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Issue 1

Great Ormond Street Hospital for Children NHS
NHS Foundation Trust

Clinical Genetics Service

Who provides your local Genetics Service?

The Clinical Genetics department at Great Ormond Street Hospital (GOSH) covers a population of 4.5 million in North and East London and Essex. The majority of our clinics are held in peripheral hospitals.

The North East Thames Regional Genetics Service is based at Great Ormond Street Hospital. 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.

NE Thames Regional Genetics Service serves the population of both North and East London, as well as the whole of Essex.

We hold regular clinics at:

- Colchester General Hospital
- Broomfield Hospital, Chelmsford
- Southend Hospital
- Basildon Hospital / Orsett Hospital
- St Margaret's Hospital, Epping
- Princess Alexandra Hospital, Harlow
- Queen's Hospital, Romford

In addition , we hold clinics at Great Ormond Street Hospital and other central London locations.

What services does the Clinical Genetics Unit provide?

The Clinical Genetics Unit offers clinical assessment and other investigations, including genetic tests, for children and adults with complex medical problems, to aid the diagnosis of genetic conditions. It provides genetic counseling for families about the likely recurrence of congenital abnormalities and medical problems.

For those families with known genetic conditions, the Clinical Genetics department provides genetic counselling to help individuals decide whether to have carrier or predictive testing and arranges this as appropriate.

The service offers prenatal testing to some families with known genetic conditions, in conjunction with the Fetal Medicine Unit, University College London Hospital.

The Clinical Genetics Unit also assesses individuals within families who are concerned about increased risk of cancer where there is a familial aggregation. In families considered to be at increased risk, gene testing may be offered and arranged, as well as advice about methods to reduce risk or screen for tumours.

Inside this issue

Special points of interest

- Regional risk reducing mastectomy MDTs
- Regular BRCA and HNPCC carrier information evenings
- DDD - Deciphering Developmental Disorders

General Genetics Referrals

Dr Jane Hurst,
Consultant in Clinical Genetics
Lead Clinician

We accept referrals from GPs and Consultants, here are examples of common reasons for referral:

•After the birth of a child with a congenital abnormality or inherited condition, for information about the condition and recurrence risks.

•Adults born with a congenital abnormality or inherited condition wanting specialist advice about the condition, including management issues and reproductive options.

•Parents of a child with significant learning difficulties which may be due to a genetic condition, referred for investigation and diagnosis.

•A person with a known genetic condition in the family, to know the risks to themselves and/or their children.

•A person or a pregnant couple with a possible genetic condition in the family wanting to know if a diagnosis can be made, and if so, their risks and options.

•A pregnant couple told that a test has given an abnormal result, wanting to talk about what the result means, and what options are available.

Who and how should you refer to us?

Essex-wide Risk-Reducing Mastectomy MDT

Three times a year a multidisciplinary meeting is held in Essex that includes the following clinicians.

Breast Surgeons

Plastic Surgeons

Psychologists

Geneticists

Breast Nurses

The purpose of this meeting is to discuss all patients in the region who want to have either unilateral or bilateral risk-reducing mastectomies. The surgical risk and risk for a recurrence and/or new primary cancer is assessed, and a consensus is reached about whether surgery should be advised. This ensures a robust and equitable service in Essex for all patients who request this surgery.

Cancer Genetics Referrals

•A person who has been diagnosed with cancer and has a strong family history of cancer, who wants to know if they and other family members are at increased risk.

•A person with a strong family history of cancer, who wants to know if they are at increased risk, and if they are, what options they have.

•A person with a known cancer predisposition gene mutation in the family who wants to discuss and arrange genetic testing so they can be advised of their own cancer risks.

For the Cancer Genetics referral guidelines, please see page 4

Genetics Clinicians and Genetic Counsellors: *September 2012*

Clinical referrals and queries are accepted from GPs, consultants, associate specialists, cancer family history clinics, antenatal services, and other health professionals.

To refer a patient, please write to:

Clinical Genetics Unit

Great Ormond Street Hospital NHS
Foundation Trust
Great Ormond Street
London
WC1N 3JH

Fax: 020 781 38141

Please ensure that a contact telephone number for the patient is provided with the referral.

For enquiries about general genetics patients, please contact the department secretaries, telephone numbers: **020 7762 6845/6856.**

For enquiries about cancer genetics patients, please contact Jacqueline Charles, telephone number: **020 7762 6831.**

Patients are sent an information leaflet with their appointment letter that explains what to expect at a genetics clinic.

BRCA carrier and HNPCC carrier information evenings

Since 2008 we have arranged regular information evenings for our carriers of inherited cancer predisposition genes, such as BRCA and HNPCC.

These evenings have been arranged in both London and Essex locations and have been very well attended by our patients.

We invite speakers, to talk on subjects such as;

Cancer risk management, research studies, how to tell your family/children about the familial predisposition, support groups.

More recently we have introduced a peer-support group part of the evening, enabling carriers to share their stories with others in a small group setting. This has been a particularly popular part of the evenings.

DDD - Deciphering Developmental Disorders Study

We are currently recruiting children in to the DDD study, which is a nationwide study run by the Wellcome Trust Sanger Institute. It is using new genetic techniques (including exome sequencing) to improve diagnosis and to increase our understanding of developmental disorders. It aims to recruit 12,000 children from around the UK with an undiagnosed developmental disorder via the 23 Regional Genetics Centres.

DDD offers families the chance of receiving a much sought after diagnosis for their child. A diagnosis may help with planning future management and care for a child as well as helping predict the risk of them having another affected child. Without a diagnosis this can be very difficult.

DDD will also contribute greatly to increasing clinicians' and scientists' knowledge of how and why these disorders occur and produce a forum to share this information and enable further future research.

North East Thames Cancer Genetics Referral Guidelines

Great Ormond Street Hospital
Tel: 0207 762 6831/Fax: 0207 813 8141

In general the person being referred should be affected or have an affected first degree relative. All affected relatives should be on the same side of the family.

Breast Cancer

***** first degree relative if related through a woman, second degree relative if related through a man.

- 2 first/second* degree relatives diagnosed at average age of 50 or less
- 3 first/second* degree relatives diagnosed at average age of 60 or less
- 4 relatives any age
- 1 first degree relative with bilateral breast cancer diagnosed age 50 or less (or any age plus 1 first/second* degree relative with breast cancer diagnosed aged 60 or less)
- Male breast cancer **plus (**on same side of family) 1 first/second* degree relative with breast cancer

Ovarian Cancer

1 relative diagnosed at any age **plus on same side of the family**

- additional relative with ovarian cancer; any age
- **Or** 1 first/ second* degree relative with breast cancer diagnosed age 50 or less
- **Or** 2 first/second* degree relatives with breast cancer diagnosed at average age of 60 or less

Anyone of **Ashkenazi Jewish heritage** affected with or 1 first degree relative with breast cancer or ovarian cancer any age

Bowel Cancer

- 1 family member with colorectal cancer under 50 years of age
- 2 family members affected with colorectal cancer both under 60 years of age
- 3 relatives affected with colorectal cancer or related cancer † at any age (one should be first degree relative of the other two).
- Familial Adenomatous Polyposis (FAP) or other polyposis syndromes
- Hereditary Non Polyposis Colorectal cancer (HNPCC)/Lynch or molecular indication (MSI or IHC results).
- More than 10 adenomatous bowel polyps, at any age, no additional family history
- More than 5 adenomatous bowel polyps, at any age, with family history of bowel polyps/cancer
- Juvenile or harmatomatous bowel polyps– please contact cancer team to discuss.

† related cancers– endometrial, ovarian, small bowel, ureter, renal, pelvis and stomach cancer

Endocrine Tumours

- All medullary thyroid carcinomas
- All MEN (multiple endocrine neoplasia) families
- Phaeochromocytomas/ paragangliomas under 50 years of age
- Familial hyperparathyroidism or hyperparathyroidism with a related endocrine tumour in another family member
- Familial pituitary tumours
- Isolated childhood onset pituitary tumour (except microprolactinoma) or pituitary macroadenoma under 30

Unusual Cancers

- Sarcoma at younger than 45 years of age
- Childhood adrenal cortical carcinoma
- Complicated patterns of multiple cancer at a young age
- Known single gene cancer syndrome in family (e.g Von Hippel-Landau, Li Fraumeni's)
- In any families with unusual patterns of cancer where there is a suspicion of an hereditary aetiology please contact us to discuss on an individual basis