ELECTIVE (SSC5b) REPORT (1200 words)

A report that addresses the above four objectives should be written below. Your Elective supervisor will assess this.

1. What genetic conditions are managed at GOSH? How do they differ from genetic conditions managed in general UK hospitals?

The aim of all the Regional Genetic Centres in England is to help those affected by, or at risk of, a genetic disorder to live and reproduce as normally as possible. The Clinical Geneticists at GOSH manage a huge variety of genetic disorders that include: chromosomal abnormalities, single gene disorders, familial cancer and cancer-prone syndromes, and certain birth defects with a genetic component. Specialist clinics provided by the team at GOSH manage conditions including: childhood deafness, retinoblastoma, craniosynostosis, Huntington's Disease, skeletal dysplasias, cleft lip and palate. Together with the rest of the Regional Clinical Genetics centres in England, GOSH accept referrals from GPs or other hospital consultants that comprise:

- **Children with a congenital abnormality or inherited condition. Parents may require detailed information about the condition, have questions about future management of the condition and may also have to consider risk of recurrence for future pregnancies.**

- **Adults with a congenital abnormality or inherited condition that want specialist advice about managing the condition and future implications including potential reproductive options for themselves and their partner.**

- **Parents of a child presenting with significant learning difficulties that could be caused by a genetic condition. If a genetic origin is discovered it may lead to an official diagnosis and offer the parents a better understanding of their child's condition and expectations for the future.**

- **Adults with a recognised familial genetic condition. Providing the patient with specific testing of their own genes enables them to understand and manage any risks to themselves as well as their children.**

- **Adults or a pregnant couple with a potential familial genetic condition. In these cases there is the opportunity to offer genetic testing and provide a diagnosis and explore possible options depending on the results.**

- **A pregnant couple may have been referred on the basis of an abnormal foetal result and wish to discuss these results and further options available to them.**

- **Adults diagnosed with cancer wanting to know if family members are also at risk**

- **Adults with a strong family history of cancer wanting to know if they and other family members are at risk**

2. How are specialist genetic services delivered at GOSH? Are they managed differently to the rest of the UK?

The genetic services at GOSH are managed and delivered in exactly the same way as genetic services are managed and delievered in any other NHS hospital in England. NHS medical genetics services usually cover areas of England with populations of between 1 and 5 million people. There are currently 23 Regional Genetics Centres in England. GOSH provides a clinical genetics service that covers a population of approximately 4.5 million in North and East London and Essex region.

Medical genetics services are offered to both children and adults and consist of two separate parts: clinical genetics and laboratory genetics.

The clinical genetics services are where doctors and genetic counsellors diagnose genetic conditions and/or assess the risk a patient will inherit or develop a genetic condition. These services are usually coordinated through a main hospital where the clinical service is based, and supplemented with regular clinics in peripheral hospitals across the service's area.

The laboratory genetics services are where scientists carry out the genetic tests. These services are not always part of the main hospital where the clinical service is based but may be located in a number of specialised laboratories anywhere in the UK. Patient samples are referred to the laboratory either directly from clinical genetics services or from other hospital services.

At GOSH the Clinical Genetics Unit, the Regional Molecular Genetics Laboratory and the North East London Regional Cytogenetics Laboratory work closely together to provide an integrated service.

3. Describe the burden of genetic disease in the UK and the role of research in its management.

It is estimated there are between 4-6,000 known genetic disorders worldwide. It is thought that 1:25 children born in the UK is affected by a genetic disorder, so approximately 30,000 babies are newly diagnosed each year. This number is probably higher as although some genetic disorders are obvious at birth, others do not become apparent until later in childhood and sometimes into adulthood. It is estimated that more than half a million children and adults in the UK are affected by a genetic disorder.

Research has become incredibly important in the management of genetic disease. The Department of Health has recently implemented the 100,000 Genomes Project in a bid to employ genomics as part of routine care in the NHS. Their initial aim is to sequence the complete genome of 100,000 patients with cancer and rare disease. Patients will hopefully be offered a diagnosis where there previously was not one, and in time it is hoped that new and more effective treatments will be obtainable based on these results. The project will also enable new medical research. Combining genomic sequence data with medical records is a revolutionary resource. It will allow researchers to study how best to use genomics in healthcare and how best to interpret the data to help patients. The results will also contribute greatly to increasing doctors and scientists knowledge of how and why these disorders occur and allow a platform to share this information and enable further future research.

Although cancer and rare diseases are very much the focus at present, in the future it is hoped that genetic contributions to common later onset disorders such as diabetes and coronary heart disease are identified, allowing genetic services to be offered for those patients at high risk.

Uncovering genetic factors that affect drug prescribing will also increasingly become an important activity.

4. Explore the field of clinical genetics as a potential future career.

This placement has been inspirational and I am excited to be entering an era where genetics and medicine will work together to provide the best care for the patient. Having witnessed the expertise and power of effective communication in often tricky consultations, I hope I can take away some of the explanation and negotiation skills with me and implement them during my own consultations.

I will definitely be considering clinical genetics as a career option.