Job description: Specialist registrar in Clinical Genetics
(Pan-Thames Region)

The Clinical Genetics Programme is run in conjunction with the London Deanery of Postgraduate Medical and Dental Education; the Pan-Thames Speciality Training Committee oversees training.

The Pan-Thames Clinical Genetics Training Scheme offers a comprehensive and structured training programme for Specialist Registrars (SpRs). There are four base training units within the programme, each of which can provide a fully comprehensive package of Training in accordance with the requirements laid down by the Royal College of Physicians. Each training unit also includes experience at peripheral hospitals as part of the training programme.

Pan-Thames secondment for Specialist Registrars in Clinical Genetics

The training opportunities in clinical genetics in London are now advertised as Pan-Thames positions. Each centre offers a general genetics service but also specialises in specific areas (see appendix). The secondment of trainees to other genetic centres maintains a broad experience in general genetics but allows flexibility for movement between the centres for experience in different areas of expertise.

Candidates apply for a SpR training opportunity attached to a designated centre. All four centres are represented at the appointments committee (as far as possible). Candidates who defer taking up their post may not be able to be accommodated at a specific centre when they return to the programme.

The SpR will spend an initial period of half of their training time at the centre to which they were appointed (Between 18 and 24 months depending on previous experience). During this time the trainee will have a basic training in Clinical Genetics including relevant history taking and pedigree construction, genetic counselling and experience in the management of common disorders in the genetic clinic. The trainee will gain experience throughout the catchment area of the centre (i.e. including peripheral clinics). There may be exposure to the laboratories where necessary to complete training. After this period the candidate should be equipped to decide about areas that they wish to gain further training in.

SpRs will then be seconded to one of the other centres for a year. Each SpR will discuss the opportunities open to them (see appendix) with the programme director and select their rotation of choice. The SpRs will be allocated their rotation positions by the STC with representatives from each centre, overseen by the Training Programme Director (TPD). Each SpR will have their rotation arranged on an individual basis. As of January 2011 there will be fixed calendar dates for rotation (Feb/Aug). While attempts will be made to give SpRs their first choice of posts, a matching system between the centres and trainees will be used where there is uneven demand for posts. If there is an irreconcilable clash, with an excess number of trainees requesting the same placement at the same time, a competitive assessment will be arranged. When SpRs are in seconded appointments they can expect to spend most of their time within the London hospital (i.e. they would not be expected to attend less accessible peripheral clinics).

After secondment trainees will return to their base centre to complete training.

Flexible trainees will be included in the rotation. Secondment will be planned at the halfway point in their training. Secondments may last proportionally longer. Flexible trainees will need to maintain flexibility about their days of work to gain maximum benefit from the system.
In addition to the formal ARCP assessments, trainees and educational supervisors need to be vigilant about the content of training and close liaison is needed between the four centres to ensure that all trainees receive adequate appraisal. Each SpR will have an educational supervisor at the centre in which they are working, but links will be maintained with their supervisor from the base centre.

It is considered desirable for trainees to undertake a period of research whilst in training. Agreement should be obtained prospectively giving a minimum of 3 months notice to the TPD, Regional Advisor and Postgraduate Dean. (Trainees should contact the Deanery Medical Workforce Officer for Clinical Genetics to request an approval form and for further advice). A period of up to a year conducting research may contribute to the overall duration of higher specialist training. Prospective accreditation should be obtained from the Royal College of Physicians, 5 St Andrew’s Place, Regents Park, London, NW1 4LB.

Trainees will have an induction, informal appraisals and will have formal assessments (RITA/ARCP) at least annually in conjunction with the Deanery. It is essential that each trainee attends the RITA/ARCP upon invitation from the Deanery and maintains an up-to-date e-portfolio for review as part of this process.

Trainees who are out of programme conducting research are also expected to go through the annual RITA/ARCP process as a condition of retaining their National Training Number. Therefore satisfactory assessment reports should be provided to the Deanery annually.

This prospectus outlines the organisation of the Clinical Genetics Training Programme in the London Deanery. A more comprehensive guide to the Specialist Registrar Training can be found in the Joint Committee on Higher Medical Training Curriculum for Clinical Genetics available from the Royal College of Physicians. This can be downloaded from their website (http://www.jrcptb.org.uk/).
Specialist Registrar in Clinical Genetics at Great Ormond Street

Clinical Genetics Department - General Information

The clinical genetics department provides a full regional genetics service for the North East Thames area, covering a population of approximately 4.5 million in North East London and Essex. The population of the Region is ethnically diverse. The genetics clinic at Great Ormond Street Hospital was the first to be established in the UK in 1946 and has developed from its close association with the hospital since then, with a large dysmorphology practice because of the specialist nature of the patients seen at Great Ormond Street.

There has been considerable expansion of the unit with particular reference to the development of a large number of outreach clinics to deliver clinical genetics locally, while maintaining a first class service within the hospital. Although the service arose with a focus on Paediatric Genetics, adult genetic care is an integral part of all clinics, and the cancer genetics service is now fully established, providing cancer genetics services to three cancer networks based within the region.

The clinical genetics department is currently situated in a self-contained building under the management of the Institute of Child Health. The general genetics service provides outpatient clinics in 17 locations across the region as well as an in-patient consultation service for Great Ormond Street Hospital. There are a number of specialist clinics based at GOSH covering dysmorphology, deafness, cleft lip and palate, 22q11 deletions and skeletal dysplasias as well as a neurogenetics clinic held at the nearby National Hospital and general clinics based in many locations, including local hospitals and Child Development Centres.

The team works closely with nearby fetal medicine units, particularly at the Elizabeth Garrett Anderson and Obstetric Hospital, (part of the University College London Hospitals), and Homerton Hospital. The department works closely with a number of specialist units at GOSH and has regular meetings with the radiology, dermatology and ophthalmology departments as well as fetal pathology and neuropathology meetings, where specialists from a number of disciplines attend.

The department is actively involved in providing teaching to undergraduates, postgraduates, trainees in Clinical Genetics and other health professionals.

The molecular genetics diagnostic and cytogenetics laboratories are recently relocated to newly refurbished adjoining accommodation in York House (part of the Great Ormond Street complex). The lab structure has recently been modified with the appointment of a single Laboratory Director (Dr Nick Lench) and closer working between the two disciplines. The laboratory and clinical components of the Genetics Service are presently managed within the Medical Unit at Great Ormond Street along with nephrology, metabolic medicine, endocrinology, gastroenterology and adolescent medicine, together with diagnostic and therapeutic services including Radiology and Pathology.

There are also very close links with academic genetics based at the Institute of Child Health. There are active scientific and clinical meetings and teaching programmes at both the ICH and GOSH.
Outline of the programme:
The trainee will gain extensive training in counselling for common genetic disorders including chromosomal abnormalities, neurogenetics including Huntington’s disease, diagnostic dysmorphology, cancer genetics, and prenatal diagnosis. Close liaison exists with both the cytogenetic and DNA laboratories and there will be opportunity for the trainee to obtain experience in the techniques of cytogenetics and molecular genetics.

Supervisors of the post: Dr M Lees

Activity/workload:
Population: 4.5 million
Total annual appointments offered 4500

Ward referrals: 5-10/week
Outpatient visits: 5-10/week

Clinical Genetics Department - The Staffing Structure

Consultant Clinical Geneticists
Dr Angela Barnicoat 9 NHS
Dr Maria Bitner-Glindzicz 3 NHS 8 academic
Dr Ajith Kumar 10 NHS
Dr Melissa Lees 7 NHS including 3 regional cleft service
Dr Alison Male 6 NHS
Dr Elisabeth Rosser 8 NHS including 2 NCG retinoblastoma
Dr Louise Wilson 6 NHS
Dr Lucy Side 5 NHS 5 academic
Dr Adam Shaw 3 NHS 6 academic (fixed term)

Genetic Associates
Ms Cheryl Berlin 0.8 WTE (Royal Free Hospital)
Ms Bernadette Farren 1.0 WTE (GOS based)
Ms Chris Harocopos 1.0 WTE (Bart’s)
Ms Kate Simon 1.0 WTE (Southend based)
Ms Anita Bruce 1.0 WTE (Southend based)
Ms Sally Taffinder 1.0 WTE (shared with UCLH)
Ms Emma Williams 1.0 WTE (North Essex GOSH based)

Junior Medical Staff
Dr K Snape (currently out of programme)
Dr R Scott
Dr J. Kenny

Administrative staff
3.0WTE Band 4 secretarial posts
1.0 WTE Band 3 administrative assistant

February 2010
Description of training:

Induction
During the first month of training the trainee will observe consultations with clinical geneticists and genetic counsellors. During this time he/she will be introduced to the techniques of genetic counselling, including taking a family history, drawing a pedigree and estimating risk.

Further training
The trainee will have responsibility for the collating of information for clinics and begin independent consultations with close supervision. For the first 6 months, he/she will gain experience in counselling for common genetic conditions (e.g. chromosomal abnormalities, common autosomal dominant/recessive and X-linked conditions). After this period of basic training, the trainee will receive more specialist training especially in dysmorphology. The trainee will be trained in counselling pregnant patients. In addition, he/she will accompany consultants on ward visits. Clinic attachments will be at both Great Ormond Street and in some of the peripheral sites covered by Genetic clinics.

Clinics:
- Great Ormond Street Hospital (4-5 per week includes Dysmorphology)
- Elizabeth Garrett Anderson Hospital (fortnightly in Fetal Medicine Unit)
- National Hospital Queen Square (fortnightly Neurogenetics)
- St Bartholomew's Hospital (weekly Retinoblastoma)
- Basildon Hospital (monthly)
- Chase Farm Hospital (monthly)
- Chelmsford Hospital (monthly)
- Queens Hospital, Romford (monthly)
- Middlesex Hospital (quarterly Adolescent Cancer)
- Newham Hospital (quarterly)
- Oldchurch Hospital (monthly)
- London Hospital (monthly)
- Southend Hospital (monthly)
- St Ann’s Hospital, Tottenham (bimonthly)
- Wood Street Children’s’ Centre (bimonthly)

Laboratory training/research
Depending on previous experience, the trainee will undertake laboratory attachments in both the DNA and cytogenetic laboratories.

Meetings
The department holds regular inter-disciplinary meetings with other specialists in Dermatology, Ophthalmology, Fetal pathology, and Radiology. The trainee will be expected to attend these meetings, and be responsible for the organisation of some of these meetings. The trainee will also participate in meetings with the clinical laboratory services, and in the weekly genetic seminars, which are shared between all NHS and academic staff. The trainee will also attend Pan Thames training days held regularly in the 5 regional genetics centres (including Cambridge) and in Central London. The trainee will be encouraged to present at the UK Dysmorphology Club, held three monthly at the Institute of Child Health, London and at national scientific meetings.

Teaching
The trainee may be required to undertake a small amount of undergraduate and postgraduate teaching.
A computerised database is used for patient data with links to the Clinical Molecular Genetics database. There is also access to the hospitals computerised patient database and links with the Cytogenetics database. There is access to LDDB and OMIM as well as Internet services. Family based notes are maintained within the Department. The central Great Ormond Street Hospital appointments centre is responsible for booking appointments at all clinics including those in peripheral hospitals. All the secretarial work is undertaken in the Clinical Genetics department at Great Ormond Street Hospital.

This post complies with the European working time directive regulations.

**Weekly Timetable**

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<th>MON</th>
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<tr>
<td>AM</td>
<td>Ophthalmology meeting Genetics clinic</td>
<td>Journal Club/ Audit/ Dermatology meeting Clinical meeting Slide review Genetics seminar</td>
<td>Admin/audit Radiology meeting (monthly) Oncology clinic Hospital grand round</td>
<td>Dysmorphology clinic (Peripheral clinics)</td>
<td>Genetics clinic</td>
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<tr>
<td>PM</td>
<td>GOSH clinics EGA clinic</td>
<td>GOSH clinic</td>
<td>Admin/clinic prep/audit</td>
<td>GOSH ward reviews</td>
<td>Genetics clinic Neurogenetics clinic</td>
</tr>
</tbody>
</table>

February 2010 6
Specialist Registrar in Clinical Genetics at Guy’s Hospital

Clinical Genetics Department – General Information

The South East Thames Regional Genetic Service is based at Guy’s Hospital. Specialist Registrars undertaking training in Clinical Genetics at this centre will take part in the pan-Thames Clinical Genetics Programme. This programme is run in conjunction with the London Deanery of Postgraduate Medical and Dental Education; the Pan-Thames Specialty Training Committee oversees training.

The Clinical Genetics Department provides a comprehensive clinical and diagnostic service for a population of 3.9 million people in South East Thames, Kent and East Sussex. Within the Trust, children’s services are based at the Evelina Children’s Hospital at St Thomas’s Hospital, and the Integrated Cancer Centre provides comprehensive cancer services for the local population. There is a strong focus on translational research following the creation of the King’s Health Partnership, Academic Health Sciences Centre (AHSC) and the comprehensive Biomedical Research Centre (BRC).

The Department is directly linked with the diagnostic genetics laboratories (DNA molecular, cytogenetics and biochemical genetics) and the Division of Genetics and Molecular Medicine within the School of Medicine of KCL. The regional population is served by clinics held at Guy’s Hospital and also by a network of peripheral clinics together with specialist clinics held at St Thomas’s and the Evelina Children’s Hospitals. The diagnostic services laboratories are innovative and have an excellent track record in supporting trainees. Excellent information services exist, both within the Division and through the NIHR BRC and across the Health Schools of KCL.

Senior staff

Dr Shehla Mohammed, Head of Service for Clinical Genetics
Dr Frances Flinter
Dr Louise Izatt
Dr Fiona Norwood (Neurogenetics)
Dr Dragana Josifova
Dr Charu Deshpande
Dr Gabriella Pichert
Dr Simon Holden
Dr Deborah Ruddy
Dr Melita Irving
Dr Leema Robert
Dr Adonis Ioannides
Dr Fiona Connell
Professor Richard Trembath
Junior Staff
Dr Neeti Ghali
Dr Anthony Vandersteen

Genetic counsellor manager
Chris Patch

Nurse consultant in preimplantation genetic diagnosis and genetic counselling
Alison Lashwood

Genetic consultant counsellor in cancer genetics
Chris Jacobs

Genetic counsellors
Sarah Ross
Sarah Rose
Sara Levene
Sally Watts
Annelise Nehammer
Anna Whaite
Anaar Sajoo
Chris Barnes
Tootie Beuser (cardiac genetics nurse)
Rupinder Jassi
Genevieve Say

Administrative staff
Lisa May PA to Dr Shehla Mohammed
25 secretarial/administrative staff

The Department is well served with IT systems, supporting both the clinical service and research. Close liaison with both the cytogenetic and DNA laboratories

Laboratory Heads
Cytogenetics    Dr Zoe Doherty
Molecular Diagnostics    Dr Steve Abbs
Biochemical Genetics    Dr Marie Jackson
Commissioning and Governance Director, Genetics Labs    Dr Gail Norbury

Senior University Staff includes:
The Prince Philip Professor of Human Genetics (Head of Division) Ellen Solomon
Professor of Molecular Genetics (Deputy Head of Division) Chris Mathew
Professor of Developmental Genetics Mary Seller
Professor of Neurogenetics Gillian Bates
Outline of the programme:
The trainee will gain extensive training in general, common genetic disorders including:

- Dysmorphology and chromosomal abnormalities: clinic exposure, ward round teaching at ECH, weekly discussion of patients (slide meeting)
- Cancer genetics: general clinics, local and regional, and specialist multidisciplinary clinics; cancer genetics meetings (monthly)
- Prenatal genetics: fetal medicine unit clinics and MDT meetings, and weekly Rapid Access Clinic; perinatal pathology

Training in specialist genetics is also offered at Guy’s:

- Neurogenetics: including paediatric and adult condition, Huntington’s disease, myotonic dystrophy
- Cancer genetics, specialist services: von Hippel Lindau, BRCA1/2 carrier clinic, genodermatoses, multiple endocrine neoplasia
- Multidisciplinary clinics: skeletal dysplasia, cardiac genetics and cardiomyopathy, ophthalmology, dermatology, metabolic, PGD, renal genetics.

The trainee will gain extensive training in counselling for common genetic disorders including chromosomal abnormalities, neurogenetics including Huntington disease, diagnostic dysmorphology, cancer genetics, and prenatal diagnosis. The trainee will participate in specialist clinics in cardiac genetics, ophthalmological genetics and prenatal clinics. In addition, a network of peripheral clinics exists, which the trainee will be encouraged to attend. Close liaison exists with both the cytogenetic, DNA and biochemical laboratories and there will be opportunity for the trainee to obtain experience in the techniques of cytogenetics, molecular and biochemical genetics.

Particular training opportunities:
Specialist training opportunities are arranged as blocks.

MULTIDISCIPLINARY CLINIC BLOCKS:

BLOCK 1: Cardiac, Eye and Skin Genetics
BLOCK 2: Neurogenetics paediatric and adult to include HD and MD clinics
BLOCK 3: Prenatal to include FMU, RAC, PGD, women’s endocrine clinic & post mortem exams
BLOCK 4: Cancer VHL, BRCA carrier clinic, skin, & endocrine (MEN)
BLOCK 5: Misc to include metabolic, skeletal dysplasia, cleft lip/palate and cochlear implant services, and renal.

Teaching
The trainee will be encouraged to take part in the many teaching sessions offered by the department at undergraduate (2nd and 4th year MB.BS, intercalated BSc.) and postgraduate levels (subspeciality teaching, MRCPCH course). Formal training in clinical teaching is offered in the Trust.
Management and Audit
The department actively participates in audit and the trainee will be encouraged to undertake audit projects with the opportunity to present at the Trust audit days. An appreciation of the management issues will be provided through attendance at service planning meetings in last training year or election onto committees.

Supervisors of the post: Dr M Irving

Activity/workload:
Population 3.9 million
Total annual appointments offered 3600
(2600 general/ 1000 cancer)
Ward referrals: 6/week
Outpatient visits: 6-10/week

Description of training:
Induction
During the first month of training the trainee will observe consultations by a variety of consultant clinical geneticists and genetic counsellors. During this time he/she will be introduced to the techniques of genetic counselling, including taking a family history, drawing a pedigree and estimating risk.

Further training
The trainee will then begin independent consultations with regular supervision. For the first 6 months, he/she will gain experience in counselling for common genetic conditions (e.g. chromosomal abnormalities, common autosomal dominant/recessive and X-linked conditions). After this period of basic training, the trainee will rotate through a series of attachments including cancer genetics, neurogenetics, and dysmorphology including cardiac, renal and ophthalmological genetics. During these attachments the trainee will be trained in counselling pregnant patients and will be expected to attend perinatal post mortem examinations as they occur. In addition, he/she will attend ward visits.

Peripheral paediatric/general clinics
Neuromuscular clinic (Guy’s) - monthly
Eastbourne (4/year)
Dartford (4/year)
Woolwich (6/year)
Brighton (8/year)
Medway (6/year)
Hastings (6/year)
Folkestone (6/year)
Maidstone (6/year)
Lewisham (4/year)
Canterbury (6/year)
Margate (4/year)
Sidcup (4/year)

Peripheral cancer genetics clinics
Faversham (4/yr)
Maidstone (4/yr)
Brighton (4/yr)
**Multidisciplinary clinics**
Endocrine Genetics (monthly)
Skin cancer genetics clinic (2/yr)
VHL (4/yr)
BRCA clinic (monthly)
Vascular skin clinic (4/yr)
Women’s Genetics Clinic (4/yr)
Huntington Clinic (monthly)
Myotonic dystrophy (6/yr)
Cardiac (monthly)
Eye clinic (monthly)
Skeletal dysplasia clinic (4/yr)

**Laboratory training**
Depending on previous experience, the trainee will undertake laboratory attachments in both the DNA and cytogenetic laboratories.

**Research**
If the SpR has an academic clinical fellowship, then 25% of their time is allocated to research/academic training. The site and project will be in joint discussion with their academic supervisor. They will be encouraged to develop a research interest and apply for substantive research funding to undertake a higher degree out of programme. Once this degree is completed, they would be eligible to apply for a clinical lectureship (50% academic/50% clinic) allowing them to complete their clinical genetics training and continuing to develop their research/academic interest.

If the trainee does not have an academic training fellowship, research opportunities are available, but may be for shorter projects only.

**Meetings**
The department holds regular inter-disciplinary meetings with the paediatric neurologists and the fetal medicine department which the trainee will be expected to attend. The trainee will also attend Thames training days held three monthly in the 5 regional genetics centres (including Cambridge). The trainee will be encouraged to present at the UK Dysmorphology Club, held three monthly at the Institute of Child Health, London and at national scientific meetings.
## Weekly Timetable

<table>
<thead>
<tr>
<th>Day of the week</th>
<th>Morning</th>
<th>Lunch time</th>
<th>Afternoon</th>
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<tr>
<td><strong>Monday</strong></td>
<td>Clinic including urgent appointment</td>
<td>Paediatric ground round</td>
<td>Clinic preparation</td>
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<tr>
<td><strong>Tuesday</strong></td>
<td>Administration</td>
<td>Research seminar</td>
<td>Clinic Joint paediatric neurology/genetics meeting (monthly) Research meeting (monthly)</td>
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<tr>
<td><strong>Wednesday</strong></td>
<td>Slide review Business and clinical meeting Journal club</td>
<td>Fetal Medicine meeting (every 4&lt;sup&gt;th&lt;/sup&gt; Wed) Cancer meeting (monthly) Joint clinic/Lab meeting (monthly) Hospital medical grand round</td>
<td>Administration Or Rapid Access clinic for pregnant patients</td>
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<tr>
<td><strong>Thursday</strong></td>
<td>Peripheral clinic or FMU &amp; ward round</td>
<td>Fetal Medicine meeting (every 4&lt;sup&gt;th&lt;/sup&gt; Thurs)</td>
<td>Peripheral clinic or Administration</td>
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<tr>
<td><strong>Friday</strong></td>
<td>Clinic preparation</td>
<td></td>
<td>Administration</td>
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Specialist Registrar in Clinical Genetics at North West Thames Regional Genetics Centre (Kennedy-Galton Centre)

Clinical Genetics Department - General Information

The Kennedy-Galton Centre (KGC) is located at Northwick Park and St Mark’s Hospital, and is part of the North West London Hospitals NHS Trust. Northwick Park Hospital is a busy District General Hospital with strong academic links to Imperial College, and St Mark’s Hospital is a specialist postgraduate teaching hospital for patients with intestinal disorders. The KCG provides the North West Thames Regional Clinical Genetics Service to a population of 3.5 million in North West London, Hertfordshire, Bedfordshire and part of Middlesex. Within the area are St. Mary’s, Chelsea and Westminster and Charing Cross teaching hospitals, the Hammersmith Hospital and Queen Charlotte’s Hospital with the Royal Postgraduate Medical School, the National Heart and Lung Institute and the Royal Brompton Hospital. There are 15 maternity units within the region, and 50,000 babies a year are born to residents. The area contains areas of affluence and inner city deprivation, and a wide ethnic mix. There are close links with the Academic Department of Physiological Genomics and Medicine based at Hammersmith Hospital. This department organises an MSc course in Human Molecular Genetics (Imperial College London).

Clinical Services
Clinics are held at Northwick Park Hospital and at 17 other locations throughout the region. There are seven consultant clinical geneticists (5.6 WTE). The genetic nurse counsellors have their own case load and provide ongoing support for families. There is close liaison between the clinical and laboratory services as they are all located within the same department. The Centre contains a major regional cytogenetics laboratory, which provides a comprehensive cytogenetics service including molecular cytogenetics (FISH, MLPA and array-CGH), and a haematological cytogenetics service. The KGC has a major molecular genetics diagnostic laboratory, which employs the full range of molecular genetics techniques and has a specialist interest in colorectal cancer predisposition syndromes. The NCG funded Ehlers Danlos syndrome (EDS) service is based jointly at the KGC and Sheffield Clinical Genetics Service. The regionally funded Congenital Malformation Register and the national Confidential Enquiry into Stillbirths and Deaths in Infancy (CESDI) are also based in the Kennedy-Galton Centre.

Outline of the programme:
The trainee will receive training in counselling for a wide range of genetic disorders, including chromosomal abnormalities, neurogenetic disorders, dysmorphology, cancer genetics and prenatal diagnosis. There will be opportunities for training in cytogenetics and molecular genetics techniques.

Activity/workload
Target population: 3.5 million
Annual appointments: 2975
Registrar’s workload:
  Ward referrals 1-2 per week
  Out patient visits 6-9 per week
Staffing Structure

Consultants
Dr Birgitta Bernhard
Dr Angela Brady (Lead Clinician)
Dr Natalie Canham
Dr Huw Dorkins (Lead Clinician in Cancer)
Dr Sue Holder
Dr Kay MacDermot
Dr Emma Wakeling (STC representative)
Professor Michael Pope (Specialist EDS Service)

Junior Medical Staff
SpRs
Dr Anju Kulkarni
Dr Angharad Roberts
Dr Bianca Desousa

F2 trainee on 4 month rotation as part of the Imperial Academic Foundation Programme

Genetic Counsellors
Carole Cummings (EDS Service/St Marks Hospital)
Vicky Kiesel
Kashmir Randhawa (Multilingual specialist)
Marion Turnbull (EDS service/ KGC)

Administrative and Support Staff
Carole Thompson (Business Manager)

Laboratory Staff
Katie Waters (Head of Cytogenetics)
Stewart Payne (Head of Molecular Genetics)
Description of Training:

Induction
Depending on previous experience, a period of observation of consultations will introduce the trainee to the techniques of genetic counselling, pedigree drawing and risk estimation.

Further Training
The trainee will be responsible for preparing the information for the following week’s clinics and will begin seeing patients independently with supervision. Initially the trainee will gain experience in straightforward common conditions and will progress to seeing patients with more complex disorders such as dysmorphic patients and predictive testing for Huntington disease and predisposition to familial cancer. The trainee will gain experience in counselling pregnant patients and will accompany the consultant on ward visits. Patients are seen at Northwick Park Hospital and at peripheral clinics.

Clinics
Clinics are held at the following locations:
Barnet General Hospital (2/ month)
Bedford General Hospital (1/ month)
Bedford Child Development Centre (2/ month)
Chelsea and Westminster (2/ month)
Ealing Hospital (1/ month)
Edwin Lobo Child Development Centre, Luton (0.5/ month)
Hammersmith Hospital (0.5/ month)
Hemel Hempstead General Hospital (2/ month)
Hertford County Hospital (1.5/ month)
Hillingdon Hospital (2.5/ month)
Lister Hospital, Stevenage (1/ month)
Luton and Dunstable Hospital (2.5/ month)
Northwick Park Hospital (8/ month)
Queen Charlotte’s Hospital (1/ month)
St Mary’s Hospital (1/ month)
Watford General Hospital (2.5/ month)
West Middlesex Hospital (2/ month)

The majority of clinics comprise a mixture of paediatric, dysmorphology and adult patients, but some are mainly cancer genetics clinics. There is also the opportunity to sit in on genetic eye clinics bimonthly at Moorfield’s Eye Clinic at Northwick Park Hospital, clinics at St Mark’s Hospital and the specialist EDS service clinics.

Joint clinics with other specialties
Neurogenetics (Hammersmith Hospital) 3 times a year
Paediatric Endocrine clinic 2 times a year
Connective Tissue clinic 4 times a year
Psychiatric/Genetic Adolescent clinic 4 times a year
Cardiomyopathy clinic (Royal Brompton) 12 times a year
Von Hippel Lindau Clinic 3 times a year

Laboratory Training and Research
The clinical section and laboratories are in the same department and there will be opportunities for both formal laboratory attachments and informal discussions with laboratory staff.
Meetings
The clinicians hold a weekly business meeting with the staff of the cytogenetic and DNA laboratories, and a weekly seminar with outside speakers is organised. There are opportunities to attend fetal medicine meetings at Northwick Park, St Mary’s, Luton and Dunstable and Queen Charlotte’s Hospitals.

Teaching
There will be opportunities for the trainee to undertake some undergraduate and postgraduate teaching, including lecturing on the MSc course in Human Molecular Genetics organised by the academic department.

Administration
The Business manager is responsible for the financial and personnel aspects of the department. The office manager supervises the secretarial staff, who run the patient administration and laboratory reporting systems, which are integrated in the Genesis computer system. The laboratory manager is responsible for the maintenance and ordering of supplies and equipment and for health and safety issues.

Information Systems
The Kennedy-Galton Centre has its own library with a good range of genetics journals. The computerised London Dysmorphology Database, Neurology Database, Pubmed and Online Mendelian Inheritance in Man are provided, with access to the Internet and on-line journals through the Imperial College of Medicine. The main hospital library is located within the same building.

The post complies with the junior doctors’ hours regulations.

Weekly Timetable

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<th>TUES</th>
<th>WED</th>
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<tr>
<td>AM</td>
<td>Genetic clinic</td>
<td>Genetic clinic</td>
<td>Departmental meeting to discuss business matters, cytogenetic and molecular results and prenatal diagnosis</td>
<td>Genetic clinic</td>
<td>Audit/ business meeting or Journal Club. Photo review. Clinical meeting to discuss the following week’s cases.</td>
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<td></td>
<td>Departmental Lunchtime Seminar</td>
<td>Hospital Grand Rounds</td>
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<tr>
<td>PM</td>
<td>Case preparation/letters/Ward visit/Audit/Research</td>
<td>Case preparation/letters/Ward visit/Audit/Research</td>
<td>Case preparation/letters/Ward visit/Audit/Research</td>
<td>Case preparation/letters/Ward visit/Audit/Research</td>
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St George's Hospital NHS Trust

Specialist Registrar in Clinical Genetics at St George's Hospital

Clinical Genetics Department - General Information

The Department provides a wide clinical training in the specialty. St George's Hospital serves the District health Authority of Merton, Sutton and Wandsworth which provides health care for a population of 280,000 people within a catchment area covering 74% of the Borough of Wandsworth and 27% of the Borough of Merton. In addition to a district role it is also the only medical school within the South West of the London region. It contains regional units for neurosciences, fetal medicine, cardiology, neonatal intensive care and paediatric surgery.

The South West Thames Regional Genetics Service was established in 1986. The service has grown considerably since then and provides an integrated clinical, cytogenetic, and molecular service on site. The Department has close links with its associated Clinical Molecular Laboratory (Head Ms R Taylor) and Cytogenetic Laboratories (Head Dr. J Taylor).

The clinical service serves a population of 3.1 million. There are monthly district clinics together with a weekly clinic at St George's as well as a number of specialist clinics.

The clinical department has close links with the academic department of medical genetics (Prof M Patton, Prof S Hodgson). The main areas of research interest are Noonan syndrome, adult polycystic kidney disease, Primary Lymphoedema, Breast and bowel cancer and the genetics of heart disease.

Outline of the programme:
The trainee will gain extensive training in counselling for common genetic disorders including chromosomal abnormalities, neurogenetics including Huntington’s disease, diagnostic dysmorphology, cancer genetics, and prenatal diagnosis. Close liaison exists with both the cytogenetic and DNA laboratories and there will be opportunity for the trainee to obtain experience in the techniques of cytogenetics and molecular genetics.

Supervisors of the post: Dr M McEntagart

Activity/workload:
Population: 3.1 million
Total annual appointments offered 4400
Ward referrals: 2-3/week
Outpatient visits: 5-10/week
Staffing Structure

Consultants
Prof M Patton
Dr T Homfray
Dr A Saggar
Dr S Mansour
Dr F Elmslie
Prof S V Hodgson
Dr M McEntagart
Dr K Tatton-Brown

Junior Staff
Dr H Webb
Dr W Jones
(Dr E Baple)

Clinical Nurse Specialists
Ms S Goff
Ms M Peterson
Ms V Attard
Mr G Brice
Ms E Winchester
Ms V Tripathi
Ms C Eddy

Administrative staff
Dr A Marsh (computer manager)
Mr I Turner (IT)
5 full time secretarial posts
4 Appointments coordinator
Records officer

Description of training:

Induction
During the first three months of training the trainee will observe consultations with clinical geneticists and genetic counsellors. During this time he/she will be introduced to the techniques of genetic counselling, including taking a family history, drawing a pedigree and estimating risk.

Further training
The trainee will have responsibility for the collating of information for clinics and begin independent consultations with close supervision. For the first 6 months, he/she will gain experience in counselling for common genetic conditions (eg chromosomal abnormalities, common autosomal dominant/recessive and X-linked conditions). After this period of basic training, the trainee will receive more specialist training. The trainee will be trained in counselling pregnant patients. In addition, he/she will accompany consultants on ward visits. Clinic attachments will be at both St George's Hospital and in some of the 15 peripheral sites covered by Genetic clinics. The trainee will be expected to travel to the peripheral clinics.

Clinics
St George's(weekly general and prenatal clinics, monthly Huntington's disease clinic, monthly neurogenetics and epilepsy clinic, cancer clinics, BRCA1/2 carrier clinic, VHL clinic, Adult endocrine and MEN clinic, NF2 clinic, thrombophilia, inherited renal disease, tuberous sclerosis
and lymphoedema clinics, paediatric endocrinology and overgrowth clinic, inherited cardiac
disease clinic)
Royal Marsden Hospital (weekly)
St Helier Hospital (monthly)
Queen Mary, Roehampton (monthly)
Mayday Hospital (3 per month)
Kingston Hospital (monthly)
Southlands Hospital (monthly)
Worthing Hospital (cancer monthly)
St Richard's, Chichester (monthly)
Epsom Hospital (monthly)
Crawley (monthly)
Royal Surrey Guildford (cancer and general monthly)
Jarvis Screening Centre Guildford (cancer)
East Surrey, Redhill (general genetics monthly)
East Surrey, Redhill (cancer genetics monthly)
Frimley Park Hospital (monthly)
St Peter's Chertsey (monthly)
RNIB college (occasional specialist clinic)

**Specialist clinics**
There are several specialist and multidisciplinary clinics at St. George’s that offer unique training
opportunities. There is a close liaison with the tertiary fetal medicine department at St George’s
with weekly joint meetings and prenatal clinics.
Other specialist clinics include:
Overgrowth clinic
Endocrine disorders
Primary lymphoedema
Neurogenetics/ Huntingtons
Hereditary cardiac disease
Marfan syndrome
Inherited renal disease
Tuberous sclerosis
Epilepsy

**Laboratory training/research**
Depending on previous experience, the trainee will undertake laboratory attachments in both the
DNA and cytogenetic laboratories.

**Meetings**
The department holds regular inter-disciplinary meetings with other specialists. The trainee will
be expected to attend these meetings. The trainee will also participate in meetings with the
clinical laboratory services. The trainee will also attend Thames training days held regularly in
the 5 regional genetics centres (including Cambridge) and in Central London. The trainee will be
couraged to present at the UK Dysmorphology Club, held three times per year at the Institute
of Child Health, London and at national scientific meetings.

**Teaching**
There is active teaching within the department with a dedicated intercalated BSc course as well
as undergraduate and postgraduate teaching.

A computerised database is used for patient data with links to the Clinical Molecular Genetics
database. There is also access to the hospitals computerised patient database and links with the Cytogenetics database will be established shortly. There is access to LDDB and OMIM as well as Internet services. Family based notes are maintained within the Department. Each trainee will have a personal computer for use in the department.

This post complies with the junior doctor's hours regulations.

### Weekly Timetable

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<td><strong>am</strong></td>
<td>RMH (2 per month)</td>
<td>Southlands (monthly)</td>
<td>Journal club</td>
<td>Thrombophilia (monthly)</td>
<td>Mayday (2 per month)</td>
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<td>(cancer)</td>
<td>Kingston (monthly)</td>
<td>Departmental clinical meeting</td>
<td>Guildford ca (monthly)</td>
<td>Frimley ca (monthly)</td>
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<td>Tuberous Sclerosis (monthly)</td>
<td>Guildford (monthly)</td>
<td>Dysmorphology pictures</td>
<td>Jarvis am (monthly)</td>
<td>Worthing ca (monthly)</td>
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<td>St Peter’s (monthly)</td>
<td>Prenatal meeting</td>
<td>Epsom (monthly)</td>
<td>Diabetic clinic (monthly)</td>
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<td>Cancer meeting</td>
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<td>ESH Redhill general (monthly)</td>
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<td><strong>pm</strong></td>
<td>RMH (2 per month)</td>
<td>Cancer genetics (weekly)</td>
<td>Prenatal clinic (weekly)</td>
<td>St George’s (weekly)</td>
<td>Frimley (2 per month)</td>
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<td>Haemophilia (monthly)</td>
<td>Guildford cancer (monthly)</td>
<td>General am (monthly)</td>
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<td>Genetics seminar</td>
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<td>Inherited renal (monthly)</td>
<td>RSCH pm (monthly)</td>
<td>Cancer pm (monthly)</td>
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<td>Adult endocrine MEN (monthly)</td>
<td>BRCA1/2 carrier clinic (monthly)</td>
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<td>Huntington’s (monthly)</td>
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<td>Crawlwy ca (monthly)</td>
<td>Kingston monthly</td>
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<td>Epilepsy genetics</td>
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<td>Inherited Cardiac disease clinic (monthly)</td>
<td>St Peter’s monthly</td>
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<td>VHL 3 monthly</td>
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<td>ESH Redhill cancer (monthly)</td>
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Appendix - Special services at each centre

- **Great Ormond Street Hospital**
  1. Dysmorphology
  2. Genetics of hearing loss
  3. Cancer Genetics
  4. Neurogenetics
  5. Fetal medicine/Genetics

  (Also combined skeletal dysplasia and intersex clinics)

- **Guy's Hospital**
  1. Cancer Genetics (including VHL clinic, genodermatoses, endocrine clinic and BRCA1/2 carrier clinic)
  2. Neurogenetics- (including multidisciplinary myotonic dystrophy and Huntington’s disease clinics)
  3. Paediatric neurology
  4. Dysmorphology/ward round
  5. Fetal medicine/preimplantation genetic diagnosis
  6. Cardiac genetic clinics

- **Northwick Park Hospital**
  1. Cancer Genetics (including St Mark's, MEN and VHL clinics)
  2. Ehlers Danlos syndrome (EDS) service
  3. Dysmorphology

- **St George's Hospital**
  1. Neurogenetics (including Huntington disease, tuberous sclerosis and peripheral neuropathy clinics)
  2. Prenatal genetics
  3. Cancer genetics
  4. Adult genetics (including renal, lipid and cardiovascular disease and thrombophilia, haemophilia)
  5. Dysmorphology