal College of Physicians is fully committed to playing a lead role in this process. A first step is for all specialty societies and their education and training sub-committees to consider the issues identified in this report. Further consideration should be given to developing a strategy to ensure that high quality care pathways, as advocated both in this document and the HGSG report, can be delivered equitably across the NHS. Physicians must learn to work closely with their colleagues in clinical and laboratory genetics, but it is they themselves who will have to apply this expanding knowledge every day.

I commend the Joint Committee on Medical Genetics, the PHG Foundation, the UK Genetic Testing Network and the National Genetics Education and Development Centre for this timely report, and thank those many individuals who took time to attend this ground-breaking meeting.

Sir Richard Thompson

President, Royal College of Physicians

Contents

1 Introduction

During the last two decades developments in the science of genetics and enormous advances in genetic technologies have altered the capability to understand diseases, make diagnoses and provide effective management.

For clinical researchers and those in the relatively small discipline of clinical genetics at the forefront of management of people with inherited disease, such progress might be said to be transformative. What is less clear is the impact that genetics and genomics has had, and will shortly have, on the practice of medicine throughout other areas of clinical practice, and how services can develop to take best advantage to improve health outcomes.

This report has its origins in work presented to the Joint Committee of Medical Genetics in 2011 in which the service implications of genetics within the specialties of cardiology and ophthalmology were considered in detail and some outline conclusions on the strategic implications for clinical services as a whole were drawn. The Royal College of Physicians (RCP) responded by asking the Joint Committee on Medical Genetics to hold a workshop including a wide range of other clinical specialties to reflect on these findings within their own service area, to consider the wider health service environment and to make recommendations for action. This report includes findings from the workshop held in June 2011 and provides key recommendations for the College.

2 Background

The future vision of genomics in healthcare and public health represents a confluence of development of three important strands: genetic technologies, clinical genetics and genomic healthcare.

A fuller discussion of these terms is included in Appendix 1. Broadly, *genetic technologies* encompass the whole range of laboratory technologies that provide detailed sequence and other information on genomes – whether related to an individual's germline, somatic cells such as the altered genome within cancer cells, or non-human genomes such as those of bacteria or parasites. The explosion of potential applications in healthcare arises from increased speed and decreasing cost of sequencing along with an understanding of the clinical relevance of emerging information for patient management.

Clinical genetics is the specialty that provides services for individuals and families affected by, or at risk of, a genetic disorder or congenital abnormality. It includes diagnostic assessment, counselling and support, genetic testing, and provision of advice to patients and the extended family. Traditionally it has encompassed chromosomal disorders, dysmorphic syndromes, teratogenic disorders and single gene disorders which may be evident in childhood or later in life. The challenge that arises for clinical genetics is that many inherited disorders, including a large number of single gene disorders, manifest as patients presenting to a wide range of clinical specialties, meaning that clinicians in these specialties need to be skilled in recognising, diagnosing and managing these conditions.

Genomic healthcare widens the range of applications of genomic technologies to include instances where they may be used to recognise a precise molecular sub-type of disease and hence fine-tune treatment, to determine disease susceptibility (where a pattern of genetic variants may be assessed alongside environmental factors) and provide stratified advice on prevention, or to understand the likely response to therapeutic interventions and personalise treatment. Genomic healthcare is thus the kernel of the expected revolution often termed *personalised* or *stratified* medicine.

Clinical genetics and genomic healthcare are thus by no means mutually exclusive. There are clear areas of overlap: for example when variable presentation of a single gene disorder may arise due to the interaction of multiple genetic or environmental modifiers; or when the clinician within a specialty such as cancer or cardiology may seek rare single gene subsets of disease such as breast cancer due to BRCA1/2 mutations in order to decide on the best treatment options. The anticipated expansion of such areas of overlap and the emerging complexity of the genetic basis of both classic 'inherited' disorders and common complex disorders provide the very basis for the assertion that genomic healthcare must be developed and that current clinical genetic services will provide an important foundation to such development.

New genetic technologies, clinical genetics and the advent of genomic medicine are set to have a major impact on health care, patient outcomes and the health of the entire population.

Taken together, new genetic technologies, clinical genetics and the advent of genomic medicine are set to have a major impact on healthcare, patient outcomes and the health of the entire population. In particular, by bringing the ability to better stratify population cohorts and provide more personalised medicine, genomic medicine complements each of the five domains of the NHS Outcomes Framework.

3 The RCP workshop

The workshop held at the Royal College of Physicians in London in June 2011 built on three reports, published between 2008 and 2011, that considered in detail the development of genetics within mainstream clinical services.

Reports analysing emerging health needs arising with regard to genetics in cardiovascular medicine, *Heart to Heart*[1], and ophthalmology, *Genetic Ophthalmology in Focus*[2], concluded in each case that there was a pressing need for expansion in capacity and capability in genetics both as a sub-specialist area and integrated across the entire specialty. A final report, *Genetics in Mainstream Medicine*[3] which considers the strategic implications of these findings, concluded that it was likely that a wide range of specialties would need to develop capacity and capability in genetics, and recommended that such services should be commissioned and provided on an equitable basis across the UK. Strategic elements to achieve this include: that specialties should each adopt an active partnership with relevant stakeholders to develop the role for genomic medicine within their clinical area; the development of education and training at all levels; the development of commissioning and relevant support; and the establishment of a supporting health services research programme. The report emphasised that the clinical and laboratory genetics specialties would need to play a leading role in these developments.

The aim of the June 2011 RCP workshop was to engage a wider clinical group in this debate, to review the current positions regarding the provision of genetics in medicine across a range of clinical areas and to make recommendations about key policy areas to enable translation of genetic advances into high quality clinical services. The meeting was organised jointly by the Joint Committee on Medical Genetics (JCMG), Foundation for Genomics and Population Health (PHG Foundation), UK Genetic Testing Network (UKGTN) and NHS National Genetics Education and Development Centre (NGEDC) in conjunction with the Royal College of Physicians.

The organising bodies and the RCP through their specialist society network identified key individuals from a wide range of clinical specialties where genetics was relevant in clinical practice, together with representatives from the clinical and laboratory genetics community, patient organisations, population health and commissioning. A full list of those attending and the specialties they represented is provided in Appendix 2.

It is estimated that 3 million of the UK population has one of 6,000 rare disorders.

1 *Heart to Heart. Inherited cardiovascular conditions services.* PHG Foundation 2009

2 *Genetic ophthalmology in focus. A needs assessment and review of specialist services for genetic eye disorders*. PHG Foundation 2008

3 *Genetics and mainstream medicine. Service development and integration.* PHG Foundation 2011

The programme was highly participative. Following introductory presentations the workshop was structured around four main questions for discussion:

1. What are the existing and preferred models of service provision for inherited disease?
2. Should a service be delivered by generalists, sub-specialists or specialist geneticists? What are the training implications for models of care?
3. What are the key components of care in inherited disease?
4. How could the new commissioning climate support pathways of care?

Each section was introduced with a series of invited short presentations, followed by structured discussion to develop a series of observations. In a final session, participants reached agreement on key policy points and recommendations for concerted action. The workshop programme is included at Appendix 3.

3.1 Workshop presentations and discussions

An introductory session set the scene about service provision for inherited diseases in the UK. Professor Sir John Burn described the importance of molecular diagnosis across a number of conditions. This illustrated the range of clinicians that would need to use and interpret genetic testing in the near future.

He gave examples of the use of germline testing for the management of an inherited disorder in an extended family and testing to determine the likely utility of inhibitors of PARP enzyme (important in DNA repair) in treatment for women with breast cancer due to BRCA1 and BRCA2 mutations.

Hilary Burton provided evidence of the need for health services to evolve and adapt to meet the challenge of genomic healthcare. Challenges in providing equitable services are associated with the high prevalence and complexity of inherited disease across a wide range of clinical areas.

It is estimated that 3 million of the UK population has one of 6,000 rare disorders, (defined by the EU as affecting fewer than 5 in 10,000 of the general population[4]). Current service provision, although excellent in some specialist centres in the UK, is highly inequitable. Opportunities were arising that may drive and encourage a wider range of clinicians to integrate genetics and genomics into their practice. Increasingly treatments will be informed by the underlying molecular diagnosis, requiring clinicians to be competent in ordering and interpreting the relevant genetic tests. Such interpretation and clinical decision-making would be supported by clinical biomedical informatics. Proposals to develop this aspect of the service are currently a central component of the strategic work of the Human Genomics Strategy Group.

3.2 Current models of care for inherited conditions

Presentations were made about several successful models of care for inherited conditions, including cardiac genetic services (the CARDIGEN service based in the Northeast of England), cancer genetics services provided in the West Midlands, a specialty led neurogenetics services based at the National Hospital for Neurology and Neurosurgery (London), a tertiary endocrine genetics services in the West Midlands, the familial

4 Council Recommendation on an action in the field of rare diseases, June 2009

hypercholesterolaemia cascade testing service in Wales, and a team based at the Royal Devon and Exeter Hospital, who are exploring ways of increasing the integration of molecular genetic testing into diabetes care. Presentations described the services they had developed to provide high quality care for inherited disease, and particularly how they had tackled inequities within their own region and sought ways to cope with the demand for specialist services. The variety of these models is captured in Table 1 and set out in detail in Appendix 4.

Table 1

Inherited cardiac disease	Led by clinical genetics
Cancer genetics	Led by clinical genetics with pathways integrated into primary and secondary care
Neurogenetics	Provided from within neurology
Endocrine genetics service	Led by endocrinology with integrated clinical genetics
Familial hypercholesterolaemia	Structured multidisciplinary pathway led by lipid clinic clinicians with family cascade service hosted by regional genetics service
Single gene diabetes	Led by diabetology with network of specialist nurses

Observations

From discussions of these examples of services for inherited conditions members of the workshop observed that:

1. Rapid advances in science and technology are taking place and those centres at the forefront of research are able to take advantage of new knowledge and technology for their clinical services.
2. Several centres of excellence have developed good practice in order to deliver high quality services for inherited diseases.
3. There is considerable inequity of access to genetic testing.
4. A variety of models has been adopted to try and improve equity. These have had to be adapted for local circumstances and according to the disease groups under consideration. However, each of these remain as locally developed services and it was suggested that a more systematic approach was warranted.
5. There is currently no systematic means of translating innovations into good practice across health services as a whole. However new national commissioning arrangements may provide a window of opportunity to address this.
6. For some of the more common conditions (for example familial hypercholesterolaemia), despite evidence-based opportunities and approval by NICE, genomic innovations had not been implemented.

Effective cascade testing in families was enhanced by the involvement of genetic counsellors or specialist nurses with genetic training.

7. Effective cascade testing in families is enhanced by the involvement of genetic counsellors or specialist nurses with genetic training with a specific remit to facilitate such testing. Effective and funded examples were provided for FH, cancer genetics, endocrine and diabetes.
8. Close working relationships of mainstream medicine and regional genetics services are advantageous, particularly regarding education, up-to-date scientific information, and standards of genetic care.
9. One model may not fit all clinical needs so it will be important to tailor services to meet specific requirements of the patient pathway for a particular condition.

3.3 Service delivery: specialisation and training implications

The necessary levels of specialisation and sub-specialisation in genetics were introduced by considering examples of the roles of generalists and specialists in three different services: haematology, obstetrics and paediatrics. Table 2 provides a summary of the main findings (the full report is available at Appendix 5).

Table 2

Specialty	Relevant areas where genetics is important	Model of provision	Training implications
Haematology	Inherited bleeding conditions Thrombophilia disorders	Comprehensive care centres in haematology departments Integrated molecular genetics services in 50% of centres	Already a significant molecular component to training, so molecular medicine will be easily facilitated
Obstetrics	All areas, where previous family history of genetic condition, risk detected in screening or unexpected anomaly	Care divided between general obstetrics and fetal medicine, depending on degree of specialisation needed	All obstetricians will require general expertise, and fetal medicine sub-specialists will require more specialised knowledge
Paediatrics	Disabling chronic diseases of childhood and developmental delay	Mostly provided by generalist paediatricians and community paediatricians. Specialisation required in some areas, e.g. inherited metabolic disease	All paediatricians will require training in genetics and some will require sub-specialist training

3.4 The role of the National Genetics Education and Development Centre

Peter Farndon described the work of the National Genetics Education and Development Centre in taking forward education in genetics for health professionals in the UK. The role of hospital doctors in making a diagnosis may require that they are able to recognise a situation where genetic testing may be of use, understand how to use genetic tests, know how to interpret the results and be able to recommend treatments on the basis of the results. The required depth of knowledge underpinning these principles may vary even within specialties depending upon the individual's practice. Although core concepts in genetics have now been developed and accepted by national bodies responsible for pre- and post-registration education, the question of whether some medical specialists need more in-depth knowledge is yet to be addressed by the specialties. It will be important to gain a detailed understanding of which sub-specialty training requires genetics and genomics (helped by

definition) and what knowledge, skills and experience they will need to work in a sub-specialist capacity. A subsequent challenge will then be to identify and expand the group of those able to undertake the roles of educators.

Observations on genetics sub-specialisation and training

From this discussion on specialisation and training the workshop observed that:

1. Patients in whom genetic issues influence care are treated by all specialties.
2. A degree of sub-specialisation with regard to genetic issues already takes place in some clinical areas (*e.g.* haematology, fetal medicine).
3. In other areas of medicine, most clinicians require some genetics knowledge (*e.g.* paediatrics) and need to maintain close links with genetics to ensure new technology is used appropriately in practice.
4. All services acknowledged that they would increasingly need to use genetics competences throughout the specialty and would also require sub-specialisation for some doctors.
5. It was seen as likely that there would always be components that lie outside the expertise of the clinical specialty and that require input from clinical geneticists. However, over time more specialties would develop sub-specialist skills to deal with potentially inherited conditions and the range and balance of conditions seen by geneticists versus clinical specialists would evolve.
6. Genetics training needs to be specific for the clinical work of each specialty, building on background knowledge achieved through basic specialist training.
7. Defining and delivering the necessary education will be a major commitment.
8. Regarding genomic information, generalists will require understanding of the core principles surrounding the potential use of genomic information and specific knowledge and guidance on the way in which genomic information is used in the clinical pathways with which they are involved.
9. The particular pathway of care for a condition will determine whether genomic information for refining diagnoses and targeting treatment is best used by specialty or clinical genetics consultants.

3.5 What are the key components of pathways of care?

Presentations were given to highlight what patients and clinical geneticists considered to be key components of care for inherited conditions. These are summarised in Table 3 and described in more detail in Appendix 6.

Table 3

Patient view point	Requirement for highly specialised centres that will ensure a definite diagnosis, appropriate pathways of care, accurate information and a route-map to help them cope with the condition. Care integrated with the rest of the service and mechanisms to cascade the process through the family if appropriate.
Clinical genetics	Starts from given diagnosis or clinical problem, package of multidisciplinary management, holistic, coordinated and evidence based. Includes which professional undertakes which tasks, (*e.g.* who initiates, performs and interprets genetic testing), what test will be done, mechanism for cascade testing. Service specifications set out roles of various professionals.

An example of how the components of a pathway of care were identified was provided by Campbell Cowan, the clinical lead for arrhythmias in the NHS Improvement – Heart Programme. The initial impetus for detailed work in the area of inherited cardiac conditions arose from a high profile Private Members Bill named 'Cardiac Risk in the Young (Screening)', following which a new chapter, focusing on cardiac arrhythmias for the *National Service Framework for Coronary Heart Disease*[5] was developed. This, and subsequent detailed work undertaken on a national basis, enabled regional service providers to make substantial progress to set out pathways of care, define tertiary and secondary levels of specialism in providing care, agree levels of management for different conditions and the pathways between them, and the specialised clinical roles that would be undertaken by cardiology and clinical genetics professionals within a multidisciplinary service.

In order to secure the provision of such specialised services on an equitable basis throughout the country a multidisciplinary group including cardiology and genetics worked at a national level to develop guidance on commissioning and promoted and secured the inclusion of inherited cardiac conditions as a specialised service under the National Definitions sets (No 13: Specialised cardiology and cardiac surgery services)[6].

3.6 Commissioning in pathways of care

Presentations from Jacquie Westwood (UKGTN) and Ed Jessup (National Commissioning Group) provided a perspective on commissioning processes and the ways in which they could formalize pathways of care in order to promote equitable, holistic and high quality care across the UK.

The mechanisms for commissioning genetics and other clinical services differ throughout the UK. Commissioning in Scotland is undertaken by 14 health boards. In Wales seven health boards are now responsible for all aspects

5 *National Service Framework for Coronary Heart Disease*. Chapter 8 Arrhythmias and Sudden Cardiac Death www.dh.gov.uk

6 This substantial work should have raised the priority for inherited cardiac conditions such that a designation process would be designed and undertaken by specialised commissioners. However, progress on this has subsequently stalled.

of planning and healthcare provision and in Northern Ireland the Health and Social Care Board, advised by the Public Health Agency, commissions services for five major trusts. The simpler structures and smaller populations in the devolved administrations allow for more expeditious and equitable introduction of service developments in these parts of the United Kingdom.

In England commissioning is currently in transition with plans to transfer responsibility for commissioning to many clinical commissioning groups (number as yet to be determined) and the NHS Commissioning Board (NHSCB) to directly commission specialised services with advice from clinical networks and senates. National commissioning for specialised services provides an opportunity to reduce variability in provision of genetic services across England through these mechanisms. This level of commissioning currently represents around 0.5% of the budget. It is envisaged that this will rise to around 10% in the new reorganised structure. The NHSCB will be geographically based in four locations across England and specialised commissioning will be aligned with these arrangements. Commissioning leads are proposed for each of the 36 specialised service national definitions.

The commissioning functions of the NHS Commissioning Board and Clinical Commissioning Groups should lead to the specification and alignment of care pathways to optimise care for patients with respect to outcomes, quality and value for money. The national model service specification[7] to be adopted for clinical genetics includes: service standards, a quality assurance framework, guidance on the workforce, clinical outcomes, data standards, patient-centred standards regarding choice, information and waiting times, and commissioning contract requirements. The model sets out principles for clinical genetics and for integrated care, which explicitly consider the roles and responsibilities of clinical genetics services with respect to other specialties. In addition to diagnosis of inherited disorders and management of familial aspects of disease, clinical geneticists are a key interface with specialist clinicians to provide genetic expertise as genetic services are expanded and embedded into clinical pathways. It is envisaged that the role of clinical geneticists will evolve to one that provides leadership, expert support and mentoring and management of particular familial issues such as predictive and cascade testing and reproductive counselling as well as continuing to provide core clinical genetic for very rare disorders.

Apart from the specialty of clinical genetics itself, there are very few services in which the genetic component is directly commissioned. Rather, services for conditions such as cardiovascular disease or neurology are commissioned as a whole and the main interaction is between commissioners and the individuals leading these services.

As a result of reorganisation the workforce in specialised commissioning will be radically reduced and the remainder will have to prioritise its attentions. Specialised commissioners are using the recent UKGTN report *Review of Commissioning Arrangements for Genetic Services and Strategic Recommendations*[8] together with common service specifications to decide how commissioning will be taken forward in 2012/13. There is also a proposal to develop clinical senates and networks advising specialised commissioners. A precedent had been set for commissioning such an infrastructure by the funding of networks such as UKGTN and the infrastructure for cancer networks.

7 SSNDS Definition No. 20 Specialised Medical Genetics Services (all ages) (3rd Edition)

8 Available at www.ukgtn.nhs.uk

Patient pathways for genetic and inherited disorders seen within a particular specialty may, or may not, have a defined management specification delivered outside the clinical genetics service. It is normal practice for the treating specialty to be responsible for the diagnostics, including the genetic diagnosis of the symptomatic patient. It would be necessary to ensure that the specification included clarity on responsibility for diagnosis of family members, (for example, including cascade testing and pre-symptomatic testing in family members). Again, the recent UKGTN report noted above provides some guidance in this area.

Examples of best practice include clinical networks, multidisciplinary teams, joint clinics and GPs with a special interest working within regional genetics centres.

There are a number of models of good practice for integrated working between genetics and other specialties and these are thought to promote efficiency and maximise clinical utility. Examples include clinical networks, multidisciplinary teams, joint clinics and GPs with a special interest working within regional genetics centres. In some circumstances (for example when many body systems are involved as in Marfan syndrome) it may be difficult to decide which is the lead specialty with regard to responsibility for diagnostic tests and, later, for coordination of care. However, providers and commissioners involved with each local agreed pathway should identify, within their own setting, the most clinically effective and cost effective structure to provide a quality service to the individual and their family. This may differ between different services.

Observations

From these examples the workshop observed that:

1. Commissioning is a key mechanism for delivering equitable high quality services that incorporate genetics and genomics aspects across the UK.
2. Commissioning of specialised services integrating genomics and genetics will be enabled by expert multidisciplinary groups who develop evidence-based recommendations about the structure and function of such services.
3. Examples were provided of general genetics and specialist pathways that had been developed. Such work will need to be replicated across a number of specialties and will require national impetus and commitment of time by a wide range of stakeholders including patients, providers and commissioners.
4. During the current reorganisation of commissioning in the NHS there is an opportunity to embed good practice for inherited disease across a range of clinical areas and to integrate tertiary, secondary and primary care levels into planned pathways. It will be important to utilise the expertise and skill-mix that resides within clinical and laboratory genetics and other specialists with an interest in inherited disorders to develop guidance on the care pathway and thereby maximise their efficiency and clinical utility.

3.7 Relevance of findings to other clinical specialties

In the final session we asked other specialties present to comment on whether they thought genetics was an important feature within their specialty, whether the specialty was ready to take it up in practice and what were the key issues that needed to be tackled. Comments were provided from perspectives of bowel disease, gastroenterology, nephrology, respiratory medicine and pediatric neurology. All specialties acknowledged the importance of genetics/genomics but were at various stages of integration into the specialty. Comments received from the other specialties present are provided in Table 4 below.

Table 4

Clinical area	Comments on readiness to integrate genetics into management
Colorectal disease	There are disease specific working groups that include genetics.
Gastroenterology	Specialist centres are ready but others are not. It was recommended that there was a need for super-specialists and there should be a formalised system of provision.
Nephrology	Inherited disorders are recognised as important and there was a rare disease working group that had published a national consensus document, *Rare Kidney Diseases: An Integrated Strategy for Patients in the UK*[9]. This highlighted the need for disease specific working groups within the specialty, the development of care pathways and specific rare disease registries to inform clinical practice. Again, although there were experts in specialist centres across the breadth of the UK there was a need for nephrologists in general to develop their expertise on inherited disease. As testing begins to inform management of more common diseases there would be an even greater urgency to prepare trainees to understand genetic variance in clinical practice.
Paediatric neurology	There was already close working between paediatricians and geneticists. The example was provided of autism, where the use of genetic testing was expected to increase but not all paediatricians would feel adequately informed to use technology effectively. Furthermore, research on genotype phenotype correlation would be required to provide evidence of clinical utility in practice.

9 http://www.renal.org/whatwedo/news/10-04-26/Rare_Kidney_Diseases_An_Integrated_Strategy_for_Patients_in_the_U_K.aspx)

Respiratory medicine	The majority of clinicians in this specialty are not ready to integrate genetic aspects of disease, although in many areas of the specialty there were individuals with genetic expertise. Increasingly in the future, the integration of genomics into such areas as asthma risk and treatment choice will mean that every physician needs to be competent to use genetics within his or her own practice and also to understand personal limitations. However, it is likely that families will continue to require the skills of clinical genetics at some stage. The British Thoracic Committee would be an appropriate organisation to receive and develop recommendations on the integration of genetics into practice.

Observation

The findings in the earlier report regarding the emerging importance of genetics, the need for development of relevant expertise for generalists and sub-specialists and the urgency of finding mechanisms to support the spread of high quality practice from centres of research expertise widely across the population as a whole was replicated across all the other specialties examined.

3.8 Views of clinical geneticists towards genetics in mainstream medicine

In a final discussion, opened at the meeting, but taken forward by email subsequently, the views of clinical geneticists regarding the subject of the genetics of inherited diseases in mainstream medicine included the following points:

1. The workshop mainly focussed on genetics and the identification and management of heritable diseases in patients and their families because this already provides real benefit to patients in many clinical specialties. A wider set of technologies and applications termed 'Genomic Medicine' would become relevant in the future.
2. There are real challenges in supporting genetics (and eventually genomics) in mainstream medicine. In particular the relatively small number of physicians with expertise in genetics within any one speciality will severely limit development in the near or even medium term future.
3. There is also a concern that non-genetic clinicians using genetic information and technology without a full understanding could result in adverse effects for patients and families. Sub-optimal or indiscriminate use of tests will not be cost-effective, and may prove very expensive, for the health service.
4. The scope and depth of education and training required to deliver genetic or genomic medicine within a specialty is currently not known and needs to be assessed jointly between the specialty and genetics experts.
5. The development of genomic medicine should build on the specialist expertise and experience of the clinical genetics specialty.

4 Discussion and recommendations

The prevailing rhetoric amongst basic science funders, researchers and many policy-makers both in UK and worldwide is that genomic medicine represents a revolution in healthcare.

It is envisaged that the use of genomic technologies to enable patient diagnosis and treatment based on information about a person's entire DNA sequence will become part of mainstream healthcare practice. Our report confirms that genomics is having an impact in many areas of clinical medicine, but that this is not so much a revolution as an evolution. Knowledge and experience is slowly gained by clinical research leaders and the process of embedding new practice in high quality care pathways throughout the UK is gradual and difficult.

The main findings and recommendations of the workshop are provided below:

1. New technologies and clinical knowledge have enabled significant progress in capability to diagnose and manage genetic and heritable disorders arising in a wide range of clinical areas. It is envisaged that this will rapidly be followed by a burgeoning of 'genomic medicine' in which wider analysis of genomic information is used to predict, prevent, diagnose and treat many common chronic disorders. It is important that the development and configuration of clinical and laboratory genetics and other specialties is optimised to meet the expected future capacity and range of needs. This should build on the strengths of existing structures and processes and aim to incorporate genomics into existing clinical pathways.

   We recommend that the relationship between specialist genetics services and a range of other clinical specialties should be developed as a key foundation for the development of genomic medicine. This will require a commitment to strengthen regional genetics services

2. With regard to innovations and high quality practice for heritable disorders, these are currently available only in a small number of specialist centres for each specialty and have not been systematically adopted across the UK. Populations thus have inequitable access to the necessary specialised services. Limited resources in terms of finance and availability of expertise mean that innovative models will need to be developed to provide the necessary services. These will vary according to the number and variety of conditions within a particular clinical area, the complexity of genotypic and phenotypic variation, the complexity and nature of long-term clinical management of patients and their families, the overall prevalence of the conditions and the expected availability of relevant expertise.

   We recommend the formal inclusion in the new commissioning structures of resourced, multidisciplinary expert groups, which may be specialty or disease specific depending on the context, able to give advice (via Public Health England or otherwise) on specifications for

Our report confirms that genomics is having an impact in many areas of clinical medicine, but that this is not so much a revolution as an evolution.

quality assured pathways to assist commissioners. These should set out requirements for high quality care integrating clinical, laboratory and long-term patient and family support and include guidance on different models of care. They should expressly include obtaining the maximum clinical benefit by cascading diagnosis and care to at-risk family members.

3. There will be a significant requirement for education for health professionals in specialties outside clinical genetics. This must include education for generalists and those who will provide more specialised advice and care. Clinicians within a specialty who have a sub-specialist interest in inherited disease or wider genomics will be an essential and leading element of the multidisciplinary team. For both generalists and specialists it will be necessary to identify the areas of specific 'genetic knowledge' and competence tailored for each specialty, to determine mechanisms to deliver the required training and to assure, by accreditation or otherwise, that practitioners have the required levels of expertise.

   We recommend that, through the Royal Colleges, the sub-specialty committees responsible for education and training develop plans for the inclusion of appropriate levels of genetics and genomics within their specialty training programmes.

4. Well-informed commissioning processes are vital to ensuring that genomics is integrated into high quality care pathways.

   The UKGTN should continue as a molecular testing network under the NHS Commissioning Board to support commissioners. We recommend that genetics training for commissioners should be provided working through national commissioning mechanisms and supported by expert public health genomic advice and the UKGTN.

5. The development of curricula and eventual delivery of teaching will require significant resources both from specialties concerned and from experts in clinical and laboratory genetics.

   We recommend that significant resources are made available for education and training in genetics across relevant clinical specialties. In particular the time commitment from specialist genetics necessary to provide support across the full range of specialties must be allocated in job planning, contracts and commissioning.

Conclusion and final recommendation

In January 2012 the Human Genomics Strategy Group[10] (HGSG) set out its vision for how the NHS could become a world leader in the development and use of genomic technology in healthcare and public health. The Group acknowledged that realising this vision will require many challenges to be met. Not least will be the challenge of delivery – the provision of genomic medicine

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as part of quality assured care pathways, by clinicians with the skills and knowledge to make effective use of the new technologies.

Our workshop, which preceded the HGSG report provides grass-roots endorsement for many of the key HGSG recommendations and embeds these in the real world of mainstream practice. We make some practical recommendations to support and strengthen genomic medicine in the UK

We recommend that, through its sub-speciality committees, the RCP promotes and supports those evolutionary changes through which genomic science and technologies can be harnessed to deliver better health.

Appendix 1 Discussion of terms

Genomic technologies encompass the whole range of laboratory technologies which can provide information on the 'genetic make-up' of an individual's germline, tissue or even non-human genomes. Traditional techniques such as karyotyping and Sanger sequencing are being superseded by newer methodologies such as array technology and next generation sequencing. Information may be generated about copy number variants, single gene mutations or multiple polymorphisms and variants. In some instances the impact of a single mutation will be very large with little modification of the phenotype from other genetic and non-genetic factors, for example the triplet expansion in the HD gene, whereas in other instances the resultant phenotypes will be the sum effect of multiple genetic polymorphisms, single gene effects and non-genetic effects, for example type 2 diabetes. The nucleotide analysis in these two instances will use most of the same technologies but the bio-informatic analysis may differ hugely.

Clinical genetics provides services for any person, or family, affected by or at risk of, a genetic disorder or congenital abnormality. These clinical services will include diagnostic assessment, counselling and support, provision of genetic advice to the extended family and management frequently through the co-ordination of multidisciplinary teams. This is delivered through integrated clinical and laboratory genetic services which are safe, efficient, appropriate, accessible and acceptable to all sectors of the community and which are of a demonstrably high quality.

Traditionally the scope has encompassed chromosomal disorders, complex dysmorphic syndromes, teratogenic disorders and single gene disorders. The boundaries between these component groups has blurred as technology increasingly reveals the underlying aetiology, such that some complex syndromes are identified as single gene disorders whereas others are due to copy number variation of multiple genes. The blurring is equally complex in the field of single gene disorders where the variable presentation of a hitherto recognised single genetic disease may show great variability in the penetrance and expressivity due to an underlying heterogeneity or the interaction of multiple genetic and non-genetic modifiers. Furthermore the response to therapeutic intervention may be greatly influenced by the underlying aetiology and these modifiers. This blurs our categorisation of what are single gene disorders and what are complex multi-factorial disorders. Therefore the demarcation of clinical geneticists providing services for single gene mendelian disorders and non-genetic services providing services for non-single gene disorders is outdated and newer models should consider how to provide up- to- date and informed care guided by the principle of holistic care of the highest quality.

Genomic healthcare refers to the provision of healthcare informed by including knowledge gained from techniques to understand the genetic aetiology or influences on the underlying disease. In some instances this might be recognition of the precise sub-type or cause; in others the interaction between the genetic and non-genetic components; in a third group the response to therapeutic intervention based on the genetic factors. These may be instigated from a wide range of specialties from GPs taking a family history of cancer, to oncologists requesting genomic expression profiles on tumours, through to clinical geneticists asking for a specific single gene test. Therefore the technology used should be applicable across disciplines and clinical diseases.

Appendix 2 List of attendees

Name	Organisation
Dr Gillian Baird	Chairman, British Academy of Childhood Disability, London
Dr Edward Blair	Consultant in Clinical Genetics, Department of Clinical Genetics, Churchill Hospital, Headington, Oxford
Dr Paul Brennan	Consultant in Clinical Genetics, Teeside Genetics Unit, James Cook Hospital, Middlesbrough
Professor Sir John Burn	Professsor of Clinical Genetics, Institute of Human Genetics, International Centre for Life, Newcastle University
Dr Hilary Burton	Director, PHG Foundation, Cambridge
Professor Kris Chatterjee	Professor of Endocrinology, Institute of Metabolic Science Addenbrooke's Hospital, Cambridge
Professor Lyn Chitty	Professor of Genetics & Fetal Medicine, Institute of Child Health, University College London
Dr Trevor Cole	Consultant Clinical & Cancer Specialist, Clinical Genetics Unit Birmingham Women's Hospital NHS Foundation Trust
Dr Ellen Copson	Consultant in Medical Oncology, Southampton University Hospital Trust
Dr Campbell Cowan	Consultant in Cardiology, Leeds General Infirmary
Professor Sian Ellard	Consultant Molecular Geneticist, Molecular Genetics, Royal Devon & Exeter NHS Foundation Trust
Dr Anthony Ellis	Consultant Physician & Gastroenterologist, Royal Liverpool University Hospital
Professor Gareth Evans	Professor of Medical Genetics, Regional Genetics Service, St Mary's Hospital, Manchester
Professor Peter Farndon	Director, NHS National Genetics Education & Development Centre, Birmingham Women's Hospital NHS Foundation Trust
Dr Neil Gittoes	Consultant Endocrinologist, Queen Elizabeth Hospital, Birmingham
Professor Andrew Hattersley	Professor of Molecular Medicine, Peninsula Medical School, Exeter
Dr Layla Jader	President of the Society for Genomics Policy and Population Health (SGPPH) and Consultant in Public Health Medicine

Dr Edmund Jessop	Medical Adviser, National Commissioning Group, NHS London
Mr Alastair Kent	Director, Genetic Alliance UK, London
Dr Graham Lipkin	Consultant Nephrologist, Queen Elizabeth Hospital, Birmingham
Professor Anneke Lucassen	Joint Lead Clinician, Wessex Clinical Genetics Service, Princess Anne Hospital, Southampton
Dr Ian McDowell	Consultant & Senior Lecturer in Medical Biochemistry, Department of Biochemistry, University Hospital of Wales, Cardiff
Professor Albert Ong	Professor of Renal Medicine, Department of Infection & Immunity, The University of Sheffield Medical School
Professor John Pasi	Professor of Haemostasis and Thrombosis, Barts and the London School of Medicine & Dentistry, London
Dr Christine Patch	Consultant Genetic Counsellor, Clinical Genetics, Guy's Hospital, London
Mr Colin Pavelin	Head, Advanced Therapies & Genetics, Health Science & Bioethics Division, Department of Health, London
Dr Imran Rafi	GPwSI Genetics and Senior Lecturer Primary Care Education, St George's Hospital, London
Mr Anthony Roberts	Consultant in Obstetrics & Gynaecology, Queens Hospital, Burton, Staffordshire
Dr Richard Sandford	Reader, Department of Medical Genetics, Cambridge Institute for Medical Research, Addenbrooke's Hospital NHS Trust
Dr Graham Shortland	Consultant Paediatrician, Department of Child Health, University Hospital of Wales, Cardiff
Dr Claire Shovlin	Senior Lecturer, NHLI Cardiovascular Sciences, Imperial College, London
Dr Peter Turnpenny	Consultant Clinical Geneticist, Clinical Genetics Department, Royal Devon & Exeter NHS Foundation Trust
Professor Mark Thursz	Professor of Hepatology, Imperial College, London
Dr Jacquie Westwood	Programme Director, UK Genetic Testing Network, London
Professor Gerry Wilson	Head of the Department of Rheumatology, University of Sheffield
Dr Ron Zimmern	Chairman, PHG Foundation, Cambridge

Appendix 3 Programme

Monday 20th June 2011 PROGRAMMME WITH PROVISIONAL TIMINGS		
9.00–9.30	Registration and Coffee on arrival	
	Welcome and introduction to the workshop **Sir Richard Thompson**	
9.30-9.45	**Session 1: Setting the scene – drivers for change** **Chairman: Sir Richard Thompson**	
9.45-10.15	Scientific and clinical advances and future potential The needs of patients The political scene including the changing NHS The evolving role of specialist clinical and laboratory genetics	**Prof Sir John Burn** **Dr Hilary Burton**
10.15-10.30	Discussion – have we missed anything?	
10.30- 11.45	**Session 2: Service provision for inherited disease** **Chairman: Hilary Burton**	
10.30-11.15	Genetics led Cancer genetics Specialty led Mixed specialty and genetic development High volume Complex disease	**Dr Paul Brennan** **Dr Trevor Cole** **Prof Nick Wood** **Dr Neil Gittoes** **Dr Ian McDowell** **Prof A Hattersley**
11-15-11-45	Discussion – what models of care will be needed in the future?	
11.45-12-00	***Coffee break***	

12.00–13.00	**Session 3: Sub-specialisation vs generalisation** **Chairman: Peter Farndon**	
12.00-12.30	A service where there is sub-specialisation in inherited disease (haematology) A service where inherited disease is important but there is usually not sub-specialisation (fetal medicine) A service where most practitioners would need to understand and manage inherited disease (*e.g.* paediatrics) A consideration from NGEDC of the comparative roles and needs of specialists and generalists	**Prof John Pasi** **Mr Anthony Roberts** **Dr Graham Shortland** **Prof Peter Farndon**
12.30-13.00	Discussion – how will necessary levels of skill be ensured?	
13.00 – 13.45	***Lunch***	
13.45- 14.35	**Session 4: Pathways of care** **Chairman Trevor Cole**	
13.45-14.15	The patient viewpoint (from GA UK project on pathways) The clinical genetics viewpoint The specialist/generalist viewpoint The commissioner viewpoint (*e.g.* UKGTN or other specialist commissioner)	**Mr Alastair Kent** **Dr Peter Turnpenny** **Dr Campbell Cowan** **Mrs Jacquie Westwood**
14.15 – 14.35	Discussion – how can pathways of care be developed to ensure high quality outcomes for patients?	
14.35-15.25	**Session 5: The new commissioning climate** **Chairman: Hilary Burton**	
14.35-14.55	GP consortia and commissioning arrangements Specialist commissioning A network commissioner	**Mrs Jacquie Westwood** **Dr Edmund Jessop**
14.55-15.25	Discussion – what needs to happen to make this work in the new NHS?	

15.25-15.40	Tea and cakes!	
15.40-16.30	**Session 5: Moving Forward** **Chairman: Trevor Cole**	
15.40-16.30	Collection of main bullet points Formulation of recommendations Agreement of process for workshop report Agreement on process for RCP positioning statement	
16.30	Close	

Appendix 4 Models of care

The **CARDIGEN cardiac genetics** service in the North East has developed a hub and spoke clinical network with clinical guidelines for referrals. The network is composed of specialist and non-specialist service elements supported by 'working rules' such as agreed staff competences, communication pathways, supervisory relationships and accountability. There is a secondary care-based family history triage service for the more common disorders such as hypertrophic cardiomyopathy and a range of tertiary cardiac genetics clinics with a cardiologist 'on-call'. Cascade genetic testing is genetic counsellor-led.

In **cancer genetics**, demand is managed in the West Midlands largely through a system in which general practitioners give patients the initial family history form and are supported through an initial triaging process regarding patient referral. In those families where the primary care practitioner is still uncertain of the appropriate route within the standardised pathway the form is reviewed by a genetics consultant or genetic counsellor and patients are referred to the appropriate health professional for follow up or seen in clinical genetics to instigate molecular testing if appropriate. The form is utilised as the referral letter if required.

This has resulted in a significant reduction of patients with near population risk of cancer being referred inappropriately or having unnecessary surveillance. At the same time there has been a steady increase of referrals of high-risk single gene families who might benefit from changed management on the basis of the genetic test. It has also allowed for cascade testing to be undertaken by the local genetic counsellor network if appropriate.

Although the programme is lead by clinical geneticists, most patients receiving surveillance are managed by local health professionals who are not genetic specialists, using pathways developed by a multidisciplinary team.

Neurogenetics presented a different problem with a large number of very rare conditions needing consideration and each with its own clinical diversity. There are a growing number of specialist neurogeneticists and, on the whole, increasing expectations amongst professionals and patients that DNA tests should be offered. These tests were, however, often perceived as being expensive, a belief that was not necessarily well-founded when compared with other tests such as MRI scans.

Increasingly tests would be more comprehensive, rapid and inexpensive. For example, it was thought that there might soon be array tests for particular conditions – for example a 'dystonia array'. However, the individual rarity of many conditions means that even experienced neurologists may have limited clinical experience of the disorder, selecting appropriate tests and providing clinical advice. Furthermore there are significant challenges in translation of molecular data into clinically useful information. Problems include: assessing the pathogenicity of variants; linking genotype with phenotype, given the inaccessibility of brain tissue to provide a final diagnosis; the need for bioinformatic solutions to provide data interpretation for clinicians; and, in turn the need for clinicians to interpret results for patients and enable them to be used in treatment decisions. It is likely, therefore, that neurogenetics will remain a highly specialised area of clinical practice.

Review of family history forms by genetics specialists in the West Midlands is reducing rates of inappropriate referrals and increasing referrals for high-risk single gene families.

The **endocrine genetics** service in the West Midlands provided an example of a tertiary service that had grown historically with close but informal links between genetics and endocrinology and with an academic focus. The service was organised as monthly one-stop multi-specialty clinics that are embedded in endocrinology. Most of the funding of the outpatients, clinical support and non-genetic investigations was provided from endocrinology and only the clinical geneticist and some of the molecular testing are funded from the regional genetics budget. The four joint clinics cover a range of sub-specialisations including general endocrine genetics, bone and calcium disorders, multiple endocrine neoplasia and reproductive medicine.

The service had worked hard to integrate important features, notably a transition service from paediatrics and a multi-specialty approach. Issues that had arisen during development included coordination of job plans, questions about the tariff for a low volume high complexity service, the use of clinic space, and how patients would best be followed up long term.

An important feature in the provision of this service was the employment of a specialist nurse who had key roles as the primary point of contact for patients, in communication (translating, emphasising and repeating messages for patients), coordination of the service with other elements including primary care, and the integration of knowledge from both specialties. The service was seen as being highly successful, well regarded by patients and strengthening relationship and knowledge between the two groups of professionals. There was also great emphasis placed on flexibility between genetics and endocrine services to offer patients the best deal. There was recognition that the relative contributions of genetics and endocrinology change over the follow up period, determined by the specific needs of the patient at any given time.

Familial hypercholesterolaemia (FH) cascade testing provided the example of an intervention involving genetic testing for a relatively common condition (1 in 500) for which good treatment options are available. Provision of cascade testing for family members was supported by NICE guidance and yet it still proved extremely difficult to get this service implemented across the UK. The Wales programme developed a very detailed care pathway with agreed regional and local management and accountability, which separately covered the work of the lipid clinic in managing the individual patient, the FH nurse for providing further patient advice and support, and the role of the genetic counsellor in undertaking the further cascade testing of the family. An important feature of the model was the use of software developed in the Netherlands to support clinical management and workflow and coordination of laboratory results. The provision of molecular testing within the whole pathway has remained a significant barrier to many other regions, especially in England.

The **monogenic diabetes group** based at the Royal Devon and Exeter Hospital provided a good example of how making a correct molecular diagnosis in Maturity Onset Diabetes of the Young (MODY) determines the clinical picture and treatment response. The majority of patients with MODY are very sensitive to sulphonylureas and can improve their glucose control by using these tablets in place of insulin. This condition accounts for approximately 1% of diabetes, with a predicted prevalence of between 20,000 and 50,000 cases in the UK. Ninety percent of patients with MODY are mistakenly thought to have Type 1 or Type 2 diabetes and this means that many patients are receiving sub-optimal treatment. There is a UK laboratory able to provide the necessary diagnostic tests, but it was noted that requests for testing vary 10 fold across the country.

It was thought important that this should be part of the routine diabetes service and that in many cases this may be independent of genetics. The group had evidence that a key to ensuring that this service is available equitably across the UK was a scheme of training using regional genetic diabetes nurses. However, following a successful DH pilot, requests to continue funding through normal commissioning routes were rejected by genetics funders, being perceived as mainstream medicine, and conversely by diabetes commissioners who perceive it is as specialist genetics. The genetic diabetes nurses are currently supported by charitable fixed-term funding.

Appendix 5 Levels of sub-specialisation for inherited disorders in mainstream medicine

In haematology, two clinical areas of managed inherited diseases are inherited bleeding and thrombophilia disorders. The general format for the provision of care for these inherited disorders is through comprehensive care centres based in haematology departments, which offer holistic care to patients. Just over half these services have integrated molecular genetics services, some of which are provided in conjunction with regional genetics centres.

In thrombophilia practice, tests for a number of common variants are widely available, but of variable utility. It is not clear whether or how they would be introduced into clinical practice. It is, however, thought likely that genetic testing will shortly be required in the context of gene therapies, which are currently in development.

Training and education for sub-specialisation is not likely to be particularly problematic in haematology, as this is a specialty with a significant molecular component both within the training programmes and throughout the subsequent delivery of service. Although many doctors who go into the specialty wish to pursue a career in haemato-oncology, all get a thorough grounding in the molecular basis of disease – and so this should provide a good basis for work in inherited disease. Therefore, integrating the developments from molecular medicine is likely to be more easily facilitated in this specialty than some other sub-specialties.

Antenatal care is provided both by generalists and by specialists in fetal medicine, both of which are involved in the consideration of genetic or inherited disorders in their practice. Patients usually present either following the previous identification of a genetic problem requiring genetic advice with a prenatal diagnostic plan, or when an unexpected anomaly is diagnosed in a current pregnancy. It is important to agree who provides care at various stages of the pathway.

Looking to the future, where there may be wider use of genetic testing as, for example, in carrier screening for cystic fibrosis or non-invasive testing using cell free fetal DNA in the maternal serum for a variety of conditions, it is clear that there will be substantial need for obstetrics and fetal medicine to apply an increasing amount of genetic expertise and therefore additional training will be required throughout the specialty. However, this training should reflect the different roles likely to be undertaken in the antenatal pathway.

In paediatrics, additional education to support the introduction of new technology such as micro-arrays is needed.

Paediatrics is a specialty that already works closely with genetics, in diagnosing and managing those disabling, chronic diseases of childhood that are genetic in origin. These include conditions such as cystic fibrosis and inherited metabolic diseases that are increasingly diagnosed through screening programmes, and many of the conditions that present with developmental delay. Much of the paediatric input for the investigation and subsequent follow up of children with developmental delay is provided by 'generalists' and community paediatricians who need to maintain close links with clinical genetics and dysmorphology expertise. Additional education to support the introduction of new technology such as micro-arrays is needed.

There are a number of areas in which sub-specialist training in genetic aspects would be helpful (for example inherited metabolic disease) but, so far, it appears that this has not been achieved within specialist training. The Royal College of Paediatrics and Child Health has the prime responsibility for this training and should be encouraged to consider genomic developments and applications generally in the paediatric curriculum and to identify the sub-specialist areas where further knowledge will be necessary.

Appendix 6 Clinical pathways of care

From the **patient point of view** there are many examples of good practice across the country, but significant problems of inequity. Patients want access to highly specialised centres that will ensure a definite diagnosis, appropriate pathways of care, accurate information and an over-arching route-map to help them understand and access support to cope with the condition. They need this care to be integrated with the rest of the service and have mechanisms to cascade the process through the family if appropriate. Pressure to systematise good practice can be effective when coming from patient and public groups. This should be recognised as an asset and these groups should be involved as a routine when providers and commissioners develop guidelines and service specifications.

From the **clinical genetics viewpoint**, pathways of care may start from a given diagnosis or for a particular clinical problem. For a given diagnosis, it is likely the pathway consists of a package of multidisciplinary management, which may include, for example a programme of surveillance or screening. It is essentially holistic, coordinated and evidence based and includes family members at risk as well as the initial diagnosed patient. When a pathway of care begins with a clinical problem, the immediate requirement is to make a diagnosis and the pathway should set out which clinical professionals should contribute to this process. In particular it may specify who initiates, performs and interprets genetic testing, whether these tests would be targeted or more general tests, and the many issues around obtaining and recording informed consent. Decisions on which tests might be employed within a given clinical pathway would be based on considerations of cost effectiveness and the clinical utility of the test in that specific case. This should include consideration of whether there are the expert skills to interpret complex or equivocal results and a mechanism is in place to cascade testing of the extended family where appropriate.

The role of the clinical geneticist in testing should be specified in the service specification and might include: general genetics expertise, bioinformatics, expert clinical diagnosis, management and investigation of families, counselling and relevant ethical expertise.

The development of recommendations for clinical pathways is an important contribution to commissioning and should be achieved through multidisciplinary activities, which may currently be based on existing clinical networks. Implementation will require education of a range of professionals, which should be reflected in their job plans, an expanded role for genetic counsellors and the training of more 'specialists with an interest in genetics.'

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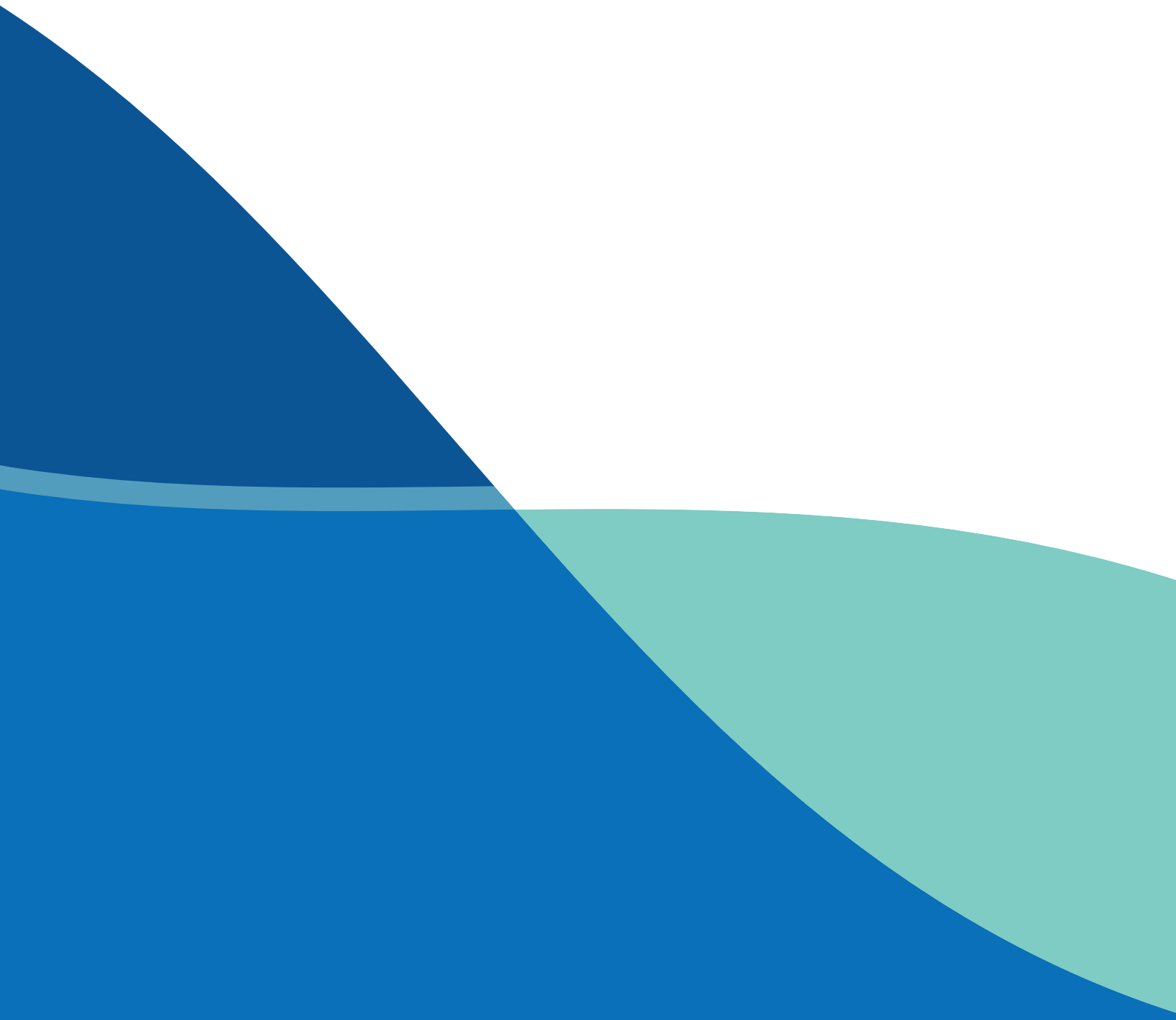
The PHG Foundation is a forward-looking policy think-tank and service development NGO based in Cambridge, UK. Our mission is *making science work for health*. We work to identify the best opportunities for 21st century genomic and biomedical science to improve global health, and to promote the effective and equitable translation of scientific innovation into medical and public health policy and practice.

We provide knowledge, evidence and ideas to stimulate and direct well-informed debate on the potential and pitfalls of key biomedical developments, and to inform and educate stakeholders – policy makers, health professionals and public alike. We also provide expert research, analysis, health services planning and consultancy services for governments, health systems, and other non-profit organisations.



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