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**COMPETENCES REQUIRED FOR APPLICANTS  
TO ATTAIN STATE REGISTRATION AS CLINICAL SCIENTISTS**

<b>MODALITY:</b>	<b>CLINICAL GENETICS</b>	<b>SUBMODALITY: (if applicable)</b>	<b>CYTOGENETICS</b>	<b>APPLICANT'S NAME:</b>	*****
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**APPENDIX 1**

**This set of documents must be completed and returned in your portfolio.**  
**Please complete the three header sections above on each page.**  
**Refer to the Specific Competences document for guidance in completing this document.**  
**Use typescript or black ink and block capitals for all sections.**

**EXPERIENCE:** The candidate should be able to demonstrate that he/she has worked in an environment that has enabled the individual to receive training and gain experience relevant to the competences set out below.

**1-SCIENTIFIC**

<b>HPC Standards of Proficiency Codes for Clinical Scientist</b>	<b>AREA OF COMPETENCE</b>	<b>INDICATE SECTION(S) IN PORTFOLIO WHERE COMPETENCE IS DEMONSTRATED</b>
3a.1p	<ul style="list-style-type: none"> <li>understanding the science that underpins the specialty (modality) and the broader aspects of medicine and clinical practice</li> </ul>	PARAGRAPHS: 1.1, 2.2, 2.4, 2.6, 2.7, 3.2, 3.3, 3.5, 3.6, 3.10 EVIDENCE: 1, 2, 3, 4, 6, 7, 8, 9,
3a.1g	<ul style="list-style-type: none"> <li>demonstrating a strong base of knowledge appropriate to the specialty and to the investigations and therapeutic options available</li> </ul>	PARAGRAPHS: 1.1, 2.2, 2.4, 2.6, 2.7, 3.2, 3.3, 3.4, 3.5, 3.6, 3.10 EVIDENCE: 1, 2, 3, 4, 5, 6, 7, 8, 9,
2b.1g 2b.1p	<ul style="list-style-type: none"> <li>experience of searching for knowledge, critical appraisal of information and integration into the knowledge base</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.5, 2.6, 3.2, 3.5, 3.10, 3.19 EVIDENCE: 1, 2, 3, 4, 6, 7, 8, 11, 15, 18, 19
2b.1g	<ul style="list-style-type: none"> <li>ability to apply knowledge to problems associated with the routine provision, and development, of the service</li> </ul>	PARAGRAPHS: 3.2, 3.3, 3.4, 3.5, 3.6, 3.7, 3.10, 3.15, 3.20, 3.21 EVIDENCE: 2, 3, 4, 11, 16
2a.1p	<ul style="list-style-type: none"> <li>ability to identify the clinical decision which the test/intervention will inform</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.6, 3.2, 3.5, 3.6, 3.10 EVIDENCE: 2, 3, 4, 14, 15
2c.1p	<ul style="list-style-type: none"> <li>ability to make judgements on the effectiveness of procedures</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.5, 3.15, 3.19, 3.20 EVIDENCE: 4, 11, 16
3a.2g	<ul style="list-style-type: none"> <li>application of the knowledge base to the specialty (modality) and to the range of procedures/investigations available</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.5, 2.6, 2.7, 3.2, 3.3, 3.4, 3.5, 3.6, 3.8, 3.9, 3.10, 3.11, 3.14, 3.15, 3.20, 3.21, 3.25 EVIDENCE: 2, 3, 4, 11

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**2-CLINICAL**

<b>HPC Standards of Proficiency Codes for Clinical Scientist</b>	<b>AREA OF COMPETENCE</b>	<b>INDICATE SECTION(S) IN PORTFOLIO WHERE COMPETENCE IS DEMONSTRATED</b>
2b.1p	<ul style="list-style-type: none"> <li>ability to provide interpretation of data and a diagnostic (therapeutic) opinion, including any further action to be taken by the individual directly responsible for the care of the patient</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.6, 3.2, 3.3, 3.5, 3.6, 3.10, 3.20 EVIDENCE: 3, 4, 14, 15
3a.1p	<ul style="list-style-type: none"> <li>understanding of the wider clinical situation relevant to the patients presenting to his/her specialty</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.6, 3.2, 3.3, 3.5 EVIDENCE: 4, 7, 8, 9, 15
2b.3p	<ul style="list-style-type: none"> <li>ability to develop/devise an investigation strategy taking into account the complete clinical picture</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.6, 3.2, 3.3, 3.5, 3.6, 3.14, EVIDENCE: 4, 14
3a.2p	<ul style="list-style-type: none"> <li>understanding of the clinical applications of his/her specialty and the consequences of decisions made upon his/her actions/advice</li> </ul>	PARAGRAPHS: : 2.2, 2.4, 2.6, 3.2, 3.3, 3.5, 3.25, 3.6, 3.14, EVIDENCE: 3, 4, 7, 8, 9
3a.2p	<ul style="list-style-type: none"> <li>awareness of the evidence base that underpins the use of the procedures employed by the service</li> </ul>	PARAGRAPHS: 1.2, 2.5, 3.4, 3.9, 3.19, 3.20, 3.21 EVIDENCE: 2, 7, 9, 10, 11, 18

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**3-TECHNICAL**

<b>HPC Standards of Proficiency Codes for Clinical Scientist</b>	<b>AREA OF COMPETENCE</b>	<b>INDICATE SECTION(S) IN PORTFOLIO WHERE COMPETENCE IS DEMONSTRATED</b>
3a.2p	<ul style="list-style-type: none"> <li>understanding of the principles associated with a range of techniques employed in the modality</li> </ul>	PARAGRAPHS: 1.1, 1.3, 2.2, 2.4, 2.5, 2.6, 3.2, 3.4, 3.5, 3.6, 3.7, 3.8, 3.9, 3.10, 3.11, 3.13, 3.15, 3.20, 3.21, 3.24 EVIDENCE: 2, 3, 4, 11, 16, 17
3a.2p	<ul style="list-style-type: none"> <li>knowledge of the standards of practice expected from these techniques</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.6, 3.2, 3.4, 3.5, 3.6, 3.14, 3.16, 3.20 EVIDENCE: 2, 3, 4, 16, 19
2b.4p	<ul style="list-style-type: none"> <li>experience of performing these techniques</li> </ul>	PARAGRAPHS: 1.3, 2.2, 2.4, 2.5, 2.6, 3.2, 3.3, 3.5, 3.6, 3.10, 3.14, 3.15, 3.20, 3.21 EVIDENCE: 2, 3, 4, 11, 16
2b.4p	<ul style="list-style-type: none"> <li>the ability to solve problems that might arise during the routine application of these techniques (troubleshooting)</li> </ul>	PARAGRAPHS: 2.5, 3.7, 3.15, 3.16, 3.20, 3.21 EVIDENCE: 3, 4, 16
2c.2g	<ul style="list-style-type: none"> <li>understanding of the principles of quality control and quality assurance</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.6, 3.8, 3.11, 3.20, 3.21, 3.22, 3.26, 3.27 EVIDENCE: 19, 21, 22
2c.1p	<ul style="list-style-type: none"> <li>experience of the use of quality control and quality assurance techniques including restorative action when performance deteriorates</li> </ul>	PARAGRAPHS: 3.8, 3.11, 3.15, 3.16, 3.22, 3.24, 3.26 EVIDENCE: 16, 22

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**4-RESEARCH AND DEVELOPMENT**

<b>HPC Standards of Proficiency Codes for Clinical Scientist</b>	<b>AREA OF COMPETENCE</b>	<b>INDICATE SECTION(S) IN PORTFOLIO WHERE COMPETENCE IS DEMONSTRATED</b>
2b.1p	<ul style="list-style-type: none"> <li>ability to read and critically appraise the literature</li> </ul>	PARAGRAPHS: 1.2, 2.2, 2.4, 2.5, 2.6, 3.19, 3.20 EVIDENCE: 1, 2, 11, 18
2b.1p	<ul style="list-style-type: none"> <li>ability to develop the aims and objectives associated with a project</li> </ul>	PARAGRAPHS: 1.2, 2.2, 2.4, 2.5, 3.20, 3.21 EVIDENCE: 1, 2, 11
2b.1p	<ul style="list-style-type: none"> <li>ability to develop an experimental protocol to meet the aims and objectives in a way that provides reliable and robust data (i.e. free of bias)</li> </ul>	PARAGRAPHS: 1.2, 2.2, 2.4, 2.5, 3.20, 3.21 EVIDENCE: 1, 2, 11
2b.1p	<ul style="list-style-type: none"> <li>ability to perform the required experimental work ability to produce and present the results (including statistical analysis)</li> </ul>	PARAGRAPHS: 1.2, 2.2, 2.4, 2.5, 3.20, 3.21 EVIDENCE: 1, 2, 11
2b.1p	<ul style="list-style-type: none"> <li>ability to critically appraise results in the light of existing knowledge and the hypothesis developed and to formulate further research questions</li> </ul>	PARAGRAPHS: 1.2, 2.2, 2.4, 2.5, 3.2 EVIDENCE: 1, 2, 11, 15
2b.1p	<ul style="list-style-type: none"> <li>ability to present data and provide a critical appraisal to an audience of peers – both spoken and written</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.5, 2.6, 3.2 EVIDENCE: 1, 2, 4, 12, 13, 15, 18, 20

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**5-COMMUNICATION**

<b>HPC Standards of Proficiency Codes for Clinical Scientist</b>	<b>AREA OF COMPETENCE</b>	<b>INDICATE SECTION(S) IN PORTFOLIO WHERE COMPETENCE IS DEMONSTRATED</b>
-	<ul style="list-style-type: none"> <li>ability to assess a situation and act accordingly when representing the specialty</li> </ul>	PARAGRAPHS: 2.2, 2.5, 2.6, 2.8, 3.2, 3.4, 3.5, 3.6, 3.9, 3.10, 3.23, 3.24, 3.25 EVIDENCE: 4, 12, 15
1b.2p	<ul style="list-style-type: none"> <li>ability to respond to enquiries regarding the service provided when dealing with clinical colleagues</li> </ul>	PARAGRAPHS: 3.5, 3.6, 3.14, 3.24, 3.25 EVIDENCE: 4
1b.4g	<ul style="list-style-type: none"> <li>ability to communicate with patients, carers and relatives, the public and other healthcare professionals as appropriate</li> </ul>	PARAGRAPHS: 2.2, 2.5, 2.6, 3.4, 3.5, 3.6, 3.9, 3.19, 3.23, 3.24, 3.25 EVIDENCE: 4, 12, 13, 14, 15, 17, 18, 20
1b.5p	<ul style="list-style-type: none"> <li>ability to communicate the outcome of problem solving and research and development activities</li> </ul>	PARAGRAPHS: 1.2, 2.5 EVIDENCE: 12, 13, 15
2b.1p 1b.5p	<ul style="list-style-type: none"> <li>evidence of presentation of scientific material at meetings and in the literature</li> </ul>	PARAGRAPHS: 2.2, 2.5, 3.2, 3.18, 3.19 EVIDENCE: 4, 12, 13, 15, 18, 20

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**6-PROBLEM SOLVING**

<b>HPC Standards of Proficiency Codes for Clinical Scientist</b>	<b>AREA OF COMPETENCE</b>	<b>INDICATE SECTION(S) IN PORTFOLIO WHERE COMPETENCE IS DEMONSTRATED</b>
2a.2g 2c.1g	<ul style="list-style-type: none"> <li>to assess a situation</li> </ul>	PARAGRAPHS: 3.7, 3.15, 3.16, 3.20 EVIDENCE: 11, 16
2b.1g	<ul style="list-style-type: none"> <li>determine the nature and severity of the problem</li> </ul>	PARAGRAPHS: 3.7, 3.15, 3.16, 3.20 EVIDENCE: 11, 16
2b.1g	<ul style="list-style-type: none"> <li>call upon the required knowledge and experience to deal with the problem</li> </ul>	PARAGRAPHS: 3.7, 3.15, 3.16, 3.20 EVIDENCE: 11, 16
2b.1g	<ul style="list-style-type: none"> <li>initiate resolution of the problem</li> </ul>	PARAGRAPHS: 3.7, 3.15, 3.16, 3.20 EVIDENCE: 11, 16
-	<ul style="list-style-type: none"> <li>demonstrate personal initiative</li> </ul>	PARAGRAPHS: 3.7, 3.15, 3.16, 3.20 EVIDENCE: 11, 16

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**7-MANAGEMENT**

<b>HPC Standards of Proficiency Codes for Clinical Scientist</b>	<b>AREA OF COMPETENCE</b>	<b>INDICATE SECTION(S) IN PORTFOLIO WHERE COMPETENCE IS DEMONSTRATED</b>
1a1.g	<ul style="list-style-type: none"> <li>Understanding of the legal and ethical boundaries of the modality, and the ethical aspects of scientific research.</li> </ul>	PARAGRAPHS: 2.5, 3.6 EVIDENCE: 8, 11
1b.1g, 1a.5g	<ul style="list-style-type: none"> <li>Ability to recognise the limits of personal practice and when to seek advice.</li> </ul>	PARAGRAPHS: 3.25 EVIDENCE:
1a.6g	<ul style="list-style-type: none"> <li>Ability to manage personal workload and prioritize tasks appropriately.</li> </ul>	PARAGRAPHS: 1.2, 2.2, 2.4, 2.5, 2.6, 3.2, 3.3, 3.5, 3.6, 3.10, 3.14, 3.20, 3.21 EVIDENCE: 1, 2, 3, 4, 5
2c.2g 1a.3g	<ul style="list-style-type: none"> <li>Understanding of the principles of clinical governance including clinical audit, accreditation requirements relevant to the modality. The importance of confidentiality, informed consent and data security</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.6, 3.2, 3.5, 3.6, 3.8, 3.11, 3.14, 3.17, 3.18, 3.19, 3.22, 3.25, 3.26, 3.27 EVIDENCE: 4, 10, 11, 15, 21
1b.3g	<ul style="list-style-type: none"> <li>Ability to contribute effectively to work undertaken as part of a multi-disciplinary team</li> </ul>	PARAGRAPHS: 2.6, 3.2, 3.3, 3.5, 3.23 EVIDENCE: 8, 21
	<ul style="list-style-type: none"> <li>Ability to supervise others as appropriate to area of practice. Understanding of the role of appraisal in staff management and development.</li> </ul>	PARAGRAPHS: 2.2, 2.6, 3.3, 3.6, 3.14, 3.27 EVIDENCE: 4, 21
1a.7g 1a.8g	<ul style="list-style-type: none"> <li>Understanding of the need for career-long self-directed learning and the importance of continuing professional development.</li> </ul>	PARAGRAPHS: 3.2, 3.3, 3.4, 3.5, 3.6, 3.9, 3.10, 3.12, 3.13, 3.19, 3.27 EVIDENCE: 4, 7, 8, 9, 10, 18, 20, 21
3a.3g	<ul style="list-style-type: none"> <li>Understanding of the need for, and ability to establish and maintain, a safe practice environment.</li> </ul>	PARAGRAPHS: 2.2, 2.4, 2.5, 2.6, 3.2, 3.5, 3.6, 3.8, 3.10, 3.11, 3.22 EVIDENCE: 4, 19
	<ul style="list-style-type: none"> <li>Understanding of the structure and organization of the department and how it fits into the local clinical setting, General understanding of the way the modality is structured and practised in other locations within the UK. Basic understanding of the importance of financial accountability, budgetary control and resource management.</li> </ul>	PARAGRAPHS: 2.2, 2.3, 2.4, 2.5, 3.20, 3.21 EVIDENCE: 7, 8

**Note:**

The above are the generic competences that must be met by all Clinical Scientists. These competences have also been mapped onto specific subjects. Copies of these can be obtained from the ACS Administrative Office and the website.

## Covering Report

### 1. Pre A-Grade Training

Evidence

- 1.1 I graduated from the University of \*\*\*\* in 2002 with a second class honours degree in Genetics. The course covered a wide range of aspects of modern genetics; in the first year I completed modules in genetics, biochemistry, physiology, microbiology, animal form and function, biodiversity and cell function. Second year modules that I studied included cytogenetics, cell cycle genetics, bioinformatics, DNA synthesis and repair, genetic analysis, evolution, molecular evolution, fundamental molecular biology and transferable skills. The third year presented the chance to complete a lab-based project (see 1.2 below). Third year modules studied included genes and development, plant molecular genetics, cancer & gene therapy, human molecular genetics, recombination and genetics of selected human disorders.
- 1.2 In the Third year of my degree I carried out a laboratory-based project within the Institute for Cell and Molecular Biosciences under the supervision of Dr \*\*\*\*. The title of the project was “Sequencing of Topoisomerase II $\beta$  and Expression Studies in Zebrafish”. This involved extensive literature and bioinformatic searching of genomic databases and the use of inverse PCR followed by sequencing to elucidate more of the sequence of the topoisomerase II $\beta$  gene. I also extracted complete RNA from zebrafish and used this to probe clone libraries to investigate the expression of topoisomerase II $\beta$  in the zebrafish brain. This project gave me a good insight into standard laboratory working practice as well as giving me experience of various genetic techniques. This project was presented as written work, as an oral presentation and in poster format.
- 1.3 In 2002 I joined the \*\*\*\* Regional Cytogenetics Laboratory in \*\*\*\* as an MTO2. I was in this post for almost a year and in that time spent 9 months within the prenatal section and 3 months in the FISH section. During this time I gained valuable experience in general laboratory working practice. More specifically I gained experience in the setting up, maintenance and harvesting of solid tissue and amniotic fluid cultures

1

and also the techniques involved in setting up and analysing FISH assays. I was also involved in the analysis of a number of solid tissue cases within this time.

## 2. A Grade Training

2.1 In October 2003 I began my training as a clinical cytogeneticist within \*\*\*\* Regional Cytogenetics Laboratory in \*\*\*\*.

2.2 My first modules covered were the 'Introductory' and 'Cytogenetics of Postnatal Referrals' modules.

In October 2003 I attended a residential course in Birmingham, which laid the groundwork for most of the background knowledge needed for these modules, however extensive personal study was also required. This course was also very useful in making contacts at other labs and finding out how other labs were structured and functioned. Case studies I produced for this module included a pericentric inversion of chromosome 10, a derivative chromosome 6 from a 6;9 translocation, a ring chromosome 18 and an supernumerary inv dup(15). I carried out an experiment comparing the effects of Ohnuki's hypotonic compared to our standard KCl on the length of chromosome preparations. I also performed an audit on the cytogenetic investigation of gamete donors, which I presented at a weekly clinical meeting. During this time I also spent 4 weeks within the Molecular Cytogenetics Section, increasing my experience of practical techniques and expanding my knowledge of FISH theory, focused on postnatal FISH referrals.

2.3 In April 2004, the \*\*\*\* Regional Cytogenetics laboratory hosted the annual ACC conference in \*\*\*\*. I was involved with the entertainment committee at this time, amongst other things helping design a treasure hunt

round \*\*\*\* for delegates. I also helped out at the conference aiding with IT and general organisation.

I completed the 'Cytogenetics of Amniotic Fluid Referrals' module The basic background theory for this module was covered in a residential course that I attended in Newcastle in May 2004 and I added to this through personal study. I also visited The \*\*\*\* Screening Centre to see foetal screening in action. The case studies I produced at this time were of a free trisomy 21, a free trisomy 13, a 10;22 translocation and an isochromosome 12p. I carried out an experiment into the long term storage of amniotic fluid samples at 5°C, -20°C and -70°C and the effect on cell viability and also performed an audit on the previous five years of amniotic fluid referrals. I spent 2 weeks within the Molecular Cytogenetics Section, focusing on prenatal FISH referrals, mainly rapid aneuploidy testing on uncultured amniocytes. During this time I spent 4 weeks working at the cytogenetics laboratory in \*\*\*\*. This gave me an invaluable insight into the structure and operation of a much smaller laboratory.

- 2.4 I spent the first six months of 2005 carrying out a project aiming to establish a test using MLPA to identify deletions and duplications in the CREBBP gene in patients with Rubinstein-Taybi syndrome. This was performed within the \*\*\*\* Regional DNA Lab under the supervision of Dr \*\*\*\*. This involved extensive literature searches to familiarise myself with the syndrome and associated genetic changes and also MLPA as a technique. Suitable patients were selected according to clinical features and cell lines were obtained and cultures to use as positive controls. All patients had originally been referred for Rubinstein-Taybi testing and therefore consent in these patients was not an issue. A lot of time was also spent optimising and troubleshooting the procedures and equipment. 24 patients were tested along with 3 cell lines. A deletion found by FISH was confirmed and two

3, 11, 1

other deletions were found. Molecular techniques such as sequencing and pulse field electrophoresis were employed to further investigate the deletions found. This project was presented at both the BSHG conference in 2005 and the ACC conference in 2006. As this project was performed within the Molecular Genetics department it also served to give me a greater understanding of the structure and role of this department.

2.5 I undertook the 'Cytogenetics of Haematological Disorders'.

3

I expanded on my knowledge of haematological disorders, the basics of which had been covered at the residential course in April 2004. I attended bi-weekly MDT meetings, presenting cytogenetic results at some of these. Case studies I produced in this module were deletion of MLL in AML, near triploidy with t(12;21) in pre-B ALL, homozygous deletion of p16 in T-ALL and a complex Karyotype with loss of chromosome 7 in MDS. I performed an audit on reporting times for cytogenetic techniques versus PCR over a 15 month period. I spent 2 weeks within the Molecular Cytogenetics Section, processing and analysing haematological samples.

2.6 My final assessment took place in December 2005 and was carried out by

\*\*\*\*

2

and \*\*\*\*. I passed the assessment successfully.

2.7 Following both the A-grade residential weeks in Birmingham and

Newcastle I was given the task of writing a report of these training courses for the ACC section of the BSHG newsletter.

### 3. Pre-registration Training

- 3.1 My pre-registration training was supervised by \*\*\*\*, with regular meetings to discuss my progress. Rotation through sections allowed me to consolidate my A-grade training and also to gain experience in areas that I had not covered in my A-grade training. I was also able to participate in other areas such as training, quality management and service development. 4, 5
- 3.2 In January 2006 I moved into the Solid Tumour section. I spent 3 months in this section and was jointly responsible for all processing of solid tumour samples. During this time I was trained in the practical aspects of solid tumour culturing and also covered a lot of theory on different tumour types and their associated cytogenetic abnormalities. I gained experience in the reporting of a wide variety of abnormal results from a range of different tumour types. I also attended weekly paediatric oncology MDT meetings as well as a biannual sarcoma MDT meeting. I performed an audit looking at cytogenetic abnormalities in neuroblastoma and their effects on patient prognosis over the previous five years and presented my results at a weekly clinical meeting. Following this clinical audit it has now become laboratory practice to FISH all neuroblastoma samples with a probe for 11q. 4, 6, 15
- 3.3 From April to July 2006 I worked within the Leukaemia section. This time served to consolidate and expand the theoretical knowledge and practical experience I had gathered as an A-grade trainee. I performed as a routine scientist within the section analysing, interpreting and reporting cases as well as carrying out routine practical work. At times I was responsible for signing referral cards, deciding which cultures were required based on the referral reasons. I regularly attended weekly MDT meetings within this time. 4, 6

3.4 In April 2006 I attended a joint ACC/CMGS Molecular Cytogenetics Workshop at Weetwood Hall in Leeds. At this meeting I presented a short talk on problems encountered when interpreting results from MLPA and problems associated with 2-colour MLPA.

3.5 At the start of July 2006 I rotated into the CVS section where I remained for 3 months.

4, 6, 14

As I had not covered chorionic villus samples in my A-grade training I spent time learning the theoretical knowledge needed for this section, some of which had been covered in the A-grade residential course. I was also completely trained in the practical aspects of CVS cytogenetics and was jointly responsible for all practical and analysis work. This involved liaison with staff in other departments as samples often had to be collected and sent, either pre- or post-culture to other centres for testing. I was also responsible for giving out short-term results via the telephone to clinical members of staff. During this time I was also responsible, on a rota, for representing the laboratory at a weekly prenatal MDT meeting.

3.6 I spent 3 months in the amniotic fluid section from October 2006 till the end of the year.

4, 6

This helped to consolidate my experiences from A-grade training; however it also enabled me to learn both the practical technique and analysis of QF-PCR for rapid prenatal diagnosis of aneuploidy, as this had been introduced as a service in April 2006. I was often responsible for giving out QF-PCR results to clinical members of staff via the telephone. Throughout these six months in the prenatal section I also supervised the solid tissue section, helping to process and analyse samples. During this time I learnt the relevant knowledge required for this section and consolidated the practical experience I had gained as an MTO2, it also gave me the opportunity to see the impact of the recently published Human Tissue Act on this section.

During my time in this section I also helped to coordinate technical staff to ensure timely and accurate processing of samples.

- 3.7 Whilst in the CVS section, there were quality issues with the amnio QFPCR results; the head of the service was on leave, however my experience with capillary electrophoresis, gleaned from my A-grade project enabled me to troubleshoot. Through a process of elimination I was able to trace the fault to faulty capillaries on the ABI genetic analyser. I replaced the capillaries thus solving the problem. 4, 16
- 3.8 During my time in the prenatal section I produced a new SOP for the 'Receipt and maintenance of immortalised cell lines'. I was given this responsibility due to my experience of growing up cell lines gained from my A-grade project.
- 3.9 In December 2006 I was able to be involved in lecturing as part of an MSc course at \*\*\*\*\* University. I gave an hour lecture on future developments in cytogenetics, covering more recent technologies such as MLPA and array CGH as well as developments on the horizon such as methylation-specific MLPA and expression arrays. I will be giving this lecture again as part of the 2007 MSc course.
- 3.10 At the start of 2007 I moved into the postnatal section, where I remained until October 2007. For the first month within the section I was introduced to breakage syndromes and the processing and analysis of these samples. During this time I was jointly responsible for breakage cases and gained experience in analysing Ataxia Telangiectasia, Fanconi's Anaemia and Bloom's syndrome cases. Liaison with other departments was necessary at this time as AT samples are sent away for irradiation as well as being treated with 4, 6,

bleomycin in-lab. Throughout my time in the postnatal section I have been responsible for the reporting of a variety of normal and abnormal cases. I also used this time to expand my theoretical knowledge of this discipline, which had been covered at the A-grade residential training weeks.

3.11 In May 2007 I was responsible for reviewing and altering the lab SOP for culture and analysis of Bloom's syndrome cases.

3.12 In early May 2007 I gave a training talk to the lab on the use of Microsoft Excel. I had  
gained extensive experience of this software through my MLPA project  
and was able to pass on useful techniques to other members of the lab.

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3.13 In early June 2007 I gave a training talk on segregation analysis to the technical members  
of staff as an ongoing training program. I am due to perform another  
training talk on Molecular tests common to cytogenetics in December  
2007.

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3.14 Throughout my time in the postnatal section I performed as duty scientist on a rota basis,  
being responsible for the coordination of technical staff to ensure timely and accurate processing of samples. I was also on a rota as the scientist responsible for the rapid processing of and reporting of urgent samples. Both of these roles require active participation in the practical duties of the section and effective communication with other departments when giving out urgent results over the telephone.

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3.15 Whilst in the postnatal section I have been heavily involved with troubleshooting the

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automatic harvesting robot. This has involved the setting up of duplicate cultures to enable assessment of the effect of changing factors such as concentration of and time in hypotonic on the quality of chromosome preparations. I was also responsible for altering the lab SOP for using the robotic harvester to provide a more thorough and easy to follow protocol. This work resulted in reinstatement of the robotic harvester, which had been unused for approximately six months.

3.16 During my time in the postnatal section there have been QA issues with the blood

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samples, with an increased number of samples achieving substandard banding quality. I queried the effectiveness of our trypsin solution and traced this back to a substandard reagent. This reagent was replaced and banding quality improved.

3.17 As part of the postnatal section I have, on a rota, been responsible for chairing the monthly section meetings and on other occasions taking minutes.

3.18 As a pre-registration scientist I have been responsible on a rota for chairing bi-monthly

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lab meetings, presenting lab figures and other relevant information. I have also presented interesting cases at these meetings.

3.19 I have presented journal reviews on a rota basis as part of a monthly Journal Club. These

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have included 'Fortuitous FISH Diagnosis of an Interstitial Microdeletion (5)(q31.1q31.2) in a Girl Suspected to Present a Cri-Du-Chat Syndrome', 'Fortuitous Detection of a Submicroscopic Deletion at 1q25 in a Girl With Cornelia-de Lange Syndrome Carrying t(5;13)(p13.1;q12.1) by Array-Based Comparative Genomic Hybridization', 'Comparative genomic

hybridization and prenatal diagnosis’ and ‘MLPA vs multiprobe FISH: comparison of two methods for the screening of subtelomeric rearrangements in 50 patients with idiopathic mental retardation’.

3.20 Throughout my pre-registration period I have been involved with the fine tuning,

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validation and introduction of MLPA for subtelomeric screening as a service. I have been responsible for the alteration of an Excel analysis spreadsheet to fit this specific test as well as subsequent adjustments needed to allow a more accurate analysis. I was also responsible for writing the SOP for this part of the technique. A great deal of troubleshooting has been required for identification of causes of erratic results; this has also needed much communication with the MLPA kit suppliers and other service providers as well as extensive literature searches. This technique is now offered as a routine service and as such I am involved in the routine practical work, analysis and reporting of all cases.

3.21 More recently I have also been responsible for further developing a service using MLPA

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to screen for deletions/duplications in Rubinstein-Taybi syndrome. This has involved further testing of previously referred patients for validation purposes. I have also ordered, grown up, extracted and labelled FISH probes with which to confirm any abnormal results found in future patients. I have written the SOP for this MLPA technique as well as performing risk analyses and COSHH assessments.

3.22 Within my time as a pre-registration scientist I have carried out two quality management

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audits. The first was an examination audit observing a technical member of staff harvesting long term solid tumour samples. The second was a vertical

audit in which I followed the audit trail of a solid tumour sample through the department from receipt to reporting.

- 3.23 For the past 2 years the Cytogenetics and Molecular Genetics departments have been preparing to move into new space within the current building, at the same time altering the use of current facilities. With respect to this I have been a member of the 'Space' committee. This has involved meeting on a regular basis both within the department to discuss the needs and hopes of the Cytogenetics department and with the Molecular Genetics department to debate and resolve conflicts of interest and ultimately produce a plan for how the facilities could be best used and how best to physically integrate cytogenetics and molecular genetics. These discussions are ongoing and hopefully movements will begin some time in mid-2008. 4
- 3.24 I have on numerous occasions been involved with the training of visitors to the department such as trainees from the molecular genetics department and registrar clinicians. This has ranged from demonstration of various technical procedures to giving short seminars on topics such as common cytogenetic syndromes. 4
- 3.25 As a pre-registration scientist it is my ongoing responsibility to respond to enquiries from service users either in person, in writing or on the telephone regarding all aspects of the service. I must also be aware of the limitations in my experience and know when to refer to more senior colleagues.
- 3.26 I have on occasion within my pre-registration period discovered incidents that have required the completion of a pre-IR1 form, such as data entry errors and SOP inaccuracies.

3.26.1 As an ongoing part of CPD I have undergone regular appraisals with a Principal Scientist. These have served to highlight my strengths and weaknesses and provide targets for me to work to.

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