JOB DESCRIPTION AND TRAINING PROGRAMME

EASTERN REGION TRAINING SCHEME

IN

CLINICAL GENETICS ST3

February 2013
1. INTRODUCTION

ST3 in Clinical Genetics - One

The post is approved by the Specialist Advisory Committee in Clinical Genetics of the Joint Royal College Postgraduate Training Board. The Postgraduate Dean has confirmed that the post has the required educational and manpower approval. The Person Specification details the essential requirements for the appointment and the qualities considered desirable.

One speciality registrar training post is currently available. It is a substantive training post in Clinical Genetics, entitling the appointee to a National Training Number.

The East Anglian Medical Genetics Service provides genetics services for the whole of East Anglia. The service is based at Addenbrookes Hospital, Cambridge. The successful candidates will participate in the clinical work of this service. Applicants should have appropriate experience in general medicine, paediatrics or other relevant specialty. Previous experience in clinical genetics is not essential.

In the event of additional places arising as locums for training, or other training appointments, these will be considered at the Appointments Committee.
2. **PERSON SPECIFICATION**

National Person Specification


3. **BRIEF DESCRIPTION OF THE TRUST**

**ADDENBROOKE’S HOSPITAL**

Addenbrookes Hospital, part of Cambridge University Hospitals NHS Foundation Trust was granted Foundation Status on 1st July 2004. It offers a full range of acute and maternity services. Community midwifery services form part of the service, ensuring continuity for those patients who are more appropriately cared for in their own communities.

Addenbrookes Hospital lies on the southern boundary of the Cambridge City, occupying a 66 acre site, which is shared by the University of Cambridge School of Clinical Medicine, the Medical Research Council, the Regional Blood Transfusion Centre and the Parke-Davies Research Institute. Close links with the University have given the hospital an international reputation for converting research and development into practical health care.

The **Rosie Hospital** is also located on this site. This purpose built maternity unit includes the Regional Neonatal Intensive Care Unit, Breast Screening Unit, Gynaecology and Genetic outpatient’s clinics.

**Day Surgery** facilities are based at Ely and outreach services are provided at the following Trusts; Lifespan, Saffron Walden, West Suffolk, Newmarket, Peterborough and Hinchingbrooke.

Addenbrookes NHS Trust employs 5000 staff and offers both a district general hospital service and is a specialist referral centre for a wider catchment population. Some special services, such as bowel/liver transplantation, draw patients supra-regionally or internationally. The Resident population of Cambridge is about 100,000. The catchment population is approximately 400,000 for most general acute specialties, but considerably higher for regional and certain other specialties.

Jane Ramsey was appointed as the Trusts Chairman on 1 November 2022 and chairs the Board consisting of executive and non-executive Directors. They are responsible for the strategic management of the Trust. The executive directors also sit on the Addenbrookes Executive Board which co-ordinates operational policies and practice, where the views of general practitioners are represented.

**UNIVERSITY OF CAMBRIDGE CLINICAL SCHOOL**

The University of Cambridge School of Clinical Medicine on the Addenbrookes site is a major centre for biomedical research and education of world leading quality. In the most recent University Funding Council Research Selectivity Exercise Cambridge shared the highest score for any Medical School in the country. Whilst the University of Cambridge has granted medical degrees since at least 1363, the university could not offer undergraduate clinical education until the Clinical School was formally established in 1975 with purpose built accommodation at Addenbrookes. In addition to these facilities comprising lecture theatres, seminar rooms and first class medical library, a postgraduate education centre was opened in the Clinical School building in 1980. The most recent HEFC teaching quality assessment of the undergraduate clinical education judged the learning facilities and the teaching in the clinical school to be of the highest quality.

The Clinical School admits 130 students annually for the clinical component of their medical education. Student teaching is organised in each department by an Attachment Director, often an NHS consultant, who is responsible to the Clinical Dean for the educational effort of that unit. The majority of students follow an accelerated 2⅓ year clinical course with a strong emphasis on bedside clinical skills as well as clinical science.

In September 1989 the first MB PhD programme in any UK medical school was established in Cambridge, in which selected students complete both their medical degree and a PhD in a five-year
course. From September 2001 a further 20 students commenced an accelerated four-year medical course for graduates.

Undergraduate medical education is delivered in partnership with the School of the Biological Sciences and a number of associated partner NHS Trusts and general practices throughout the eastern region. The university offers three integrated undergraduate medical education programmes leading to the MBBChir degree:

1. **standard course**: six years including a compulsory BA (Medical and Veterinary Sciences Tripos).
2. **MBPhD course**: extended standard course of nine years including the BA and a research period intercalated between years 4 and 5 leading to a PhD.
3. **Cambridge Graduate Course in Medicine (CGC)**: accelerated four year programme for graduates with a first degree in any academic discipline.

The university admits 280 students to the standard course. Initially based in the School of Biological Sciences, all students undertake two years of core biomedical science study, with additional input from the Clinical School to contextualise their scientific study and provide some early clinical experience. Year 3 is a further year of academic study leading to the University's primary degree, the BA. Students then progress to clinical studies either in the School of Clinical Medicine, one of the London medical schools or Oxford. Over the last five years, Cambridge has become the clinical school of choice for our students, resulting in competition for clinical places. From 2012 intake, the School will admit 160 students to continue study in Cambridge; we hope to increase this number over the next few years.

The Clinical School offers students a first class scientific approach to medicine combined with learning and teaching programmes that emphasize the development of excellence in the clinical, communication, practical and professional skills required for good medical practice. Based at the Cambridge University Hospitals NHS Trust (Addenbrookes Hospital) on the Biomedical Campus, the School promotes the benefits of learning medicine in a research – rich environment, offering all students opportunities to pursue their interests through the student–selected components and other project, research and independent study opportunities. Teaching also occurs in a network of partner NHS Trusts throughout the eastern region and a large number of associated general practices. These are linked by an enthusiastic and committed Clinical Sub-Deans group, the Clinical Skills Unit network and the School's virtual learning environment, enabling simultaneous curriculum delivery across a wide range of sites. The School provides staff development and regular education and Quality Assurance visits to promote these partnerships in teaching and learning.

Each year, between five and ten students admitted to the standard course are accepted onto the MBPhD programme. At the end of year 4, these students intercalate a PhD, with groups across the university and other local research institutions offering studentships. Throughout their doctoral studies, students continue to receive clinical supervisions and seminars. Having completed their PhD, MBPhD students are integrated into the final two years of the CGC clinical programme.

The CGC provides a graduate entry route for 20 academically able students whose first degree may be in any subject, not restricted to biomedical science. During the first two years students undertake the standard core biomedical science programme, spending the vacation time following a bespoke programme of clinical study based at West Suffolk Hospital and a small number of specially selected GP practices. The final two years are largely integrated with the standard programme with the final year for CGC and MBPhD students based at West Suffolk.

The School maintains close links with our local NHS postgraduate education community through the CUHP and good working relationships with the Postgraduate Deanery team and East Anglia Foundation School. Looking forward, the establishment of strong partnerships in the local health economy provide security for student placements and continuity between undergraduate and postgraduate medical education. Establishment of the Clinical Academic Training Office within the School has further enhanced opportunities for seamless clinical academic education, particularly providing career structure and guidance for students wishing to pursue combined clinical and research – based training.

The University provides exciting and varied programmes for medical students. Outcome measures have demonstrated significant improvement in national ranking when Clinical School graduates are asked about their preparedness for being a doctor (placing Cambridge now in the top 10 of all UK medical
schools for this indicator) and over 90% of our students have been placed in their first choice Foundation School since the introduction of the national selection process. Most importantly, we now have very high levels of student satisfaction, both in internal feedback and in the National Student Survey, rising from 67% overall satisfaction in 2008 to 93% in 2011.

The Cambridge Biomedical Campus located on the southern edge of Cambridge, encompasses Addenbrookes Hospital, a major University Teaching Hospital with 1100 beds and a comprehensive range of regional services, the Clinical School and its 12 departments and associated Institutes, the Medical Research Council Laboratory for Molecular Biology (LMB) and the new Cancer Research UK Cambridge Research Institute which will house some 350 researchers from the University and Cancer Research UK. Research on cancer is also being undertaken at the nearby Strangeways Research Laboratory. In addition to the LMB, there are five other MRC Units in Cambridge, of which four are located on the Addenbrookes site. Within the local environment are the University’s main biological science departments on the Downing Site in the centre of Cambridge, and to the south the Babraham Institute (funded by the Biotechnology and Biological Sciences Research Council) and the Hinxton Campus with the Wellcome Trust Sanger Institute and the European Bioinformatics Institute. This co-location in Cambridge of a major teaching hospital, medical school and several large research institutes on a single campus, together with these surrounding strengths in biological science in a 5 mile radius offers a unique environment for the highest quality biomedical research. Addenbrookes Hospital in collaboration with the University of Cambridge has been awarded funding of £65m for 2007–2012 to become a National Institutes of Health Research Biomedical Research Centre (BRC) under the new Department of Health initiative Best Research for Best Health. Genomics is one of the research themes of the BRC. The Addenbrookes “2020 vision” plans for a further 70 acre expansion of the campus for additional clinical and research facilities. This includes the relocation of Papworth Hospital, a centre of excellence for cardiothoracic medicine and surgery and the UK’s main centre for heart and lung transplantation.

The Clinical School is a member of Cambridge University Health Partners, a partnership between the University of Cambridge, Cambridge University Hospitals NHS Foundation Trust (the main acute trust for Cambridge), Papworth Hospital NHS Foundation Trust (a specialist Cardio-Thoracic Trust) and the Cambridge and Peterborough NHS Foundation Trust (the regional Trust responsible for Mental Health).

Cambridge University Health Partners has been awarded ‘Academic Health Science Centre Status’ in recognition of its excellence in research education and clinical service, and of the close working relationships between the partners which enable this excellence to be achieved. Cambridge University Health Partners is based on the Cambridge Biomedical Campus.

Through the University of Cambridge, CUHP has access to a wealth of pre-clinical excellence, both in Clinical School Departments and Institutes, and in the School of the Biological Sciences. The School’s research strategy has identified a number of strategic themes, which are aligned with the strategic themes of the School of the Biological Sciences and our NHS partners. These are:

- Cancer
- Cardiovascular science and medicine
- Developmental and regenerative biology and medicine
- Epidemiology and public health
- Functional genomics, systems biology and genetic medicine
- Infection and immunity
- Medical Imaging
- Metabolic medicine, integrative and comparative physiology
- Neuroscience, psychology and mental health
- Women’s health

GENERAL INFORMATION

Cambridge is one of Britain’s smallest cities but also one of the fastest growing. The Arts Theatre within Cambridge is thriving and there are many musical activities to enjoy. The Fitzwilliam Museum is world famous.
For those with children of school ages, there is a full range of public and private education institutions covering all age groups.

Communications with the rest of England have much improved in recent years. Cambridge is served by the national motor way network and regular train services to London King’s Cross or London Liverpool Street have a journey time of less than one hour.

Within Addenbrookes, the main concourse offers excellent shopping facilities; an advice centre; Bank; café; clothes boutique; dry cleaners; financial advisory services; florist; hairdressing salon; mini-market; newsagent; The Body Shop; gift shop; solicitor and travel agents. There is a Food Court which offers “fast-food”, as well as conventional options 24 hours a day.

In addition the Frank Lee Leisure Centre provides comprehensive facilities for swimming, squash, a multi-sports hall, a floodlit outdoor multi-sports facility and the Profiles Fitness Suite.

The Cambridge University Postgraduate Medical Centre has catering and bar facilities as well as the library, lecture theatres and seminar rooms.

Within the University of Cambridge, there is an unrivalled range of educational facilities, diverse cultural, sporting and other leisure activities.

4. THE DEPARTMENT OF CLINICAL GENETICS

4.1 Description of Department

The East Anglian Medical Genetics Service provides a comprehensive genetics service for a population of over 2 million. This comprises a clinical genetic service for diagnosis and genetic counselling. The service is delivered via out-patient clinics, with some ward referrals. Referrals are currently at the rate of 6,000 per annum. Ward referrals are seen by arrangement. The genetic counsellors work with the clinicians in the provision of the service.

The clinical laboratory services (cytogenetics and molecular genetics) provide diagnostic tests, prenatal diagnosis and carrier detection for a wide range of chromosomal and single gene disorders. A maternal serum screening programme is provided for the prenatal detection of Down’s syndrome, neural tube defects and other disorders which is used by 80% of mothers in the region. There is close liaison with the Regional Biochemical Genetic Screening Service based at Addenbrookes.

The cytogenetics laboratory is well equipped for modern techniques including fluorescence in situ hybridisation (FISH) and microarray. It produces over 3,000 chromosome analyses per year of which 1,200 are for prenatal diagnosis. Similarly, over 3,000 DNA diagnoses are provided each year by the molecular genetics laboratory. Other specialised DNA laboratories are starting to join the Molecular Genetic laboratory – increasing the range of tests available. Increasingly the laboratories are becoming a single genetic laboratory with shared staff and expertise.

Computer services have been developed to assist with clinic booking, patient information and follow-up, test in progress, amniocentesis and other audit, dysmorphology database searching and pedigree plotting. Computer access is available to the various genome and protein databases, OMIM and relevant journals and literature searching tools.

4.5 Staff of the Department
NHS Consultant Dr Joan Paterson
NHS Consultant Dr Helen Firth
NHS Consultant Dr Sarju Mehta
NHS Consultant Dr Soo-Mi Park
NHS Consultant Dr Ruth Armstrong
Clinical Lead, Reader in Neurogenetics, Honorary Consultant  Dr Lucy Raymond
University Reader and Hon. Consultant Dr Richard Sandford
Professor of Medical Genetics and Hon. Consultant Dr Geoff Woods
Honorary Consultant Dr Jenny Carmichael
University Lecturer and Hon. Consultant Dr Marc Tischkowitz
University Clinical Lecturer (ST post) post vacant

Academic Clinical Fellow Dr Ana Lisa Taylor Tavares
Academic Clinical Lecturer Dr Kate Baker
Specialist Registrar in Clinical Genetics Dr Katherine Schon
Specialist Registrar in Clinical Genetics Dr Deepak Subramanian

Genetic Counsellors
Consultant Genetic Counsellor Ms Ann Kershaw
Senior Counsellor Ms Vicky Wiles
Lead Cancer Counsellor Dr Sue Kenwrick
Counsellor Ms Felicity Wadrup
Counsellor Ms Gaya Connelly
Counsellor Dr Sue Kenwrick
Counsellor Ms Jacqueline Hodgkinson
Counsellor Ms Lisa Hughes
Counsellor Ms Mabella Farrer
Counsellor Dr Amy Taylor
Counsellor Ms Sarah Wilcox
Counsellor Ms Bev Speight
Counsellor and DDD research assistant Mr Jonathan Roberts

Senior Laboratory Staff
SDU Director Dr Stephen Abbs
Head of Genetics Laboratory Dr Becky Treacy
Head of Cytogenetics Dr Ingrid Simonic

Associated Academic Staff
Acting Head of University Dept of Medical Genetics. Prof John Todd
Wellcome Trust Senior Fellow in Clinical Science Dr David Rubinzstein
Wellcome Trust Senior Clinical Research Fellow Dr Fiona Karet
Wellcome Trust Senior Clinical Fellow Dr Evan Reid

5. THE WORK OF THE DEPARTMENT
The Clinical Department and Molecular Genetics and Cytogenetics Laboratories are situated on Level 6 of the new Addenbrookes Treatment Centre.

Out-patient clinics are held at:
Addenbrookes Hospital, Cambridge
Ipswich Hospital
Hinchingbrooke Hospital, Huntingdon
Peterborough Hospital,
King’s Lynn Hospital
West Suffolk Hospital, Bury St Edmonds
Norfolk and Norwich Hospital, Norwich
James Paget Hospital, Great Yarmouth
Saffron Walden Community Hospital

Consultants and SpR’s usually have a mix of Addenbrookes clinics with no more than two peripheral clinic allocations.
There is a close tie between the research work of the University Department and NHS Clinical Genetic Departments. Much of the work is translational. There are currently ten joint academic/clinical positions. The fields of endeavour are; adult neurodegeneration, paediatric mental neurodevelopment, type 1 diabetes and genetic kidney diseases.

5.1 Time Table of Activities

The departmental timetable is arranged on a monthly cycle to accommodate peripheral clinics in district general hospitals throughout the region. This timetable therefore serves as an example of time allocation in an average week.

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<thead>
<tr>
<th>Time</th>
<th>Activity</th>
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<tbody>
<tr>
<td>Monday</td>
<td>General Genetics Clinic</td>
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<tr>
<td></td>
<td>Post-clinic work</td>
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<tr>
<td>Tuesday</td>
<td>General Genetics Clinic</td>
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<tr>
<td>Wednesday</td>
<td>Post-clinic work</td>
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<tr>
<td>Thursday</td>
<td>Research Departmental Meeting, laboratory liaison and seminar</td>
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<tr>
<td>Friday</td>
<td>Audit</td>
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<td></td>
<td>Clinic preparation</td>
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The appointee will be responsible to the Clinical Director, currently Dr Lucy Raymond. Day to day issues are dealt with by the Educational Supervisors and Training Programme Director, Dr Soo-Mi Park and Dr Richard Sandford.

6. STUDY/MEETINGS

Weekly departmental meetings are held on a Thursday with a format of:
- Departmental seminar 1.30-2.30
- Laboratory liaison and Pregnancy meeting 2.30-3.00
- Clinic review 3.30-5

In addition trainees are encouraged to attend:
- The weekly Addenbrookes staff round on Wednesday 13.00 – 14.00
- Fetal medicine meeting on Wednesday 8.15-9
- NICU meeting Wednesday 12.30-1.30
- Cancer genetics meeting Wednesday 2-3
- Neuromuscular genetics meeting once a month
- Paediatric Neurogenetics meeting once a month
- Dysmorphology club at the Institute of Child Health (3 monthly meeting)
- Pan-Thames training days in Cambridge and the four London centres (3 monthly)
- RSM dysmorphology meetings (3 monthly) (Senior Trainees)
- National Cancer genetics meetings
7. POSTGRADUATE CENTRES

Clinical Tutor: Dr Arun Gupta 01223 217105
Postgraduate Centre Manager: Mrs Mary Archibald 01223 217060
Royal College Specialty Advisor Dr Richard Sandford 01223-216446
Postgraduate Dean Professor Simon Gregory 01223 743305

8. RESEARCH OPPORTUNITIES

The department has a large academic base. Current interests of the dept. include neurodegenerative disease, pain, diabetes, renal disease, eye disease, Tuberose sclerosis, X-linked learning disability and neurodevelopment. There is ample opportunity for trainees to develop their research interests and there is a strong track record of successful fellowship applicants.

9. SPECIAL EXPERIENCE

The department provides a genetic service to the whole of the East Anglian population, so there is excellent exposure to the whole range of genetic disorders in the general clinics. In addition specialist clinics are held in:

- Prenatal diagnosis
- Cardiac genetics
- Eye genetics
- Neurogenetics
- Cancer genetics
- Dementia and Huntington’s disease
- Endocrine (adult and paediatric)
- Renal
- Tuberous Sclerosis
- VHL
- 22q11
- NF2

There are weekly liaison meetings with the Neonatal Intensive Care Unit, and monthly meetings with Fetal Medicine and Paediatric and Adult Neurology

10. TRAINING PROGRAMME

The appointee to the substantive post will receive a National training number.

11. HOURS OF DUTY

Full-time. No out of hour’s duties