## Contents

<table>
<thead>
<tr>
<th>Section</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Executive Summary</td>
<td>2</td>
</tr>
<tr>
<td>Why choose the UK to provide genomic services?</td>
<td>3</td>
</tr>
<tr>
<td>Planning and Service Development</td>
<td>7</td>
</tr>
<tr>
<td>Clinical Services</td>
<td>9</td>
</tr>
<tr>
<td>Laboratory Services</td>
<td>13</td>
</tr>
<tr>
<td>Education and Training</td>
<td>17</td>
</tr>
<tr>
<td>Appendix 1 – Ethical Framework</td>
<td>22</td>
</tr>
<tr>
<td>Appendix 2 – Membership of the UKGTN Task and Finish working group</td>
<td>23</td>
</tr>
</tbody>
</table>
UK Genetic Testing Network (UKGTN)

UKGTN is a national advisory organisation for NHS genetic testing services. It was set up by the Department of Health in 2002 to promote equity of access to gene testing within the NHS. It is a collaborative of clinicians, scientists, patient representatives and commissioners and has a membership of laboratories.

Over 60 colleagues from the UK clinical genetics community provide advice to four working groups in the delivery of the annual work programme. The member laboratories are, in the main but not exclusively, associated with NHS Regional Genetic Centres within NHS tertiary Trusts. The laboratories apply to be members of UKGTN and are accepted providing they meet the agreed stringent quality criteria.

The UKGTN is supported by the project team, advisors and chairs of the working groups. The accountability is through the UKGTN Clinical and Scientific Advisory Group that has a wide representation from the member nations, professional bodies, the Department of Health and Patient Groups. The work of the UKGTN influences policy development, provides advice to commissioners, assures quality of laboratories and the Network services they provide and evaluates and recommends new genetic tests for NHS service.
Executive Summary

In April 2014 the UK Genetic Testing Network (UKGTN) and Healthcare UK hosted a conference to assess whether there would be interest in taking forward international business opportunities for NHS genomic services. This brochure has been developed with colleagues in clinical genetics from across the UK to provide information about the services that are currently available and that the NHS could offer internationally.

A wide breadth of services are available including business and planning, clinical and laboratory services and training and educational courses. An ethical framework provides an overview of the standards that these UK services adhere to in order to provide quality assured services.

The UK is a recognised leader in genomics research, treatment and care. It is at the forefront of the genomics revolution which could radically transform the way that patients are diagnosed and treated. In late 2012 the Prime Minister announced the launch of the 100,000 Whole Genomes Project which will aim to sequence 100,000 whole genomes from NHS patients by 2017. NHS genetic services are available in all four UK countries with 23 Regional Genetic Centres that each run as multidisciplinary teams. Providers are regularly assessed to assure quality standards are maintained.

The UK genetics services can provide consultancy expertise within and outside of the UK. Expertise is available in health needs assessment, financial planning, policy development, project management, technology appraisals, service improvement, workforce planning and patient/laboratory information systems.

Clinical genetic services deal with families, often over several generations, and can provide expertise for heritable diseases to any age group affected by, or at risk of, genetic disorders. The aim of NHS clinical genetic services is to offer high quality, clinically effective services with equity of access to the population. Technological and scientific advances related to genomic medicine mean that increasingly clinical genetic services are becoming integrated in mainstream medicine.

The UK NHS laboratory genetics community collaboratively provides genetic testing for more than 1000 disorders and their associated genes. Information on all recommended tests and provider members within the UK are collated via the UKGTN (www.ukgtn.nhs.uk/). In addition to providing a broad portfolio of tests the UKGTN laboratories have expertise in technology development, sample processing and analysis, quality assurance systems, IT systems, bioinformatics, interpretation and clinical reporting.

UK education and training facilitates genetic centres to run as multidisciplinary teams. This includes formal graduate degrees (pre registration), postgraduate studies (post registration) and continued professional development (clinical practice) for the workforce. Modernising Scientific Careers introduces training programmes at four main career level entry points for scientists. The Joint Royal College of Physicians Training Board provides ongoing training for clinicians and genetic counsellors are supported with training from the Association of Genetic Nurses and Counsellors. Health Education England has a Genomics Education Programme and short courses are run by the British Society of Genetic Medicine and its affiliated organisations or through UK centres.

NHS providers work to an ethical framework that provides an overview of ethical standards and UK legislation to ensure quality of patient care. This includes standards in the areas of direct to consumer testing, predictive testing for children, sex selection for social reasons, training and qualifications, the Human Tissue Act 2004, the Data Protection Act 1998 and consent.
Why choose the UK to provide genomic services?

The UK National Health Service Clinical Genetic Services have an international reputation for excellence. The NHS brand provides high levels of trust and credibility both nationally and internationally. Organisations that are part of the NHS family provide reassurance that the services are delivered in line with NHS values. The NHS has dedicated services in all four UK countries, England, Scotland, Northern Ireland and Wales.

The UK is a recognised leader in genomics research, treatment and care. It is at the forefront of the genomics revolution which could radically transform the way that patients are diagnosed and treated. In late 2012 the Prime Minister announced the launch of the 100,000 Whole Genomes Project which will aim to sequence 100,000 whole genomes from NHS patients by 2017.

All services are underpinned by an evidence base that includes:
- analytical validity
- clinical utility
- ethical/legal and social considerations

Number per annum
- 23 Regional Genetic Centres
- >30 Genomic Laboratories
- >4,800 Clinically relevant genes tested
- 40,000 Clinical referrals
- >3,700 Specialist Genetic Clinics
- 200,000 Genetic test results issued

Number evaluated/developed since 2004
- >1,000 Genetic disorders with new evaluated tests
- >500 New tests evaluated (many new panel tests are testing for a number of different disorders)
- >450 Testing Criteria (clinical guidance for testing)
- >45 Next Generation Sequencing panels for multi gene tests

www.ukgtn.nhs.uk
Quality standards in the NHS are world class. Providers are regularly assessed to ensure standards are maintained. They follow best practice and achieve value for money through effective use of resources. The UK is respected for adhering to an ethical framework, regulations and laws that respect the dignity of individuals at all times – see Appendix 1 Ethical Framework.

Staff working in NHS genetic services are appropriately trained and registered with the necessary registration board for example General Medical Council Specialist register, Genetic Counsellor Registration Board, Nursing and Midwifery Council, Health and Care Professions Council.

The NHS is value for money. The NHS in the UK has been found to be one of the most cost effective systems when compared to the USA and 17 other Western countries.\(^1\) The UK was also ranked second in an assessment of health systems in seven countries published by the Commonwealth Fund.\(^2\)

Expert clinical advice guaranteed. The UK promotes integrated working across academia, the sciences and clinical disciplines to deliver services that are at the cutting edge of new discoveries. Clinical care pathways include primary care, local hospitals, regional centres and specialist clinical centres. Specialist services have a depth of expertise about a range of genetic conditions. Expert knowledge is supported by high quality training programmes that recognise that rare diseases require additional skills. Each year in the UK more than 3700 joint specialist clinics (clinical genetics working with other clinical specialties) take place.

NHS genetic services aim to offer the highest clinical standards working within a clinical framework with adherence to codes of ethical practice and conduct. This applies to both medical and non medical practitioners and is also consistent with international codes and standards.

GMC Good Medical Practice

---

Typical professional staff numbers and qualifications for a Genetic Centre serving a 3 million population

The 23 Regional Genetics Centres each run as a multidisciplinary team. A typical Regional Genetic Centre service is illustrated above, detailing the numbers and qualifications of staff to serve a 3 million population and staffing numbers across the UK.
What is available?

An overview of the wide range of services is catalogued by the UK Genetic Testing Network (www.ukgtn.nhs.uk/our-work/ukgtn-reportsguidelines/ukgtn-guide-to-centres/).

The listings give information about the clinical specialties and laboratory services available. Uniquely the NHS evaluates all new genetic tests for rare disorders prior to service implementation. Therefore clinicians can be confident about the quality and clinical utility of the tests offered.

Case study
Monogenic diabetes

Exeter is the world’s leading centre for diagnosing monogenic forms of diabetes which accounts for 2-3% of all diabetes. The research led by Professors Sian Ellard and Andrew Hattersley has identified many novel genetic forms of diabetes in patients referred from >80 countries. They showed that patients with the most common HNF1A MODY subtype respond best to treatment with sulphonylurea tablets, although most of these patients are misdiagnosed with type 1 diabetes and inappropriately treated with insulin injections. The genetics laboratory in Exeter provides clinical advice as to the most appropriate testing strategy, immunological/biochemical assays, comprehensive genetic testing for all known monogenic subtypes, full clinical interpretation of test results and advice on treatment for patients and their relatives.
As a world leader, the NHS has clinical genetic service providers that are able to provide consultancy services to support the planning, development and establishment of clinical genetic services at local, regional and national levels.

Specialist health planning can be provided to develop the specifications for necessary services for differing populations or customer base.

Clinical and laboratory services are well established to deliver innovative, cost-effective, evidence-based models of integrated care. Underpinning the growth and development of these services are processes to establish the population health needs, analyse current services, review clinically effective and cost effective interventions and services and develop clinical guidelines and audits.

Working in a consultancy capacity, NHS genetic services can provide high quality multidisciplinary expertise required to support the range of services outlined. Expertise includes but is not limited to, clinical genetics, genetic counsellor, clinical scientist, laboratory scientist, clinical informatician, bioinformatician, public health and healthcare finance.

If there was international interest in current NHS services these could be delivered in the UK and for some elements delivered in the country where they are required.

UK Genetic consultancy services can provide:
- Comprehensive review of local population health needs assessment
- Strategic services and business planning:
  - Financial planning
  - Policy development
  - Project management
  - Technology appraisals
- Service improvement
- Workforce planning to identify current and future capacity requirements
- Patient and Laboratory information systems

www.ukgtn.nhs.uk
Health Needs Assessment
The experience and expertise in meeting complex health demands of diverse populations has enabled NHS genetic services to deliver effective and equitable services that meet the needs of local populations. We have the skills and knowledge to assess and advise on the prioritisation of the health needs of your population and enable resources to be used most effectively.

Strategic Services and Business Planning
NHS expertise is ideally placed to help plan and deliver genetic services to support significant scientific and technological developments and evolving healthcare needs. Consultancy services can be provided to undertake the development of business models, technology appraisals, policy development, financial planning and project management.

Service Improvement
The ability to transform services that lead to significant and sustainable improvements in quality and productivity are essential in delivering positive patient experience and cost efficient services.

The experience and expertise of high impact changes within the NHS can be drawn upon to develop a framework to assist in reviewing existing services to improve service delivery and optimise the allocation of healthcare resources.

Workforce Planning
As scientific knowledge and technology expands consideration must be given to the future shape and size of the workforce. Capacity must be planned against service requirements. We can assist in helping define your current and future workforce.

Patient and Laboratory Information Systems
A number of different integrated clinical and laboratory Genetics IT systems have been successfully implemented across the UK. The variation in these systems, and the unique relationship between NHS providers and industry, means that a wide range of expertise is available to help develop robust and effective health management information systems to support the specialised system requirements of genetic services.
Clinical Services

Clinical genetic services deal with families, often over several generations, and can provide expertise for heritable diseases to any age group affected by, or at risk of genetic disorders.

The aim of NHS clinical genetic services is to offer high quality, clinically effective services with equity of access to the population. Technological and scientific advances related to genomic medicine mean that increasingly clinical genetic services are becoming integrated in mainstream medicine.

Although there is a network of clinical genetic services across the whole NHS, addressing the requirements of local populations has led to the development of flexible models of service delivery. There is therefore experience of a range of models of service based on local expertise and population needs.
The Referral Pathway

Patient

Referral to Genetics from healthcare professional

Cancer Genetics  General Genetics  Prenatal Genetics
The woman sees her local family doctor and is referred to her breast clinic where she sees a breast nurse trained and supported by the genetic centre. The breast nurse assesses the family history, organises screening and refers to the genetics clinic for an appointment if appropriate.

**General**

A married couple are first cousins. They do not have any known family history of medical problems. After birth, their baby was found to be floppy and started fitting. He also has a 'hole in the heart'.

The local paediatrician has assessed the child and prescribed medication to control the seizures. The paediatrician refers the child and parents to a Clinical Genetics doctor to assess the child and investigate in order to diagnose the underlying condition which is likely to be genetic. A confirmed diagnosis will resolve the anxiety of uncertainty, will help the future management of the child, and will provide the couple with an accurate recurrence risk and option of testing in pregnancy, should they want this.

**Prenatal**

A couple have a little boy who was born with cataracts in his eyes and suffering with kidney problems. A specific diagnosis was suspected, which would predict that the boy is likely to show subsequent severe learning and behavioural difficulty as well as deteriorating kidney function. After a detailed genetic test a mutation was identified in the gene which causes this condition, Lowe syndrome. The mother is pregnant and the couple want to know if the fetus has inherited the same condition.

The couple see a Genetic counsellor who explains that this condition only causes these problems in boys as the genetic alteration is located on the X chromosome. A baby girl could be a carrier of the condition but would not suffer from the medical problems like their son does. If the fetus is male, it could be similarly affected, but also has a chance of at least 50% of not inheriting the condition.

The actual risk for the fetus would depend on whether or not DNA from the mother's blood indicates that she is a carrier, but the risk never drops to zero.

The genetic counsellor first offers a blood test to the mother as it is now possible to find out the sex of the baby in the womb from analysing the mother’s blood alone. If the baby is female, no invasive testing would be required. The result comes back showing the baby is male. The couple are then offered an invasive test taking a biopsy of the placenta to find out whether the male baby is affected or unaffected, by testing for the same genetic change as in their son. The result of this test shows the baby is unaffected. The couple carry on with the pregnancy as normal.

The mother is also offered DNA testing from her blood to see if she is a carrier of the genetic change, but even a 'normal' result does not eliminate a small risk in future pregnancies.

Having the diagnosis in the son provides answers to the parents, enables the option of a test in the pregnancy if they wish, and also enables advice to be given to the wider family as a reliable carrier test is available to female relatives on the mother’s side.
Summary of services

A bespoke service can be created from the menu below in order to meet your country’s needs. The NHS would be happy to consider requests if there are particular clinical services you would like to be made available.

**Clinical Services**
- Teleclinics (telephone/Skype)
- International Specialist clinics
- Remote genetics advice
- Genetics counselling services
- Multidisciplinary clinics

**Teaching / Training**
- Shadowing clinical geneticists or counsellors
- Work-place secondments
- Accredited courses, seminars and conferences
- Focused module training (Cancer, prenatal, cardiac genetics etc.)
- Training of local physicians

**Management**
- Consultancy services (setting up genetic services abroad)
Laboratory Services

The UK NHS laboratory genetics community collaboratively provides genetic testing for more than 1000 disorders and their associated genes. Information on all recommended tests and provider members within the UK are collated via the UK Genetic Testing Network (UKGTN).

Each laboratory within the network is accredited by the United Kingdom Accreditation Service with clinical pathology accreditation (CPA) and either has or is working towards ISO15189 standards.

The laboratories provide a broad portfolio of genetic tests for their local population, together with tests for disorders in which they have specialist expertise that they provide on a national and international basis. In addition many laboratories offer genomic tests related to cancer beyond the UKGTN directory.

From this unique resource the UK can offer a menu of genetic testing or support for the wider international community including:

- laboratory technology development
- sample processing and analysis
- quality assurance systems
- IT systems
- bioinformatics
- interpretation and clinical reporting
- training and support.

External Quality Assessment

Laboratory organisations that contribute to the UKGTN offer testing in a variety of ways on diverse platforms but each test or technology is scrutinised by external quality assessment (EQA) to ensure results for patients are accurate and reproducible. Not for profit organisations provide external quality assessment schemes for genetics within the UK, and are also available to laboratories worldwide:

National External Quality Assessment Scheme (NEQAS) – www.ukneqas.org.uk

European Molecular Quality Network (EMQN) – www.emqn.org

Testing services

Each laboratory within the UK Genetic Testing Network is able to offer end to end support for the testing from sample receipt through processing to analysis and interpretation. The end product will be a clinical interpretive report to be distributed to the referring clinician for them to deliver the outcome to the patient.

www.ukgtn.nhs.uk
Types of testing and testing scenarios

- **Diagnostic tests**
  - Targeted tests for single gene
  - Gene panel test two to several hundreds of genes
  - Clinical exome or whole exome tests
  - Whole genome analysis
  
- **Predictive tests**
  - Carrier tests
  - Prenatal tests

**My patient has multiple congenital abnormalities. I don’t know what the diagnosis is but it is likely to be genetic. Can you test my patient?**

**Diagnostic testing** to confirm the molecular or chromosomai basis of the congenital abnormalities using one or more of the various types of test listed above.

**Has my patient got inherited disorder X?**

**Diagnostic testing** to confirm the molecular or chromosomal basis of an inherited or sporadic disorder (e.g. Neurofibromatosis type 2 or Prader Willi syndrome) using one or more of the various types of test listed below.

**Will my patient develop the inherited disorder that is present in their relative?**

**Predictive testing** also known as pre-symptomatic testing – used to detect a familial gene mutation associated with a disorder that appears after birth, often later in life.

The person having the test has no features of the disorder at the time of testing, but has a family member with a genetic disorder where the mutation has been identified, e.g. testing for a familial mutation in the MSH2 gene that causes hereditary non-polyposis cancer (HNPCC).
Is my patient a carrier of the inherited disorder present in their family?

Carrier testing is used to determine if a person is a carrier for a specific autosomal recessive disorder such as Cystic Fibrosis. For example, where one partner has a family history of a recessive disorder it may be requested by couples who are considering becoming pregnant to determine if they carry a mutation therefore if there is a risk of their child being affected by that genetic disorder.

My patient is pregnant will her baby be affected by the inherited disorder present in her/her partner’s family?

Prenatal diagnosis — testing a fetus or embryo for a genetically inherited disorder which is at high risk of inheriting e.g. Cystic Fibrosis, Duchenne muscular dystrophy, Fragile X syndrome. Confirming a diagnosis which has been indicated by another testing method (e.g. Down syndrome in a pregnancy predicted to be at high risk in a screening programme).

Targeted tests:
- Gene tests – identifies variations or mutations in a specific gene that can lead to a genetic disorder.
- FISH tests – identifies copy number or rearrangement of targeted sequences.

Gene panel test:
(from 2 to several 100 genes) interrogates a set of genes associated with a particular phenotype or group of phenotypes to identify variations or mutations that can lead to a genetic disorder.

Exome:
studies all the genes known to cause genetically inherited diseases (clinical exome) or the whole coding region of the genome (whole exome) to identify variations or mutations that could be associated with a genetic disorder.

Whole genome:
- analysis of chromosomes or DNA for large genetic changes, such as an extra copy, deletion or rearrangement of all or part of a chromosome, that causes a genetic condition.
- whole genome sequencing (research) – to generate the complete genetic information for an individual; to look for associations and potential new variations relating to disease.

Laboratory education and training

Modernising Scientific Careers introduces training programmes at four main career levels for the healthcare science workforce.

- Associate/assistant – NVQs and foundation degrees (or equivalent) underpinned by an awards and qualifications framework
- Practitioner Training Programme (PTP) – undergraduate level
- Scientist Training Programme (STP) – postgraduate level, pre-registration training
- Higher specialist scientific training (HSST) – doctoral level, post registration training

This model could provide modular training packages for all or part of the curriculum either by enabling overseas staff to train in UK laboratories or by supporting this training in the host organisation.
Summary of services

Bespoke laboratory services can be tailored to your needs.

**Laboratory Services**
- Testing clinical samples and issuing clinical interpretation reports
- Assistance in the interpretation and reporting of results
- Remote Laboratory advice
- Clinical trials support
- Technology transfer
- Access to translational research in centres of excellence

**Teaching / Training**
- Shadowing Clinical Scientists and Healthcare practitioners
- Work-place secondments
- Accredited courses, seminars and conferences
- Focused module training

**Management**
- Consultancy services (setting up Laboratory Genetic services abroad)
The UK has a proud tradition of excellence in education within science, medicine and healthcare whether for clinical service or research and at undergraduate and postgraduate level.

This excellence underpins the quality of:

- provision of services
- research
- courses (taught and on-line)
- training opportunities for visitors to the UK & provided by UK staff abroad.

For service provision:

- UK NHS staff are subject to national professional registration,
- the UK ensures that staff competence and quality is maintained through a system of regular staff appraisal and revalidation

The UK therefore offers unparalleled training opportunities for all staff roles for clinical genetic services.

---

**Training opportunities in the UK:**

- Work placements
- Courses
- Conferences

**Academic support:**

- Curriculum planning
- Set examination standards
- Examine trainees (PhD, diplomas, counselling)

**Local training opportunities:**

- Support & mentoring
- Joint clinics
- Teaching (seminars, courses)

**Management:**

- Adapt UK based training model for training of scientists, counsellors and clinical geneticists locally

**e-based training:**

- Webinars
- e-link consultations
- Online courses and resources (guidelines)

---

www.ukgtn.nhs.uk
UK training facilitates Genetic Centres
to run as multidisciplinary teams

The case history here illustrates how a trained multidisciplinary professional team approach achieves the best for a patient and their family:

Mrs S, aged 30 years, is referred to the genetic service. Her father and grandfather both died from bowel cancer around age 50 years, and now a cousin aged 40 years has developed this.

The genetic counsellor takes a full family history, and Mrs S obtains consent from the cousin to access his medical information. This confirms colorectal cancer, but no genetic testing.

The cousin agrees to see the clinical geneticist, who notes that the bowel cancer has the characteristics for the hereditary type, discusses the familial implications with the cousin, and takes blood for DNA testing.

The Lab genetic scientist uses a UKGTN-approved gene panel test of 14 genes which can underlie hereditary colorectal cancer.

The results are not clear cut, as there are several different unknown variants, but the Bioinformatician evaluates these through multiple e-based resources, finding that only one variant is predicted to have a high likelihood of being pathogenic.

Mrs S is seen by the consultant and counsellor and offered DNA testing for this variant. She proves to have inherited this, and on consequent colonoscopy is found to have very early, but surgically resectable, bowel cancer.

Ten years later she remains well and continues to have two yearly bowel screening. Her daughter will be counselled about genetic testing when she is an adult.
Organisation of Education and Training

Undergraduate, postgraduate and ongoing education & training in each genetic profession is organised on a national basis, following an overall themed direction.

This is made possible through UK-run courses and conferences. Many of these are open to participants from outside the UK.

For substantive courses:
See www.educationuk.org/global/

<table>
<thead>
<tr>
<th>Course name</th>
<th>No. of UK academic institutions offering course, (and level):</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Undergraduate</td>
</tr>
<tr>
<td>‘Genetics’</td>
<td>32</td>
</tr>
<tr>
<td>‘Molecular Biology’</td>
<td>25</td>
</tr>
<tr>
<td>‘Molecular Genetics’</td>
<td>3</td>
</tr>
<tr>
<td>‘Genomics’</td>
<td>–</td>
</tr>
<tr>
<td>‘Bioinformatics’</td>
<td>–</td>
</tr>
<tr>
<td>‘Medical Genetics’/</td>
<td>–</td>
</tr>
<tr>
<td>‘Genetic Medicine’</td>
<td>–</td>
</tr>
<tr>
<td>‘Genetic Counselling’</td>
<td>–</td>
</tr>
</tbody>
</table>
National UK Professional Training:
Laboratory scientists & bioinformaticians:

Clinicians:
• For opportunities for international applicants: www.rcplondon.ac.uk/international/supporting-doctors-train-uk

For Genetic nurses/counsellors:
• Association of Genetic Nurses and Counsellors – www.agnc.org.uk

e-learning
Information about e-learning opportunities are available from:
• National School of Healthcare Science – www.nshcs.org.uk
• Genetics and Genomics Education Centre – www.geneticseducation.nhs.uk
• Health Education England Genomics Education Programme – www.genomicseducation.hee.nhs.uk

Case study: Prof AW from South Africa
Consultant medical geneticist and university associate professor
• attended 2014 BSGM on Clinical Genetic Society scholarship
• presented work on: sickle cell disease, and on hearing loss in Cameroon
• visited two UK genetic centres

His aims were:
• to improve his own expertise through exposure to UK clinical genetic services
• to establish networks to enhance the training capacity and opportunities which AW’s own centre can provide in the task of instigating genetic services across sub-Saharan Africa

Outcomes:
• He gained valuable experience in diagnostic dysmorphology
• He has a commitment to enable South African trainees to attend a UK Dysmorphology course
• He has been able to develop plans for collaborative molecular research in sickle cell disease

In his words:
“I’m grateful for this wonderful opportunity given by the CGS international scholarship, and wish to any willing international colleague a similar experience.”

Shorter courses and conferences
The British Society of Genetic Medicine (BSGM) holds an annual conference, which in 2015 is to be held jointly with ESHG (European Society of Human Genetics).

Some sponsored scholarships are available for visitors working in isolated situations in their own country to attend and present at the BSGM meeting.
Many other open short courses or conferences, are organised through the constituent societies of BSGM, affiliated organisations, or through individual UK centres.

For further details see: www.bsgm.org.uk or individual sites.

**Case example: The Manchester Dysmorphology course**

- Every 2 years in Manchester
- Trainees in Clinical Genetics
- Equips trainees with essential skills in recognising patterns of dysmorphology
- Course is affiliated with the European Society of Human Genetics
- Many international trainees
- Unique opportunity to:
  - meet international peers
  - establish links for future affiliations
  - form friendships sustainable throughout careers.
Appendix 1

Ethical Framework

All work undertaken by the NHS in the UK must meet published ethical standards and be in accordance with UK legislation. NHS providers work to the guidelines provided below to ensure the quality of patient care.

1. Direct-to-consumer testing
   Laboratories will only accept requests for gene tests from a healthcare professional who takes full responsibility for counselling the patient, explaining why the test is being done, obtaining informed consent and feeding back the results afterwards.

2. Predictive testing in children
   Laboratories will not accept samples from children for predictive testing for adult onset disorders, in line with international guidance.

3. Social sex selection
   Laboratories will not provide information about the sex of a pregnancy if they believe that the test is being done purely for social reasons and likely to lead to a termination of a pregnancy on the basis of the sex.

4. Training and qualifications
   The standards and competencies that clinical and laboratory staff from other countries are required to achieve in order to earn any qualification must be exactly the same as it is in the UK. It may be possible to develop tailor-made, short training packages that aim to provide someone with the targeted set of skills that they require for their job, without them needing to work towards a complete qualification. In these circumstances it may be appropriate to provide them with something in writing that explicitly states which skills they have acquired, and how these were obtained, while making it clear that this does not equate to any particular qualification.

5. Reimbursement
   The NHS is not a charity and must be fully reimbursed for every cost incurred.

6. Legislation
   Any work undertaken must be done in accordance with UK/European legislation and professional guidelines. This includes:
   - The Human Tissue Act 2004
     www.hta.gov.uk/legislationpoliciesandcodesofpractice/legislation/humantissueact.cfm
   - The Data Protection Act 1998

7. Consent
   It is a general legal and ethical principle that valid consent must be obtained before genetic testing can take place. This principle reflects the right of patients to determine what happens to their own bodies, and is a fundamental part of good practice. A healthcare professional (or other healthcare staff) who does not respect this principle may be liable both to legal action by the patient and to action by their professional body. Employing bodies may also be liable for the actions of their staff.

Author:
Professor Frances Flinter – Chair Medical Genetics Clinical Reference Group and Consultant Clinical Geneticist
Appendix 2

Membership of the UKGTN Task and Finish working group

Chair:
Dr Shehla Mohammed MD, FRCP – Consultant Clinical Geneticist, Head of Genetics Service Guys & St Thomas’ NHSFT, Clinical Advisor to UKGTN

Business and Policy advice:
Dr Mark Kroese – Public Health Advisor, UKGTN & Programme Director, PHG Foundation
Colin Pavelin – Health Science and Bioethics, Department of Health, England
Jeanette Thorpe – Business Manager, Department of Clinical Genetics, the General Infirmary at Leeds

Clinical advice:
Dr Tazeen Ashraf – Specialist Registrar, Guys & St Thomas’ Clinical Genetic Services
Dr Christine Patch – Consultant Genetic Counsellor/Manager, Department of Clinical Genetics, Guys Hospital

Scientific advice:
Professor Mike Griffiths – Director, West Midlands Regional Genetics Laboratory, Birmingham Women’s Hospital NHSFT
Darren Grafham – Head of Laboratory, Sheffield Children’s Hospital NHSFT
Dr Jo Whittaker – Scientific Development Advisor, UKGTN

Education and Training advice:
Dr Peter Lunt – National School of Healthcare Science

Healthcare UK team:
Professor Rory Shaw
Dr Aphrodite Spanou
Vicky Waite
Ronald Fraser

UKGTN project team:
Jacquie Westwood – Director
Jane Deller – Programme Manager

www.ukgtn.nhs.uk
The UKGTN established a task and finish group to produce this brochure and would like to thank members of this group who co-authored different sections. We are grateful for their guidance and advice in its development. The full membership is listed in appendix 2.

**Authors**
Why choose the UK to provide genomic services?
Jacquie Westwood & Jane Deller

Planning and Service Development
Jeanette Thorpe & Dr Mark Kroese

Clinical Services
Dr Shehla Mohammed, Dr Tazeen Ashraf & Dr Christine Patch

Laboratory Services
Professor Mike Griffiths, Dr Jo Whittaker, Darren Grafham

Education and Training
Dr Peter Lunt & Dr Tazeen Ashraf

Ethical Framework
Professor Frances Flinter
Please send enquiries to:
Healthcare UK
1 Victoria Street
London SW1H 0ET

Or email:
Healthcare.uk@ukti.gsi.gov.uk