NHS
Guy's and St Thomas'
NHS Foundation Trust

Visiting Professional Programme: Clinical Genetics

Introduction

The Guy's and St Thomas' NHS Foundation Trust Clinical Genetics Visiting Professional Programme (VPP) is designed to provide international visiting professionals with the opportunity to experience our highly acclaimed genetics department first hand. The programme is a unique chance to be fully integrated within a genetics service which is at the forefront of clinical activity, training and research; regional genetics clinics offer services to diagnose and assess the risk of families inheriting a genetic condition; the genetic counselling service involves explanation and risk assessment with any available options for family members, the genetics clinic is part of the Division of Medical and Molecular Genetics at King's College London and we also take part in the 10,000 Genomes Project which focuses on cancer and rare diseases.

Our Genetics department is divided into a range of specialist or multidisciplinary services and clinics including: Bardet Biedl Syndrome, BRCA family services, Breast cancer risk assessment, Connective tissue disorder, Dermatology (cancer), Dermatology (general), Endocrine, Eye genetics, Fetal cardiology, Fetal medicine unit (FMU), Sickle cell and thalassaemia, Haemophilia genetics, Huntington's disease, Inherited Cardiac Conditions, Metabolic, Myotonic dystrophy, Neurofibromatosis (NF1 and NF2), Paediatric neurology/genetics, Pre-implanation genetic diagnosis (PGD), Pre-natal diagnostic service, Rapid access, Skeletal dysplasia, Tay Sachs, Von Hippel Lindau (VHL) and Xeroderma Pigmentosa (XP). We offer specialised visiting professional programmes within all of our services for those that wish to hone their expertise in a subspecialty area of interest in genetics. Alternatively visitors may apply to rotate throughout our genetics department and we even offer the opportunity for visitors to gain professional certification depending of the duration of their stay.

The programmes are designed by our clinical leads to provide unique development experiences which enable visiting professionals to broaden their clinical expertise and learn new skills that can be transferred back to their work place. We are excited to deliver exclusive access to observe and work alongside our renowned genomics faculty and to provide an opportunity for international professionals to gain insight into our Trust and the role that genetics consultants play within the NHS.

Who Should Apply?

This programme is intended for non-U.K. trainee geneticists, specialist registrars and consultants. Visitors have the choice of applying for a hands-on clinical practical attachment or observership programme. Those wishing to apply for a practical attachment must have GMC registration. You can apply for GMC registration directly through the main GMC website (www.gmc-uk.org), alternatively The Royal College of Physicians run a medical training initiative which facilitates GMC registration and provides sponsorship for a Tier 5 visa. You can find out more about their Medical Training Initiative by visiting their website (www.rcplondon.ac.uk/education-practice/advice/medical-training-initiative).

Duration

We are able to offer visiting professional programmes for periods from 12 weeks up to one year. The programme's duration can be tailored to the specific learning requirements of those visiting the Trust.

Format

The Clinical Genetics programme is delivered within the hospital work place and it includes exposure to genetics clinics, inpatient ward rounds, multidisciplinary team meetings, and access to training courses depending on the length of programme that visitor's request.

Visiting professionals will be aligned with supervisors from within the clinical teams who will ensure that all of the learning and development needs and expectations are met. The supervisors will also ensure that visiting professionals are totally integrated in the day to day service within our genetics team, regardless of the speciality and duration that visitors apply for.

Overall Programme Aims

The programme is aimed at those who wish extend their clinical expertise in genetics, and who wish to learn more about the treatment pathways and specialist technology within Guy' and St Thomas' NHS Foundation Trust.

The programme aims to include:

- Direct experience and exposure to outpatient based services through our 27 specialist and multidisciplinary genetics clinics
- Participation in multidisciplinary team meetings
- Access to monthly lectures and internally organised courses
- Involvement in genetic counselling including diagnosis, testing and counselling with family members on risk assessments and available options (attachments only)
- Opportunities to receive bespoke training from our expert clinicians and to gain a professional certification
- The opportunity to create networks and close links for future learning and development through peer mentorship from junior and senior doctors
- Support of a small project/audit with an intention of presenting in international meetings or for publication
- Access to and the chance to actively contribute to clinical research with the Division of Medical and Molecular Genetics at King's College London
- Experience of the NHS and working in a genetics centre with advanced clinical and technical expertise amongst an internationally renowned faculty of experts.

Sample Weekly Clinics

	Monday		Tuesday		Wednesday		Thursday		Friday	
AM/PM	Type	Location	Type	Location	Type	Location	Type	Location	Type	Location
AM	General	Whitstable	General	Genetics	Derm	Genetics	General	Medway	General	Genetics
	General	Genetics	General	Genetics	Psych	Genetics	General	Medway	General	Genetics
	General	Genetics	General	Eastbourne	GEL	Genetics	General	Canterbury	General	Genetics
	General	Genetics	General	Genetics					General	Genetics
	General	Brighton	General	Genetics			General	Canterbury		
	General	Maidstone	General	Genetics			General	Hastings	Cancer	Maidstone
			General	Genetics			General	Brighton	Cancer	Maidstone
	Cancer	Cancer	General	Genetics			General	Canterbury	Cancer	Maidstone
	Cancer	Jersey					General	Brighton	Cancer	Genetics
	Cancer	Genetics					General	Hastings		
	Cancer	Genetics	Cancer	Genetics						
	Cancer	Brighton					Cancer	Genetics	CRAS	Kings
	Cancer	Brighton								
	Cancer	Brighton	CRAS	Lewisham			Cancer	Medway	Eye	Genetics
	Cancer	Cancer					Cancer	Endocrine	GEL	Genetics
			Endocrine	End MDT			Cancer	Genetics	Psych	Genetics
	GC	Jersey	PGD	Genetics						
	GC	Whitstable	PGD	Genetics			GC	Medway	HD	Faversham
	GC	Genetics	General	Genetics			GC	Genetics	General	Ashford
	GC	Whitstable	PGD	Genetics			GC	Genetics	General	Ashford
	GC	Hastings	PGD	Genetics						
							CTD	Cardiology	Mixed tel	Telephone
	PGD	Genetics					MMR MDT	Genetics		
	CRAS	Genetics								
	GEL	Genetics								
	Tay Sachs	Genetics								
	R&D	Genetics								
PM			General	Genetics	General	Genetics	Cancer	Genetics		
	Cancer	Genetics			Rapid access Gen	Genetics	General	Genetics		
	General	Genetics	Cancer	Genetics	Rapid access Cancer	Genetics				
	General	Maidstone								
			Mixed	Genetics						
			PGD	Genetics						
	GC	Genetics	GC	Genetics						
	GC	Genetics	GEL	Genetics						
	GC	Genetics	Mixed	Genetics						
			Mixed tel	Telephone						
	GC	Genetics	Mixed	Genetics						

Expert Faculty

Management team

- Dr Deborah Ruddy – Head of service
- Dr Muriel Holder – Deputy head of service (general genetics)
- Dr Anjana Kulkarni – Deputy head of service (cancer genetics)

Consultant clinical geneticists

Clinical geneticists or consultant geneticists are medical doctors with specialist experience in genetics.

- Dr Adam Shaw
- Dr Ana Beleza
- Dr Charu Deshpande
- Dr Dragana Josifova
- Dr Fiona Connell
- Dr Leema Robert
- Dr Louise Izatt
- Dr Melita Irving
- Dr Shehla Mohammed
- Professor Frances Flinter
- Professor Phil Beales

Specialist registrars

All clinical genetic specialist registrars are doctors who have undertaken several years of postgraduate training in general medicine or paediatrics and have completed their membership examinations (MRCP or MRCPCH). Many of our trainees also have higher research degrees.

Our trainees have the chance to experience each sub-speciality within genetics over a four-year training programme, during which they will lead clinical consultations, manage inpatient referrals, shadow consultants, attend multi-disciplinary meetings, deliver teaching, conduct audit and be involved in genetic research. The main areas of focus during the training programme are: paediatrics, dysmorphology, cancer genetics, cardiac genetics, neurogenetics and prenatal genetics.

- Dr Helena Carley (SpR – ST4)
- Mina Ryten (NIHR – fellow)
- Rachael L. Jones (maternity leave)
- Tazeen Ashraf (SpR – ST4)

Genetic counsellors

- Alison Lashwood* – consultant genetic counsellor and clinical lead in PGD
- Belinda Lotter – trainee genetic counsellor

- Cecilia Compton – genetic counsellor
- Charlotte Tomlinson* – genetic counsellor
- Dr Vishakha Tripathi* – consultant genetic counsellor in cancer genetics
- Eshika Haque* – genetic counsellor
- Genevieve Say* – genetic counsellor (maternity leave)
- Roberta Rizzi – locum genetic counsellor
- Rupinder Jassi* – genetic counsellor (maternity leave)
- Sally Watts* – lead genetic counsellor and manager
- Sara Levene* – principal genetic counsellor
- Sarah Rose* – genetic counsellor
- Sarah Ross – genetic counsellor

* indicates professional registration with the Genetic Counsellor Registration Board

Other staff

- Colin Evans – cardiac genetics nurse
- Dee Howley – cardiac genetics nurse
- Dr Clare Firth – clinical psychologist
- Gail Norbury – commissioning and governance director for genetics laboratories
- Jackie Aw Fong – cardiac genetics nurse
- Tootie Bueser – BHF cardiac genetic nurse

Lab staff

- Anne Bergbaum – Operations lead admin, H&S
- Dr David Cregeen – Operations lead Metabolic Genetics
- Dr David Ellis – Operations lead Monogenics
- Dr Deborah Ruddy – Clinical Lead
- Dr Kathy Mann – Operations lead prenatal genetics & reproductive medicine
- Dr Michael Yau – R&D Lead
- Dr Pam Renwick* – Operations lead PGD
- Dr Wook Ahn* – Operations lead Bioinformatics & developmental delay
- Elaine White – Office manager
- Gail Norbury – Commissioning and Governance Director
- Jeremy Skinner – Quality manager
- Marie Jackson – Scientific lead Biochemical Genetics
- Michael Neat – Scientific Cancer Lead
- Nicola Foot – Operations lead cancer genetics
- Prof Graham Taylor – Scientific Director
- Richard Hall – Service Delivery manager

Viapath *part trust

Genetics Service

The Guys and St Thomas' NHS Foundation Trust's Regional Genetics service serves a population of 4.9 million in South East England and aims to provide a comprehensive and integrated clinical and laboratory service. As one of the largest genetics services in the UK and a member of SEEGEN, (South East of England Genetics Network), our focus remains on delivering and continuing innovative models to enhance patient care. We remain committed in developing a responsive and flexible service.

We also keen to ensure that patients benefit from our strong track record in new gene discoveries, translational research and emerging treatment modalities for patients with both rare and more common genetic disorders.

We retain a strong ethos in both undergraduate and postgraduate teaching and training. Over the past few years we have solidified our links with a number of specialities as genetics moves into mainstream medicine.

The service is part of the GRIDA directorate (Genetics, Rheumatology, Infection, Dermatology and Allergy) which comprises 395 whole time equivalent staff across both the Guy's and St. Thomas' campuses. The department provides large ambulatory services for severe chronic inflammatory disease of the skin and joints, musculoskeletal services, complex connective tissue disorders such as lupus, complex skin cancer, clinical diagnostic services for infectious diseases, the regional clinical genetics service and clinical care for sexual health including HIV. The directorate is also a key provider of clinical diagnostic specialist laboratory services within GSTS, including the areas of virology, microbiology, dermapathology and genetics.

Clinical Genetics is the medical specialty which provides a diagnostic service and "genetic counselling" for individuals or families with, or at risk of, conditions which may have a genetic basis. Genetic disorders can affect any body system and any age group. The aim of Genetic Services is to help those affected by, or at risk of, a genetic disorder to live and reproduce as normally as possible.

Genetic disorders include:

- Chromosomal abnormalities, which cause birth defects, mental retardation and/or reproductive problems.
- Single gene disorders such as cystic fibrosis, muscular dystrophy, Huntington's disease and sickle cell disease.
- Familial cancer and cancer-prone syndromes such as inherited breast or colorectal cancer and neurofibromatosis.
- Birth defects with a genetic component such as neural tube defects and cleft lip and palate.

In addition a large number of individuals with birth defects and/or learning disabilities are referred and investigated for genetic factors. Individuals identified through childhood or pregnancy screening programmes also require genetic services. In the future, as the genetic contributions to common later-onset disorders such as diabetes and coronary heart disease are identified, genetic services may be required for those at high risk. Testing for genetic factors that affect drug prescribing will also increasingly become an important activity.

What are Regional Genetics Services?

Specialist genetic services have developed in the UK largely as regional centres of expertise. All support the network of Regional Genetics Centres and the need for close functional interaction between centres and between elements of the service within an individual centre. Specialist genetic services can be distinguished from other medical services by the fact that they deal with families, often over several generations, and that they can provide genetic expertise for any age group affected by, or at risk of, disorders in any body system.

The clinical team includes Clinical Geneticists (medical doctors who increasingly may have subspecialty expertise), junior doctors in training, Genetic Counsellors (sometimes referred to as genetic associates or genetic co-workers) and, in some centres, genetic family register staff. Services are delivered in clinics in the regional centre, in outreach clinics in district general hospitals, in ward or hospital department consultations.

Following referral to the service for most patients, a clinic appointment with a member of the clinical team is organised (and any follow up appointments as necessary) to allow examination, investigations, diagnosis and further management. A detailed summary letter is sent to the family and to hospital and primary care doctors. In some situations the genetics team maintains long-term contact with the family. Services are offered to the extended family as necessary.

Referrals as appropriate are triaged for mainstreamed genetic services such as the haemoglobinopathy service or the familial hypercholesterolemia service.

Most regional genetics services have developed areas of specialist expertise in areas of mainstream medicine to deliver coordinated care for patients. Specialist services provide both diagnostic and management services for patients with rare conditions with regular multidisciplinary meetings. Some specialist services also maintain patient registers and databases for ongoing care of patients.

Genetic counselling

Because diagnosis and testing will often have an impact on the wider family, the genetic counselling process includes an explanation and risk assessment with any available options for family members.

It can take time to adjust to new diagnoses and to decide on the best possible course of action. Our team provides up-to-date and unbiased information. We ask for an update when the affected person and family members reach reproductive age, and before a new pregnancy. Testing, risk-reducing treatment and research options change over time.

Commitment to research

The genetics clinic is part of the Division of Medical and Molecular Genetics at King’s College London. We are committed to genetic research and academic excellence.

100,000 Genomes Project

The South London Genomic Medicine Centre (GMC), hosted by Guy's and St Thomas', is one of the centres taking part in the 100,000 Genomes Project.

The national programme will focus on cancer and rare diseases and will enable pioneering research to decode 100,000 human genomes, a scale not seen anywhere else in the world.

Genetics clinics

The specialist clinical genetic service provides services to the South East of England south of the Thames, a population of approximately 4.7 million. It is provided by Guys and St Thomas' NHS Foundation Trust located on the Guys hospital site. Most clinics are based centrally at guys Hospital for patients referred by clinicians in this area.

Peripheral Clinics: The service holds a number of clinics in local DGH's and Child Development clinics. These peripheral clinics aim to improve access to genetic services and provide patients and families a choice of location for a genetics assessment.

Outreach Clinics: The monthly Outreach Clinics in Whitstable, Hastings and Brighton remain very popular with our patients and we have high demand for appointments at these venues. We have reconfigured the clinic to ensure most appropriate utilisation of available slots. We have negotiated rooms in health centres allied to GP practices enabling us to see patients of all ages in a local site more convenient to them.

The genetics service also runs specialised subspeciality services including:

- Cancer
- Cardiac
- Fetal medicine
- PGD
- Skeletal dysplasia
- NCG funded services

The Genetics Service runs a range of specialist or multidisciplinary clinics, held at Guy's and St Thomas' and Evelina London Children's Hospital, including:

- Cancer Dermatology Clinic
- Dermatology (general)
- Endocrine genetics
- Eye genetics
- Metabolic clinic
- Paediatric neurology/genetics
- Rapid access clinic for pregnant patients
- Tay Sachs
- Von Hippel Lindau (VHL)

Specialist Services

Biochemical Genetics Laboratory

The biochemical genetics laboratory, established in 1973, offers a regional, and in some cases national, specialist service for the diagnosis of over 50 genetic biochemical disorders.

It collaborates closely with clinical, biochemical and genetics services at Guy's and St Thomas' Trust and Viapath to facilitate the diagnostic pathway.

The laboratory has a special interest in lysosomal storage disorders, particularly mucopolysaccharidosis, but also carries out testing for disorders of galactose metabolism, urea cycle disorders, amino/organic acid disorders, and peroxisomal disorders. Prenatal diagnosis is available for many of these disorders, and we regularly test samples from around the UK and overseas.

The laboratory is also a national centre for Tay-Sachs disease carrier screening in the Ashkenazi Jewish population. Testing is carried out by a combination of enzymology, metabolite analysis (e.g. urinary glycosaminoglycans) and molecular genetic testing. The Biochemical Genetics laboratory is UKAS accredited medical laboratory No 8688, part of the Supra-Regional Assay Service (SAS) and a member of the UK Genetic Testing Network (UKGTN). Tissue culture facilities are a vital component of the biochemical genetics service and many cell lines are referred to other specialist centres in the UK or abroad for rare disorders.

The biochemical genetics laboratory carries out the following tests:

- Urine analysis: glycosaminoglycans (mucopolysaccharides) and oligosaccharides.
- Galactose metabolism: galactose-1-phosphate uridyl transferase (classical galactosaemia), galactokinase.
- Mucopolysaccharidosis: All types (Hurler, Scheie, Hunter, Sanfilippo, Morquio, Maroteaux-Lamy, Sly).
- Glycoprotein disorders: Aspartylglucosaminuria, Schindler, α & β-mannosidosis, Fucosidosis, Mucolipidosis / I-cell, sialidosis.
- Sphingolipidoses: Tay-Sachs, Sandhoff, GM1 & GM2-gangliosidosis, metachromatic & Krabbe leucodystrophy, Fabry, Gaucher, Niemann-Pick A/B/C, Wolman, cholesterol ester storage (LIPA deficiency).
- Neuronal ceroid lipofuscinosis.
- Peroxisomal disorders.
- Other: Citrullinaemia, argininosuccinic aciduria, ornithine transcarbamylase, carbamyl phosphate synthetase, propionic acidaemia, pyruvate carboxylase, maple syrup urine disease, Pompe disease (glycogen storage disease type 2).
- DNA Tests: ACADM (MCAD), ARSA, CTSA, CTSD, DHCR7, GALC, GALT, GALK1, GALE, GLB1, GCDH, GLA, GM2A, HADHA (LCHAD), IVA, LIPA, SMPD1, PC.

BRCA Family Service

The Guy's BRCA Family Service (BRCAFS) was established to develop a streamlined model of multidisciplinary care and management of families with mutations in BRCA1 and BRCA2. The primary aims are to: Offer every BRCA1/2 mutation carrier in our catchment a clinical review, Devise an individually tailored counselling and risk management strategy, Address any concerns or worries individuals and their families might have, Update BRCA1/2 mutation carriers on the newest clinical developments by discussing appropriate surveillance and risk-reducing options, Ensure that the chosen surveillance and risk-reducing options are implemented, Offer BRCA1/2 mutation carriers participation in research trials, Offer clinical and peer-led psychosocial support

The service consists of five core elements: A multidisciplinary BRCA clinic, Patient register, Patient information update days, Patient newsletter, Facilitated peer support groups for female carriers All patients with a pathogenic BRCA1 and BRCA2 mutation identified through our service are referred to the BRCAFS.

A multidisciplinary meeting during the clinic ensures patients are given consistent advice based on national guidelines and published evidence. Patients with a cancer diagnosis are able to access advice which contexualises their age-related risks of a further BRCA-related primary cancer in comparison to their competing risks based on current disease prognosis. This model is publicly funded through the National Health Service (NHS) as are all the screening and risk reducing surgery options.

BRCA support groups allow for access to facilitated peer support for patients and run three monthly at our sites at Guy's, Maidstone and Brighton. The groups are facilitated by family history nurses and genetic counsellors and oversight is provided by the BRCA steering committee that has three patient representatives who also attend the support groups.

The BRCAFS currently looks after approximately 1600 individuals with a BRCA1 or BRCA2 mutation and is the largest patient group within the cancer genetics service.

Cancer

Cancer is reported to affect around 1 in 2 individuals in their lifetimes in the United Kingdom and family history may be contributory in around a fifth of cases. Up to 5% of all cancers may result from a high or moderate penetrance mutation in a cancer predisposition gene, but most probably go unrecognised. The Cancer Genetics Service at Guy's Clinical Genetics department sees individuals with or at risk of hereditary cancer and serves a population of approaching 5 million, across Southeast London, Kent and East Sussex.

In addition to daily clinics at Guy's hospital, the service provides regular outreach clinics for Brighton, Maidstone, Hastings, Whitstable and Jersey.

Cancer Family History Clinics

The cancer family history clinics provide access to cancer risk assessment for a large number of patients in the region. These clinics are run by Clinical Nurse Specialists in the local hospitals and supervised by genetic counsellors. Training, supervising and overseeing these clinics generates a great deal of work for the Genetic Counsellors but is extremely important in ensuring high quality referrals and close links with local cancer teams. There is a clear protocol for the training, supervision and administration of these nurses and family history clinics.

Cardiac

Inherited cardiac conditions are a common cause of preventable ill-health and sudden death, often in young people. Inherited cardiovascular conditions (ICCs) are a group of genetic disorders that primarily affect the heart, including cardiac electrical systems and blood vessels. Sometimes, the first indication that an ICC is present in a family is the sudden and unexpected death of an apparently healthy child, teenager or young adult - other relatives may also be at risk. Recent advances in genetics and cardiology are now making it possible for health professionals to offer much more accurate diagnosis, improved clinical management and better risk assessment for families affected by ICCs - avoiding tragic and unnecessary deaths

Our ICC service is one of the largest in the UK and we cover all aspects of inherited cardiac diseases including aortopathies, neuromuscular disease, cardiomyopathies and rare metabolic disorders for both adult and paediatric patients. The service was originally set up in 2008 with a single monthly clinic. It has subsequently grown to become one of the largest services in the UK. The patients and their families are often very complicated with a lot of emotional and psychological issues as well as complex medical problems. Currently, we see over 5000 patients a year in our service.

The service covers the following patient groups:

1. Patients with cardiomyopathy (hypertrophic, dilated, restrictive and right ventricular)
2. Patients with inherited arrhythmia syndromes (long QT, Brugada, CPVT etc)
3. Patients with aortopathy (e.g. Marfan and connective tissue disorders, vascular Ehler Danlos syndrome and non-syndromic familial aortopathies)
4. Family screening for patients at risk of inherited disease
5. Patients with neuromuscular disease and cardiac involvement (e.g. Muscular dystrophy, myotonic dystrophy)
6. Patients with metabolic disease (e.g. Fabry's disease, mitochondrial disease)

Other rarer syndromes

Patients are usually seen in dedicated ICC or CTD/ aortopathy clinics for coordination of care with the support of cardiac genetic nurses, cardiologists, electrophysiologists and cardiovascular geneticists.

Connective Tissue Disorder and Aortopathy Service (CTD and aortopathy)

Our CTD and aortopathy service has developed in the last decade with geneticists working closely with all members of the cardiovascular department, this includes the paediatric cardiology service, adult congenital heart disease (ACHD), obstetric service and adult and paediatric surgical specialities to provide comprehensive patient care. In addition to CTD and aortopathy clinics, we also provide specialist joint cardiac obstetric clinics and obstetric care. .

We offer one stop diagnostic clinics for patients that have suspected inherited arteriopathies or inherited connective tissue disorders, including Marfan syndrome and Loeys Dietz syndrome. Ongoing follow up and medical management of patients is done in liaison with our paediatric and ACHD colleagues in dedicated aortopathy follow up clinics. Together with our interventional, surgical and electrophysiology colleagues, we also provide a comprehensive range of procedures including complex congenital cardiac surgical repairs, catheter intervention and hybrid procedures.

FMU

Fetal Medicine (input from Obstetrics, Fetal Medicine and Genetics): This is coordinated by Dr Muriel Holder, with additional input from Dr Ana Beleza and a team of dedicated genetic counsellors (Alison Lashwood, Charlotte Tomlinson). Close communication between the group and the fetal medicine unit (FMU) at St Thomas' Hospital allows rapid, appropriate assessment and monitoring of women whose fetus is at risk of a genetic condition or congenital anomaly syndrome. There are two regular multidisciplinary fetal medicine meetings. One is held weekly and one is held monthly. Both are attended by the team, allowing discussion of the cases with which we are (or have been/will be) involved. Fetal medicine clinics with cases for the genetics team are held throughout the week, but largely concentrated on Tuesdays and Thursdays, after these meetings. The weekly **Rapid Access Clinic** is held in the Department of Clinical Genetics, where there is further input from the extended genetic counselling team, Consultants and the specialist trainees. A network of FMUs around the region is established to facilitate advising other specialists and providing

genetics support. Close links and similar practices are currently in place in Medway. The group is active in research studies conducted jointly between the FMUs and Clinical Genetics.

HD Service

Huntington Disease (HD) multidisciplinary clinic (input from neurology, neuropsychology, psychiatry, genetics and the Huntington Disease Association): The multi-disciplinary HD service is delivered via a multidisciplinary clinic which is run monthly at Guys hospital, and at an outreach clinic (in the Kent area) – for patients who are less able to travel, twice per year. As part of the HD service, patients are invited to take part in both observational studies and in commercial disease modifying drug trials.

Inherited cardiac conditions (ICC) Service

A multidisciplinary team of clinicians run five dedicated clinic for the care of patients with cardiomyopathies and inherited heart rhythm conditions such as long QT syndrome, brugada syndrome and catecholaminergic polymorphic ventricular tachycardia. We treat and manage these patients through inherited cardiac condition clinics (ICC), inherited heart muscle condition clinics, inherited cardiac arrhythmias clinics and neuromuscular conditions clinics. The clinics are supported by access to a range of testing services and facilities, such as an advanced echocardiography service and cardiovascular imaging service.

Routine monthly MDTs supported by clinicians in local DGHs are well established. These MDTs are used for seeking expert opinions and planning coordinated care of these patients in line with national published guidance.

Mismatch Repair Deficiency Service

The integrated mismatch repair deficiency testing service for Lynch Syndrome and hereditary bowel cancer was set up in 2010 as collaboration between clinical genetics, cellular pathology, colorectal surgery, medical oncology and the molecular genetics laboratory. The purpose is to improve diagnostic efficiency, cost effectiveness and management of patients and families suspected of having a mismatch repair defect.

Cases are referred by the Clinical Genetics or Histopathology departments according to established criteria. The risk assessment includes reviewing histology reports, obtaining a family history, confirmation of diagnoses in relatives, requesting tumour blocks where appropriate and testing according to prior risk using defined testing algorithms. The results are reviewed at the monthly multidisciplinary team meeting to agree interpretation of results, need for any further investigation and recommendations for cancer surveillance. To date we have issued over 1000 reports.

We continually monitor our service and have an iterative approach to our algorithms in line with the rapid technical and clinical developments. Outcomes include a more standardised, cross-discipline service for patients, awareness of when further investigations would alter the surveillance and management of the family, closer monitoring of diagnostic yield and improved through-put.

Molecular Genetics Service

Our laboratory provides a clinical molecular genetics service for a range of genetic disorders. We are a UKAS accredited medical laboratory (No 8688) and a member of the UK Genetic Testing Network (UKGTN).

Referrals are generally made via the UKGTN and testing for some specific diseases is commissioned by the National Commissioning Group for Genetics. However referrals are accepted from healthcare professionals from around the world. If the test is not available locally, the DNA is extracted and forwarded to the appropriate laboratory that does offer the test.

We have a history of introducing new technologies that improve the genetic testing service in collaboration with the Guy's and St Thomas' Trust (GSTT) Clinical Genetics centre and other healthcare partners.

Genetic testing is provided by DNA sequencing (Sanger and Next Generation), MLPA, Genotyping and RNA analysis as indicated.

Aliquots of DNA are stored for future use whenever possible. Samples are also accepted for DNA banking where analysis is currently not available, but where this may become possible in the future.

The laboratory offers testing on a range of disease and specialises in testing for:

- Duchenne and Becker Muscular Dystrophy
- Alport Syndrome
- Congenital Muscular Dystrophy
- Congenital Myopathy
- Skeletal Dysplasia

In addition, the laboratory provides testing that contributes towards specific multidisciplinary services such as for Lynch Syndrome, Glycogen Storage Disease and Nucleotide Excision Repair.

Guy’s and St Thomas’ NHS Foundation Trust Genetics Department currently provides DNA tests for the following disorders:

- Alport Syndrome
- Alzheimer disease, familial, type 3
- Amyotrophic Lateral Sclerosis type1 (motor neurone disease)
- Breast/ovarian cancer
- Congenital muscular dystrophy type 1A (merosin deficient)
- Congenital muscular dystrophy type 1C
- Congenital muscular dystrophy (Large-related) type 1D
- Congenital myopathies
- Duchenne/Becker muscular dystrophy
- Familial dysautonomia (Riley Day syndrome)
- Fanconi Anaemia
- Fragile X syndrome
- Fukuyama congenital muscular dystrophy
- Huntington disease
- Muscle Eye Brain disorder
- Primary Pulmonary Hypertension
- Rigid spine syndrome
- Spastic paraplegia, autosomal dominant
- Spinal Muscular atrophy
- Spinal Muscular atrophy with respiratory distress

- Ullrich congenital muscular dystrophy
- Walker-Warburg syndrome
- X-linked Infantile Spasm syndrome
- X-linked VACTERL with Hydrocephalus syndrome
- Other disorders

Multi-disciplinary

We participate in an increasing number of multi-disciplinary clinics which include the following:

A **Dental Genetics Clinic** is held every three months, providing a multidisciplinary assessment and management planning service for children with developmental dental anomalies, with input from clinical genetics (Melita Irving), Dr Mike Harrison (paediatric dentist) and Prof Martyn Coubourne (orthodontics). Development of this collaboration in clinical service and academic research is supported by a Biomedical Research Centre grant.

Dermatology Genodermatoses Clinic (input from dermatology and genetics): This monthly clinic provides a regional diagnostic and follow-up service for patients with skin malformations and cancer predisposition. Patients with a wide range of conditions which predispose to benign or malignant skin tumours (e.g. Gorlin Syndrome, Ferguson-Smith disease, Hereditary Leiomyomatosis and Renal Cell Carcinoma, Birt-Hogg-Dube, Muir-Torre and Familial Atypical Mole and Melanoma syndrome) are seen in this clinic. Clinical expertise is provided by Dr Anju Kulkarni (Consultant Clinical Geneticist) and Dr Katie Lacy (Consultant Dermatologist).

Endocrine Genetics Clinic (input from endocrinology and genetics): This monthly clinic provides a regional diagnostic and follow-up service for patients with a wide range of genetic conditions which predispose to endocrine tumours (e.g. Multiple Endocrine Neoplasia, Hyperparathyroidism-Jaw Tumour Syndrome, Hereditary Paraganglioma/Phaeochromocytoma syndrome). Clinical expertise is provided by Dr Louise Izatt (Consultant Clinical Geneticist), Dr Ben Whitelaw (Consultant Endocrinologist) and Sarah Rose (Genetic Counsellor).

Genetics Skin Clinic (input from dermatology and genetics): Dr Holder runs a joint clinic/MDT meeting every 2 months with Pr Jemima Mellerio (Consultant Dermatologist). This clinic provides a regional diagnostic and follow-up service for patients with dermatological/genetic conditions. There is also an MDT meeting attended by the dermatology clinic, Dr Muriel Holder from the Clinical Genetics team, the plastic surgery team and the pathology team every month to discuss cases seen in dermatology, which require further input from different specialists.

Metabolic-Genetics Family Clinic (input from Paediatric and Adult Inherited Metabolic Disease and Genetics): This fortnightly clinic held at St Thomas' Hospital. The clinical genetics input is provided by Dr Charulata Deshpande, and the adult and paediatric metabolic teams. The clinic is dedicated to adults with an inherited metabolic disease for routine follow-up, genetic counselling and also arranging appropriate genetic tests including prenatal tests. The clinic also sees patients with complex metabolic problems for a joint diagnostic assessment and arranging appropriate investigations. Additional appointments are made available for joint consultation with colleagues in Paediatric Metabolic Medicine as well as in patient assessment. In addition, there is a monthly multidisciplinary meeting involving all clinicians and the Biochemical Genetics labs to discuss the patients, results of investigations and ongoing management and also opportunities for service development. There is an increasing demand from families for one-stop service to arrange all the necessary surveillance including radiological investigations so that the care is coordinated at the specialist centre and there are plans to formalise this service in the near future.

Neurogenetics (input from Paediatrics and Genetics): The paediatric neurogenetics clinic runs on a regular basis with input from Dr Elizabeth Wraige, Paediatric Neurologist, Dr Shehla Mohammed, and physiotherapy support as appropriate. The clinic sees patients with complex neuromuscular/genetic conditions.

Ophthalmology and Genetics: The multidisciplinary genetics eye clinic has continued to operate successfully, on a monthly basis. It now has a well established team of two consultants (clinical geneticist and ophthalmologist) and a dedicated genetic counsellor. It offers approximately 60-70 appointments per year and provides diagnosis and counselling for patients and families affected or at risk genetic ophthalmological conditions. When required, further counselling sessions can be arranged with our genetic counsellor, especially for individuals with newly diagnosed conditions associated with poor prognosis for their vision.

Vascular Skin Clinic (input from dermatology, plastic surgery, radiology and genetics): A joint clinic is occurring every two months with Dr Carsten Flohr (Consultant Dermatologist), Miss Aina Greig (Plastic surgeon), the interventional radiology team and Dr Muriel Holder (Consultant Geneticist). This clinic provides a regional diagnostic and follow-up service for patients with vascular dermatological/genetic conditions. There is also an MDT meeting attended by the dermatology clinic, Dr Muriel Holder from the Clinical Genetics team, the plastic surgery team and the pathology team every month to discuss cases seen in this clinic, which require further input from different specialists.

Von Hippel Lindau (VHL) Disease (input from Genetics, Nephrology, Neurology, Ophthalmology, Urology, Endocrinology, Radiology and Neurosurgery): The clinic is staffed by Dr Deborah Ruddy, Sally Watts (senior genetic counsellor), and Sarah Rose (genetic counsellor). The multidisciplinary VHL screening service is run via a clinic which is run four times per year, and provides a comprehensive follow-up service for a large cohort of families affected by Von Hippel Lindau Disease. 60 families are now known to the VHL service.

NCG funded service

We either host, or are actively contributing to, a number of NCG (National Specialist Commissioning Group) funded multidisciplinary clinics. These include:

- **Bardet Biedl syndrome (BBS) clinic:** BBS is multisystem disorder, generally associated with developmental difficulties and with renal, endocrine and retinal involvement leading to reduced visual acuity/blindness and often chronic renal failure. Patients have very complex needs and associated morbidity. The clinics are overseen by Dr Shehla Mohammed and Professor Phil Beales.
  Now in its third year, the clinic is run in close collaboration with the LMBBS Society (patient support group) and the Guy's Genetics Department. The Guy's clinic sees only adult patients with children being seen at Great Ormond Street Hospital. Birmingham Children's Hospital is a partner in NCG service and undertakes both paediatric and adult clinics on site. We also continue to receive a significant proportion of new referrals from Moorfields Eye Hospital, where ophthalmologists are aware of the existence of the clinics. More recently, we have received a number of new referrals from GPs and clinical genetics, renal, endocrine or paediatric consultants nationwide as awareness of the service has been raised.
- **Xeroderma pigmentosa (XP):** We also contribute actively to the Xeroderma Pigmentosa (PG) service with monthly adult, paediatric and transition clinics being held at St Thomas' (Dr Shehla Mohammed). Clinics are held at St John's Institute of Dermatology (at St Thomas' Hospital) which opened in 1863. It is now one of the world's leading centres for patients with skin disease and the largest clinical dermatology department in the UK and an internationally recognised centre of research into skin diseases. XP is a rare autosomal recessive disorder of DNA repair, characterised by photosensitivity leading to exaggerated and dramatic sunburn reactions, progressive pigmentary change, increasing lentigines, and a very high incidence of UV induced skin and mucous membrane cancer. There is 10,000 fold increased risk of multiple skin cancer as well as ocular and neurological disease.
  The model of care is unique with shared care in collaboration with local dermatology services, providing highly specialist advice as part of a coordinated clinical network to optimise clinical outcomes.

- **Neurofibromatosis-2 Service:** Guy's & St Thomas' is one of only four nationally commissioned centres in the UK for the management of patients with Neurofibromatosis type 2. The multidisciplinary team includes input from Neurology, Neurosurgery, ENT & Skull-base surgery, Ophthalmology, Clinical Genetics, Neuroradiology, Oncology, Audiology, Physiotherapy, and Psychiatry.

Preimplantation Genetic Diagnosis (PGD)

Preimplantation genetic diagnosis (PGD) is available to couples that are at risk of having a child with a specific genetic or chromosome disorder.

It involves the use of assisted reproductive technology (ART), which is also offered to couples with fertility problems. The aim is to obtain and fertilise a number of eggs. Once fertilised, the embryos develop for six days and then a number of cells are removed from each embryo. The genetic material (DNA or chromosomes) within each cell is then tested for the genetic or chromosome abnormality. One unaffected embryo is then transferred to the uterus with the hope that it will implant and form a pregnancy. If successful, the baby should not be affected by the disorder it was tested for.

Guy's and St Thomas' Centre for Preimplantation Genetic Diagnosis leads the field of fertility and Preimplantation Genetic Diagnosis (PGD), with an international reputation for excellence. The centre also offers NHS PGD treatment provided couples meet the criteria for funding. The expertise of the prenatal and clinical genetics teams give patients access to the best advice and the opportunity of talking through the diagnostic and treatment options available to them.

PGD multidisciplinary meetings are held each week to discuss ongoing clinical cases. Patients must be referred by a clinical geneticist for PGD and, provided PGD is suitable, will be offered an appointment within 8 weeks of referral.

Skeletal

Skeletal Dysplasia Service: This multidisciplinary, tertiary referral service is coordinated by Dr Melita Irving and Dr Moira Cheung through the Evelina London Children's Hospital. For patients who would benefit from a combined approach, the multidisciplinary skeletal dysplasia clinic is held once a month and attended by Dr Moira Cheung, paediatric endocrinologist, Miss Elizabeth Ashby, paediatric orthopaedic specialist, Ms Jill Massey, specialist OT, Miss Katie Phillips, specialist physiotherapist, and Melita Irving - with a pre-clinic meeting chaired by Dr Lloyd. This clinic provides the opportunity for diagnostic discussion, management planning and research, the latter supported by the local collaborative group and the Pan Thames Skeletal Dysplasia Group, meetings of which are held at Guy's twice a year. This has produced high quality research, published in a peer-reviewed journal with high impact factor, with the support of the Biomedical Research Centre genomics facility. The clinical service is supported by a molecular service, offering whole exome sequencing, as well as targeted gene testing, e.g. for achondroplasia. It is run by Viapath and accepts samples from and providing advice for patients from around the UK.

The dedicated **Achondroplasia clinic** runs twice a month at the Evelina London Children's Hospital. It offers a review and anticipatory surveillance service for children with achondroplasia and complex hypochondroplasia. It is run with input from paediatric endocrinology, paediatric respiratory paediatrics and specialist occupational therapy and physiotherapy, with additional input from a wider range of specialists dependent upon onward referral indications (e.g. ENT, orthopaedics, orthodontics). From this clinic, patients are being recruited to commercial students, sponsored by BioMarin, with recruitment to a phase 3 trial planned for spring 2017.

SLaM

SLaM provides clinical services in seven London boroughs with a combined population of nearly two million people, delivering mental health and substance misuse services for people living in the London boroughs of Croydon, Lambeth, Lewisham and Southwark. SLaM provides in-patient care at the Bethlem Royal Hospital, Maudsley Hospital and Lambeth Hospital in addition to over 180 community base sites across London. We run a monthly joint psychiatry/genetics clinic (at the Maudsley hospital) for patients who are suspect of having a syndromic psychiatric diagnosis.

Fees

A course fee per week will be applied. This excludes accommodation and travelling expenses. Participants will be provided with a certificate at the end of their attachment.

Registration and enquiries

To register for the Clinical Genetics Visiting Professional Programme please complete the application form at www.guysandstthomasevents.co.uk and return to:

Events

Guy's and St Thomas' NHS Foundation Trust

vpp@gstt.nhs.uk

Telephone: + 44 (0)20 7188 7188 extension 55865

Guy's and St Thomas' NHS Foundation Trust

Guy's and St Thomas' NHS Foundation Trust is one of the largest Foundation Trusts' in the UK. It consists of St Thomas' Hospital, Evelina London Children's Hospital and Guy's Hospital.

The Trust provides a full range of hospital services, as well as specialist services including cancer, cardiothoracic, women and children's services, kidney care and orthopaedics. Guy's is a major centre for cancer and renal services with the UK's largest kidney donor programme, and is also a leading centre for genetics, stem cell and allergy research and cleft lip and palate. St Thomas' is a leading centre for the treatment of cardiovascular disease, stroke, HIV and dermatology.

The Trust has one of the largest critical care units in the UK and one of the busiest A&E departments in London.

It has an annual turnover of £1.2 billion and employs 13,500 staff.

Last year, the Trust handled over 2 million patient contacts, including:

- 1.07m outpatients
- 85,000 inpatients
- 88,000 day case patients
- 192,000 accident and emergency attendances
- 859,000 in community services
- 6,847 babies delivered

The Trust has 665 beds at St Thomas', 288 at Guy's, 144 at the Evelina London Children's Hospital and 64 in the community.

NHS statistics show that our patient survival rates are nearly 25 per cent better than the national average. This is one of the lowest standardised mortality rates in the NHS and provides an important indication of the quality of care provided by our clinical staff.

In 2013, the Dr Foster Hospital Guide awarded us Trust of the Year for safe care.

Care Quality Commission (CQC) rating

Guy's and St Thomas' achieved a 'Good' overall rating by the Care Quality Commission (CQC) with several services rated 'Outstanding'.

CQC inspectors visited the Trust's hospital and community services from 7-10 September 2015 with further unannounced visits taking place later the same month.

In its report, the CQC praises staff who its inspectors found to be 'highly committed to the Trust and delivering high quality patient care'.

Guy's and St Thomas' NHS Foundation Trust values

Our values help us to define and develop our culture – what we do and how we do it.

Making people aware of the Trust values, making them part of our culture and demonstrating them through the organisation helps us develop a shared way of acting.

Our values are a key part of everything that we do as an organisation, from being included in staff job descriptions and in our publications, through to our work on leadership development and performance management. Our five values are listed below:

1. Put patients first
2. Take pride in what we do
3. Respect others
4. Strive to be the best
5. Act with Integrity

Every day we aim to provide the most exceptional care to our patients from dedicated staff who follow our five values. Take a look at our video at http://gti/services/organisational-devt/values-behaviours/our-values.aspx to see what our values mean to them.