EUROPEAN SOCIETY OF HUMAN GENETICS

European School of Genetic Medicine

Basic and Advanced Course in Genetic Counselling

Bertinoro, Italy, April 28 – May 3, 2016

Bertinoro University Residential Centre
Via Frangipane, 6 – Bertinoro, Italy

Course Directors:
F. Forzano (Great Olmond Street Hospital, London, UK),
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EUROPEAN GENETICS FOUNDATION

Basic and Advanced Course in Genetic Counselling

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BASIC AND ADVANCED COURSE IN GENETIC COUNSELLING

University Residential Centre, Bertinoro (Italy)
April 28 - May 3, 2016

Arrival: April, 27th

Thursday, April 28th – BASIC

Morning Session

9.00 – 9.45	Introduction to the course, Ice-breaking session, Workshop instructions **F. Forzano , A. Tibben, G. Romeo**
9.45 – 10.45	Genetic services: aims, process and outcomes **C. Patch**
10.45 – 11.15	**Coffee Break**
11.15 – 12.15	Inheritance models and risk assessment **D. Turchetti**
12.15 – 13.15	Cytogenetics: current status and future perspectives **J. Baptista**
13.15 – 14.15	**Lunch Break**

Afternoon Session:

14.15 – 15.15	Molecular analysis: old and new diagnostic tools **M. Iascone**
15.15 – 16.15	Prenatal diagnosis: scenarios and issues **F. Forzano**
16.15 – 16.45	**Coffee break**
16.45 – 17.45	Cancer genetics: scenarios and issues **D. Turchetti**
17.45 – 18.15	General discussion

Friday, April 29th – BASIC

Morning Session

9.00 – 10.00	Basic concepts on dysmorhology **F. Forzano**
10.00- 11.00	Practical ethics: consent, confidentiality and disclosure **C. Patch**
11.00 – 11.30	**Coffee Break**
11.30 – 12.30	Genetics of intellectual disability **F. Forzano**
12.30 – 13.00	Setting the agenda **C. Patch**
13.00 – 14.00	**Lunch Break**

Afternoon Session:

14.00 – 15.30	Concurrent Workshops
15.30 –16.00	**Coffee Break**
16.00 – 17.30	Concurrent Workshops

Saturday, April 30th – BASIC & CROSSOVER

Morning Session

9.00 - 11.00	Concurrent Workshops
11.00 – 11.30	**Coffee Break**
11.30 – 13.00	Concurrent Workshops
13.00 – 14.00	**Lunch Break**

Afternoon Session:

14.00 – 15.00	Introduction to the course Revision of basic Patient-Centered counselling skills **A. Tibben**
15.00 –15.30	**Coffee Break**
15.30 – 17.00	Skills practice (I) **A. Tibben**

Sunday, May 1st – ADVANCED

Morning Session

9.00 – 9.45	Why do we need counselling skills? **E. Razzaboni**
9.45 – 10.15	Skills practice (II) **All faculty**
10.15 – 11.00	Counselling for predictive testing **A. Tibben**
11.00 –11.30	**Coffee break**
11:30 –12.30	Discussion of difficult cases brought by students **All faculty**
12.30 – 13.30	**Lunch break**

Afternoon Session:

13.30 –14.15	Skills practice (III) **All faculty**
14.15 –15.00	Breaking the news: theory **A. Tibben**
15.00 – 15.30	**Coffee break**
15.30 – 16.30	Skills practice (IV) Breaking the news **A. Tibben**

Monday, May 2nd – ADVANCED

Morning Session:

9.00 – 9.15	Family dynamics: theory **E. Razzaboni**
9.15 – 10.30	Family dynamics: awareness **E. Razzaboni**
10.30 – 11.00	**Coffee break**
11.00 – 12.15	Genetic screening and testing in children **C. Patch**
12.30 – 13.30	**Lunch break**

Afternoon Session:

13.30 –14.15	Grief and loss issues – theory **A. Tibben**
14.15 – 15.00	Psychological issues in antenatal screening and testing **E. Razzaboni**
15.00 –15.30	**Coffee break**
15.30 – 17.00	Cross cultural perspectives **K. Kharusi**

Tuesday, May 3rd – ADVANCED

Morning Session:

9.00 – 10.00	Discussing difficult issues with clients **All faculty**
10.00 – 11.00	Skills practice (V) **All faculty**
11.00 – 11.30	**Coffee break**
11.30 – 12.30	The Counsellor end: self-awareness tools, occupational stress and burnout syndrome **E. Razzaboni**
12.30 – 13.30	**Lunch**

Afternoon Session:

13.30 –14.30	Using supervision effectively **A. Tibben and E. Razzaboni**
14.30 – 15.00	Evaluation, Closing Remarks, Farewell **F. Forzano, G. Romeo, A. Tibben**

Thursday, April 28

Aims Processes and Outcomes of Genetic Counselling

C Patch
Dept of Clinical Genetics, Guys and St Thomas NHS Foundation Trust London
Florence Nightingale School of Nursing and Midwifery. KCL. London

In the past twenty years the demand for clinical genetic services and genetic counselling has increased enormously alongside the major advances in genetic science. Although accurate genetic counselling relies on a firm medical diagnosis, accepted definitions of genetic counselling also emphasise the educative and counselling components.

Advances in the science related to genomic medicine also raises questions about what is genetic counslling and how do our existing models fit with the knowledge genereted through the scientific developments.

There may seem to be little in common between the science of genetics and counselling. Individual genetic counselling clients will often have questions and concerns about a genetic illness that could be important for themselves or their families. They may not only require information that is technically correct, but also some assistance to understand the information provided and to appreciate its relevance to their own lives, values and emotional reactions.

Genetic counselling can act as a bridge between the science of genetics and the understanding and feelings of its clients. Genetic counselling is a communication process that deals with the occurrence, or risk of occurrence, of a (possibly) genetic disorder in the family. The process involves an attempt by appropriately trained person(s) to help the individual or the family to

> (1) understand the medical facts of the disorder;
>
> (2) appreciate how heredity contributes to the disorder and the risk of recurrence in specified relatives;
>
> (3) understand the options for dealing with the risk of recurrence;
>
> (4) use this genetic information in a personally meaningful way that promotes health, minimizes psychological distress and increases personal control;
>
> (5) choose the course of action which seems appropriate to them in the view of their risk and their family goals, and act in accordance with that decision,
>
> (6) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder' (Eurogentest 2009).

Genetic counselling should always be based on a diagnosis that is as accurate as possible. This increasingly involves interpretation of complex genetic analyses. The activities that take place within a counselling session include:

Taking a family medical history which is necessary to provide reliable information

Giving and interpreting genetic information with skill, presenting it in a non-judgmental way.

Supporting the patient or client particularly when they are making difficult decisions or at times of stress related to their genetic issues.

In the UK, most genetic counselling in provided in Regional Genetic Services by multi-disciplinary teams including medically trained specialist clinical geneticists and genetic counsellor colleagues. These colleagues are supported by laboratory scientists. Genetic diagnoses are usually made by clinicians - clinical geneticists or other medically qualified doctors - but clients can often be helped by discussions with non-medical genetic counsellors. In this session we will introduce the framework of genetic counselling and put it into context with the aims of this course.

Resources

Eurogentest Recommendations for genetic counselling related to genetic testing.

http://www.eurogentest.org/fileadmin/templates/eugt/pdf/guidelines_of_GC_final.pdf (accessed 5th April 2015).

Middleton A, Hall G, Patch C Genetic Counselors and Genomic Counselling in the United Kingdom Molecular Genetics & Genomic Medicine 2015 3(2) p 79–83

Inheritance models and risk assessment

Daniela Turchetti
UO Medical Genetic, Univ. of Bologna,
Policlinico S. Orsola-Malpighi,
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Single-gene disorders (Mendelian disorders) are caused by mutations in one or both members of a pair of autosomal genes or by mutations in genes on the X, or rarely on the Y, chromosome (sex - linked inheritance). These disorders show characteristic patterns of inheritance in family pedigrees. If a disease is expressed in the heterozygote, then it is dominant, whereas if it is only expressed in the homozygote, it is recessive.

In autosomal dominant disorders, It is equally likely that a child will receive the mutant or the normal allele from the affected parent, therefore there is a 1 in 2 or 50% chance that each child of a heterozygous parent will be affected. Conversely, in autosomal recessive disorders, affected individuals have inherited two mutant alleles (one from each parents). The commonest situation is that the parents of the affected individual are healthy carriers (heterozygotes) for the mutation: the risk of having an affected child is 25% for every pregnancy. In X-linked recessive inherited disorders, heterozygous females are usually clinically unaffected (carriers) but may pass the

condition to the next generation. One-half of the daughters of a female carrier will be carriers as well, while one-half of the sons will be affected. These disorders are never transmitted by unaffected males. On the contrary, affected males transmit the X-chromosome, therefore the mutation, to all the daughters, while their sons, who receive the Y-chromosome, never inherit the mutation.

Although the large majority of single gene disorders follows typical Mendelian mechanisms of inheritance, there are several genetic conditions that have inheritance mechanisms that, while essentially Mendelian in nature, are atypical, for instance with genetic anticipation, incomplete penetrance, variable expression or pseudoautosomal inheritance. These and other non-classical Mendelian mechanisms, as well as non-Mendelian inheritance, will be presented in the lecture.

Cytogenetics: current status and future perspectives

J. Baptista
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The early days

The study of chromosomes and their structure is the subject of cytogenetics. The year 1956 marked the beginning of modern human cytogenetics when Tjio and Levan defined the number of chromosomes in man as 2n=46 and Ford and Hamerton confirmed this finding. In 1959, the first chromosome abnormalities were described: Jacobs and Strong reported an additional chromosome X in a case of Klinefelter syndrome, Lejeune *et al.* showed the presence of an extra chromosome 21 in Down syndrome and Ford *et al.* demonstrated a 45,X karyotype in a case of Turner syndrome. These first discoveries were rapidly followed by other cytogenetic reports that established the medical applications of cytogenetics.

Because of the limitations of the early methods used in clinical cytogenetics, numerical abnormalities, in which the chromosome complement is different from 2n=46 were the first type of chromosome abnormality described. However, the existence of a number of large structural rearrangements, including Robertsonian translocations, was also documented at this time, but it was the introduction of chromosome banding techniques that initially allowed much more detailed characterisation of structural chromosome rearrangements.

Fluorescence in situ hybridisation (FISH)

Although conventional cytogenetics is an essential technique in the identification and characterisation of chromosome rearrangements, this methodology has a resolution limited by the capacity of the human eye of ~3 to 5Mb. More recently, the advent of FISH has permitted the study of chromosomes at resolutions significantly higher than afforded by conventional cytogenetic analysis. The technique is based on the hybridisation of a labelled DNA or RNA probe to patient genomic DNA. Radioactive isotopic labels were used initially, but were later replaced with fluorochromes, rendering the technique safer and easier to use. Furthermore, the availability of fluorochromes of different colours enables the testing of more than one probe simultaneously. This technical advance allied to the increasing accessibility of probes generated by the Human Genome Project promoted significantly the use of FISH.

Array Comparative Genomic Hybridisation (Array CGH)

Recently, the application of DNA probes to microarrays has emerged as a powerful technology in genetics studies. Array CGH enables the detection of copy number changes by competitively hybridising differentially labelled test and reference DNA to arrays of spotted and mapped clones. Thus, the technique allows the rapid screening of the whole genome at a resolution determined by the density of the markers spotted onto the array.

Next Generation sequencing (NGS)

NGS enables a fast and cost effective way to determine the whole DNA sequence of an individual, hence allowing for the identification of the whole catalogue of DNA variants in a given genome. Alternatively, a NGS assay might be designed in order to target not the whole genome, but specific genomic regions, for example the exons and in that case the assay is named exome sequencing. Irrespective of the chosen design for a NGS experiment, the final outcome is that data on all variants present in a DNA sequence is obtained and quite often the amount of data generated is overwhelming.

Future perspectives

The field of Human Genetics has greatly benifited from technological advances. At this point in time, it seems obvious that the main challenge faced by human geneticists rests with the interpretation of the data obtained, specially by array CGH and NGS assays. Although a set of criteria has been put in place to aid in this interpretation in many cases a definitive answer just cannot be given to patients. Hopefully, we will be in a position to tackle these shortcomings when a large enough number of individuals' genomes has been analysed. Thus far, the study of normal individuals has demonstrated that human genome variation is considerable and further studies are necessary to help to gauge its full contribution for human diversity and susceptibility to disease.

Molecular analysis: old and new diagnostic tools

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Technological advances in molecular genetics had signed the pace of progress in our ability to diagnose genetic diseases. Molecular genetic tests usually study single genes or short lengths of DNA to identify variations or mutations that lead to a genetic disorder. Now this approach is changing due to the recent introduction of new sequencing technology in clinical practice. The lesson will focus on old and new techniques used to diagnose diseases caused by different pathogenetic mechanisms, in particular:

- diseases caused by dynamic mutations (triplets expansion in the promoter region of the FMR1 gene, Fragile X syndrome)
- diseases caused by imprinting defects (methylation-specific PCR for evaluation of methylation status of a DNA region and use of STR markers to detect deletions or uniparental disomy, Prader-Willi /Angelman syndrome).

- diseases caused by total or partial deletions/duplications of single genes (MLPA, multiplex ligation-dependent probe amplification, Alagille Syndrome).
- diseases caused by known point mutations (single bases substitutions or indels) detectable by targeted methods (real-time PCR for detection of single known mutation or reverse dot blot and similar for arrays of known mutations, Cystic Fibrosis).
- diseases without hotspots mutations detectable only by DNA sequencing.

At the basis of almost all molecular analyses, there is the **polymerase chain reaction** (**PCR**). This is a technology used to amplify a single or a few copies of a piece of DNA across several orders of magnitude, generating thousands to millions of copies of a particular DNA sequence. Developed in 1983 by Kary Mullis, PCR is now a common and often indispensable technique used in research and diagnostic labs for a variety of applications. The method relies on thermal cycling, consisting of cycles of repeated heating and cooling of the reaction for DNA melting and enzymatic replication of the DNA. Primers (short DNA fragments) containing sequences complementary to the target region along with a thermostable DNA polymerase are key components to enable selective and repeated amplification. As PCR progresses, the DNA generated is itself used as a template for replication, setting in motion a chain reaction in which the DNA template is exponentially amplified. PCR can be extensively modified and is at basis of the majority of tests used in a molecular genetics lab. PCR and capillary electrophoresis allows the detection of triplets' expansion or by amplification of chromosome specific markers the detection of parental origin of chromosomes to detect uniparental disomy. One o the modifications of PCR, is represented by **Methylation-specific PCR** (**MSP**), used to detect methylation of CpG islands in genomic DNA. DNA is first treated with sodium bisulfite, which converts unmethylated cytosine bases to uracil, which is recognized by PCR primers as thymine. Two PCRs are then carried out on the modified DNA, using primer sets identical except at any CpG islands within the primer sequences. At these points, one primer set recognizes DNA with cytosines to amplify methylated DNA, and one set recognizes DNA with uracil or thymine to amplify unmethylated DNA. MSP is, for example, used in Prader-Willi/Angelman syndrome genetic testing.

MLPA (**Multiplex Ligation-dependent Probe Amplification**) is a multiplex PCR method detecting abnormal copy numbers of genomic DNA segments (usually exons). Although for most hereditary conditions, (partial) gene deletions or duplications account for less than 10 % of all disease-causing mutations, for many other disorders this is 10 to 30% or even higher still. The inclusion of MLPA in clinical settings can therefore significantly increase the detection rate of many genetic disorders. Although MLPA is not suitable for genome-wide research screening, it is a good alternative to array-based techniques for many routine applications. Typical for MLPA is that it is not target sequences that are amplified, but MLPA probes that hybridise to the target sequence. The MLPA reaction can be divided in five major steps: 1) DNA denaturation and hybridisation of MLPA probes; 2) ligation reaction; 3) PCR reaction; 4) separation of amplification products by electrophoresis; and 5) data analysis.

During the lesson, particular emphasis will be given to old and new **sequencing techniques** that are revolutionizing the approach to genetic testing. Sequencing technologies have evolved rapidly over the past 5 years. **Semi-automated Sanger sequencing** has been used in clinical testing for many years. It is based on chain-termination method developed by Frederick Sanger in 1977. The Sanger method was soon automated and was the method used in the first generation of DNA sequencers and is still considered the gold standard of clinical sequencing. However, its limitations include low

throughput and high cost, making multigene panel laborious and expensive. Recent technological advancements have radically changed the landscape of medical sequencing. **Next-generation sequencing (NGS)** technologies utilize clonally amplified templates, which are then sequenced in a massively parallel fashion. This increases the throughput by several orders of magnitude decreasing the cost of sequencing. NGS technologies are now being widely used in clinical setting. Three main levels of analysis, with increasing degrees of complexity, can now be performed via NGS: disease-targeted gene panels, whole-exome sequencing (WES), and whole-genome sequencing. All have advantages over Sanger sequencing in their ability to sequence massive amounts of DNA, yet each has challenges for clinical testing.
For example, the results of NGS genetic tests are not always straightforward, which often makes them challenging to interpret and explain. During the lesson an overview of current limits of clinical application of NGS will be addressed.

Prenatal diagnosis: scenarios and issues

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All the couples face a 3% risk of having children with congenital anomalies.
Prenatal screening refers to different kind of testing offered during pregnancy, which include a combination of US scans, metabolites dosage and fetal DNA analysis on maternal blood, aimed to investigate a portion of this risk, primarily related to the more frequent chromosome aneuploidies, as trisomy 21, and malformations, as neural tube defects.

Prenatal diagnosis refers to testing offered to selected couples who are at risk of specific disorders, or to pregnancies identified at high risk through prenatal screening.
The aim of prenatal screening and diagnosis is to identify fetal anomalies in order to drive the management of the pregnancy and to allow the parents to make autonomous reproductive choices.
Parental personal views and feelings, cultural and ethical issues, time constraint and uncertainties on prognosis make this setting particularly challenging.

The explosion of available techniques of genome analysis is now opening up new scenarios in which a thorough fetal genome could be prenatally investigated, thus enriching the debate on which the scope of prenatal testing should be as well as the ethical issues implicated.

Cancer genetics: scenarios and issues

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Cancer is always a genetic disease, as it is the result of multiple genetic defects in cells. In the majority of cases, the accumulation of genetic changes in a tissue is random, and in this case the tumour is termed sporadic. In a fraction of cases, however, all the cells of the body carry an inborn genetic defect, which increases the chance that certain tissues would become cancerous. This type of cancer susceptibility can be passed down to the offspring, and cancer occurring in such predisposed individuals is therefore regarded as "hereditary".
Observation of large populations of individuals revealed that as much as 5-10% of cancer cases show marked familial clustering suggesting hereditary cancer predisposition. This is a small fraction of the total cancer burden, if compared to those attributed to dietary risk factors (35%) and to smoking (30%). Nevertheless, if one estimates that 5-10% of the most common cancers, like breast, colorectal and prostate cancer, are associated with a genetic predisposition, it becomes clear that the absolute number of hereditary cancer cases is significant. Moreover, the identification of cancer genetic syndromes allows for the identification of individuals at increased risk, who can benefit from specific prevention strategies.

Genes involved in hereditary cancer predisposition belong to three main classes:
1. **Oncogenes** are genes that are normally involved in cell growth and proliferation and cause cancer when they are over-expressed, amplified, or mutated (gain of function).
2. **Tumour suppressor genes**, on the other hand, normally regulated cell growth, and only result in malignant progression when their negative control is impaired (loss of function).
3. Similarly to tumour suppressor genes, also **DNA repair genes** cause cancer predisposition through a loss of function, which allows for multiple genetic defects to accumulate in the cell genome, leading to the malignant phenotype.

Unlike oncogenes, a monoallelic mutation of which is sufficient to cause cancer, tumor suppressor and DNA repair genes generally require that both the alleles are mutated for cancer to develop.
There are very few instances of oncogenes involved in hereditary cancer syndromes: RET mutations cause Multiple Endocrine Neoplasia 2, while mutations in MET are responsible for Familial Papillary Renal Carcinoma Syndrome. Conversely, mutations in tumor-suppressor genes account for the majority of cancer syndromes, such as the Breast Ovarian Cancer Syndrome, caused by mutations in BRCA1 and BRCA2 genes, and Hereditary Melanoma, caused by mutations in the CDKN2A gene. Talking of repair genes, a dysfunction of the mismatch repair caused by mutations in one of the responsible genes results in Hereditary Non-Poliposis Colorectal Cancer (HNPCC, also known as Lynch Syndrome).
Recognizing the hereditary cases among all cancer patients is sometimes easy, when the family history is highly suggestive, but can be difficult in many cases. The usefulness of specific familial and/or clinical features in the identification of inherited cases will be discussed in the lecture. In addition, for some cancer types, such as breast and colorectal cancer, probabilistic models are available to predict the probability of mutations in specific genes and to assess individual cancer risk.

Genetic testing may find the causative mutation in a number of families, which helps identify asymptomatic carriers in the family; nevertheless genetic heterogeneity (rare or undiscovered genes) and the existence of possible alternative mechanisms of gene alteration (undiscovered type of mutations) hamper the ability of genetic testing to detect the underlying defect. Limitations of testing must be taken into account when counseling people from cancer-prone families and planning prevention and surveillance. In the near future, next-generation sequencing technologies are expected to lead to an enormously increase in the detection rate of genetic tests. At the same time, however, we will face increasing troubles with interpretation and communication of genetic test results, as variants of unknown significance will become more common, a number of "incidental findings" will be detected and penetrance of rare genes will be uncertain.

Whenever the efficacy of available risk-reduction strategies is not definitely demonstrated, a non-directive approach should be adopted in counselling patients at increased risk for cancer. The purpose of counselling may include helping the individual explore feelings about his or her personal risk status and make a healthy adjustment to that risk status. Either alone or in consultation with a mental health provider, professionals offering cancer genetic counselling attempt to assess whether the individual's expectations of counselling are realistic and whether there are factors suggesting unusual risk of adverse psychological outcomes after disclosure of risk and/or genetic status. To limit the chances of adverse consequences of risk assessment and communication, in addition to a continued follow-up by the counsellor, the availability of psychological support, preferably provided by mental health professionals with experience in cancer genetics, is recommended.

References:

- Offit K.: Clinical Cancer Genetics. Risk Counselling and Management, Wiley-Liss 1998
- Genetic Cancer Risk Assessment and Counseling: Recommendations of the National Society of Genetic Counselors Journal of Genetic Counseling, Vol. 13, No. 2, April 2004
- http://www.nci.nih.gov/cancerinfo/pdq/genetics/overview
- http://www.nci.nih.gov/cancerinfo/pdq/genetics/risk-assessment-and-counseling
- Robson. M. and Offit. K.: Management of an inherited predisposition to breast cancer. N Engl J Med 2007; 357:154-62
- Hendriks YMC et al.: Diagnostic approach and management of Lynch Syndrome (Hereditary Nonpolyposis Colorectal Carcinoma): A Guide for Clinicians. CA Cancer J Clin 2006; 56: 213-225
- Stadler ZK et al: Cancer Genomics and Inherited Risk. J Clin Oncol 2014; 32: 687-698.

Friday, April 29

Basic concepts on dysmorphology

F. Forzano
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The term “Dysmorphology” has been coined by Dr. David W. Smith in the 1960's to generally define the study of human congenital malformations, particularly those affecting the “morphology” (anatomy) of the individual.
A few years later, Dr. Jon Aase, a former Dr Smith’s student, elaborated much more this concept and stated that "As a scientific discipline, Dysmorphology combines concepts, knowledge, and techniques from the fields of embryology, clinical genetics and pediatrics. As a medical subspecialty, dysmorphology deals with people who have congenital abnormalities and with their families."

The clinical examination of the morphology of referred patients has proved essential for the delineation of hundreds of syndromes and has been a key tool for the discovery of many “disease genes”. A structural defect is in fact an inborn error in morphogenesis, and the study of these anomalies ultimately lead to an extended knowledge on genetic mechanisms which regulate normal embryonal development too.

The dysmorphological assessment relies on a careful analysis of congenital anomalies. While major malformations are obvious at birth and usually lead to a prompt referral for a medical evaluation, minor malformations have no clinical consequences and can easily be neglected. However, the recognition of these minor malformations might be the essential clue for the detection of a genetic condition, which can allow to establish the more appropriate intervention for the child and the whole family.

Since the evaluation of minor malformation is largely subjective, new computer-based 3D techniques have recently being developed to analyse facial features in an objective, operator-independent way and to assist clinical training in pattern recognition.
Databases like OMIM, London Medical Databases, Possum are useful tools commonly used by dysmorphologists to achieve a diagnosis in difficult cases.
With the introduction of new cytogenetic and molecular testing, the traditional path from phenotype to genotype in dysmorphology has now become a two-way road.
In fact large scale testing of patients with developmental problems has brought to the identification of several ‘new’ microdeletion/duplication syndromes through so called ‘reverse dysmorphology’, that is, using a genotype to phenotype approach.

The combination of all these new tools and techniques makes Dysmorphology nowadays a very exciting and dynamic branch of Clinical Genetics. The parallel improvement in both phenotyping and genotyping and their continuous reciprocal interaction will ultimately lead to a profound

knowledge on pathogenesis of a number of diseases and also on physiological development and functioning.

Practical ethics - consent, confidentiality and disclosure

C. Patch
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Principles regarding consent for procedures and protecting the confidentiality of medical information are enshrined in codes governing ethical practice. They are also subject to statutory oversight which may vary according to the area of administration. It can be argued that medical genetics is no different from other medical specialties. However the practice of clinical genetics may give rise to situations where issues of consent and confidentiality do require special consideration. In relation to consent for procedures the key aspects are that i) the person understands the nature and risks of the procedure to which they are consenting and ii) that the person gives consent without coercion.
In this session we will consider cases where there may be special issues relating to consent and confidentiality. The text below is adapted from 'Applied genetics in health care'. In genetic healthcare settings, consent most often relates to:

1.Taking a family history
Consent can generally be assumed if the proband provides the information requested, providing that the process and reason for taking the pedigree have been explained. However, when using the pedigree to counsel other family members, the confidentiality of the original proband must be respected. For this reason, it may be appropriate to take a new pedigree when seeing a different branch of the family.

2. Obtaining specific medical history from the proband and/or other relatives
It is frequently necessary to request medical notes on the proband in order to advise him or her properly, consent must be sought to view or request medical records. The purpose of viewing records of other family members must be explained to them and written consent obtained.

3. Obtaining blood or tissue samples
Permission to take a sample must be explicitly given by the client. This is sometimes written consent, but if the procedure has been explained the co-operation of the client in giving the sample is usually deemed to be evidence of consent. For example, if a client lifts his sleeve and presents his arm after being asked to consent to a blood sample, this would be evidence that the client has given consent.

4. Performing genetic tests
The exact nature of the tests and the implications of the result must be explained to the client . It is good practice to give the client written information as well as a verbal explanation, and written evidence of consent must be recorded. Risks associated with genetic testing might include the

discovery of false paternity, this should be mentioned if a possibility. Other aspects of consent for genetic tests include whether consent is given for the sample to be stored and the possible outcomes of the test. Separate consent should be obtained for use of the sample in research and to share the results with relatives in the process of their own testing.

5. Issues arising from Genome Sequencing

The advent of whole genome approaches to genetic analysis in the research setting and in the clinic has led to number of different analyses of how to approach the issue of 'health related' actionable incidental findings. This discussion is still ongoing with different approaches being suggested. The spectrum ranges from a bio-informatically targeted approach to analysis based on the clinical question which minmises the possibility of uncovering the information unrelated to the genetics test, to an opportunistic screening approach deliberately targeting genetic variants that have actionable health consequences. Whatever approaches are used there are challenges for consent and confidentiality.

Confidentiality

Confidentiality of personal information is a basic tenet of healthcare and is considered so important to the rights of the client that it is enshrined by law in many countries. However, there may be provision under some statutes for the healthcare professional to disclose the client's confidential medical information, if not disclosing would result in serious but avoidable harm to others. This is the case in UK law. A good example covered in law would be where a person had a serious infectious disease that was putting others in the community at risk.

In a genetic healthcare setting, the situation may be complex, as the information about the genetic structure of one individual may (and often does) have implications for other family members. Where this occurs, the proband is usually encouraged to share the information with relatives who may be affected, especially if screening or treatment is available that would reduce the health risk. It is usual to offer support in the form of written information that can be given to relatives and contact details so that they can seek more information and guidance from the genetics team if they wish.

When an individual refuses to share information with relatives, there is always an underlying reason that might not be obvious to the practitioner. The situation is rarely urgent, and effort spent in gaining the proband's confidence and allowing time for psychological adjustment to their status can often be helpful in enabling the proband to share the information. However, this is not always the case and then the decision about whether to break confidentiality may arise.

Resources

American College of Medical Genetics and Genomics ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing
Update published April 1st 2014
https://www.acmg.net

Joint Committee on Medical Genetics Consent and confidentiality in genetic practice: Guidance on genetic testing and sharing genetic information. A report of the Joint Committee on Medical Genetics 2nd edition 2011 (accessed 14th April 2014) http://www.bsgm.org.uk/media/678746/consent_and_confidentiality_2011.pdf

Middleton A et al Position statement on opportunistic genomic screening from the Association of Genetic Nurses and Counsellors (UK and Ireland) (2014) Eur J Hum Genet. 2014 Jan 8. doi: 10.1038/ejhg.2013.301

PhGFoundation Clinical whole genome analysis: delivering the right diagnosis http://www.phgfoundation.org/reports/15237/ (accessed 14th April 2014)

Skirton H, Patch C and Williams J (2005) Applied Genetics in Health Care. Taylor Francis. Abingdon 2005

United States Department of Health and Human Services. Office for Civil Rights- HIPPA. Medical Privacy – National standards to protect the privacy of personal health information (accessed 14th Aril 2014) http://www.hhs.gov/ocr/hipaa/finalreg.html

Genetics of intellectual disability

F. Forzano
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Intellectual Disability (ID) is a common condition which affects 1-3% of people worldwide, and is currently defined as "an impairment of general mental abilities that impact adaptive functioning in conceptual, social and practical domains" with onset in the developmental period.
A genetic cause can be found roughly in a half of the cases, being much more likely as the IQ progressively decrease.
Among the genetic causes it is possible to make a gross distinction between multiple genes defects and single genes defects.

The first group include genomic imbalance (deletion or duplication) which involves various contiguous genes and which can be identified through standard karyotyping (resolution 3-5 Mb) or molecular karyotyping (FISH and array-CGH, resolution 25Kb-1Mb). The contribution of each of the genes located within the critical region to the phenotype can be different, and sometimes it is possible to identify one major gene responsible for the core phenotype. Almost 10-15% of the patients affected by genetic related ID carry an overt or subtle chromosomal abnormality.

The second group includes a few hundreds of genes spread throughout all chromosomes. More than 200 genes are located on the X chromosome. Among these, FMR1 is responsible for the most frequent inherited form of MR, Fragile-X syndrome, which affects 1 in 6000 people. On the contrary, all the other genes individually account for a very small proportion of ID disorders.
The functions of the genes involved in ID can be very diverse and include the structure, the function or the metabolic environment of neurones.

To search the causes of ID is important for many reasons: to define a prognosis, to start a proper care plan, to provide a specific recurrence risk and to get a proper support to the family.

Guidelines on the evaluation of mental retardation have been established through Consensus Conferences, one of the foremost has been from the American College of Medical Genetics in 1997. As the research advances, new genes are identified and new techniques available, thus improving both knowledge and tools that can drive clinicians in the diagnostic process. It's now emerging that ID can be the end result of a number of different abnormal pathways, no-one of them overriding the others, which underlie the huge complexity of our intellectual processing. So unraveling the causes of ID phenotypes will ultimately be important to understand how the brain develops and works and eventually to find out possible specific treatments.

Saturday, April 30

Revision of basic Rogerian Counselling skills

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In all healthcare settings, the use of counselling skills is helpful in enabling the client to discuss their health concerns and engage in shared decision-making about investigation and/or treatment. The core competencies for genetic health professionals include eliciting the client's concerns and exploring the psychosocial influences that have relevance to the genetic counselling for each family (AGNC, 2004). However, there will be sessions when the need to use counselling skills is more apparent, such as when the client is making difficult decisions or during periods f adjustment to changed circumstances. Active counselling may be undertaken by genetic counsellors who are appropriately trained to assist the client when the psychosocial issues are impeding adjustment to their genetic situation and therefore adversely affecting the client's quality of life.

Rogerian, or Person-centred, counselling

In every session of genetic counselling, it is important that clients are able to express their own concerns, questions and reactions, and to feel that the genetic practitioner had heard and addressed them appropriately. One model that is suitable for counselling in a genetic counselling context is the person-centred model based on the seminal work of Carl Rogers (1961). The central tenet of the model is the belief that each person has the ability to solve his/her own problems an work through difficult situations using one's own resources. Support from another person enables the client to explore the situation in a safe emotional environment.

The aim of person-centred counselling is to facilitate the client to achieve self-actualisation through enhancing self-belief. The counsellor aims to hold the client in unconditional positive regard, and to demonstrate this. The empathic relationship is central to the counselling work.

Person-centred counselling is very appropriate in a genetic healthcare setting, as the practitioner does not profess to be 'an expert', who can solve the client's problems, but rather a supporter whose role is to reinforce the client's self-belief. Rogers described the 'core conditions' necessary for helpful relationship.

Core conditions

Genuineness

The counsellor is real to him or her self and to the client. To achieve this, the counsellor requires a considerable degree of self-awareness and a belief in the equality of the client.

Empathy

On description of empathy is being able to 'walk in the other person's shoes'. Whereas sympathy involves feeling sorry for the other person, empathy is more connected with trying to understand how the client feels, and communicating that understanding.

Warmth

Understanding the client is not facilitative unless that can be conveyed. The 'golden standard' for the person-centred counsellor is the ability to hold every person in unconditional regard. Whilst this itself a challenge, it helps to reduce value judgements of the client and therefore increases the likelihood that the client will free to make the decision that is best for them.

Basic counselling skills that we will discuss and practice are:

- Open questions
- Reflections of feelings
- Paraphrases of content
- Summaries of the dialogue
- Non-verbal communication
- Silence

Sunday, May 1

Why do we need counselling skills?

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Bad news may be defined as "any information which adversely and seriously affects an individual's view of his or her future". Bad news is always, however, in the "eye of the beholder," such that one cannot estimate the impact of the bad news until one has first determined the recipient's expectations or understanding. Different models of communication will be explored and learned to achieve communicational, emotional and relational skills. Furthermore, specific difficult issues in genetics will be analysed, such as: risk perception, autonomous decision, emotional impact and cultural differences.

Counselling for predictive testing

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Genetic counselling has been perhaps the most important way of assisting families with a hereditary disease in managing the consequences of the disease, and in helping individuals at-risk to find creative solutions for their problems. The increased awareness of the genetic aspects of a disease, and genetics in general, together with the more widespread availability of genetic centres have contributed to a more appropriate approach for those who ask for assistance in making important life decisions. Clinicians involved with families with a hereditary disease may prefer to refer their patients to a clinical genetics centre to address the genetic questions. The way such questions are dealt with can have a profound impact on the attitude of individuals at risk, their partners and children, and on further relatives. Before the availability of predictive or susceptibility testing, general counselling of the genetics of a hereditary disease was the most important issue that led individuals at risk to visit the genetic counsellor. Currently, people often apply for general genetic counselling when they have only recently first learned of a hereditary disease in their family, although many of them come with the intention to discuss predictive or prenatal testing. Most people seen for genetic counselling regarding a hereditary disease are the asymptomatic children of an affected patient, seeking reassurance for themselves and their (future) children. Sometimes people apply for predictive testing because they have the opinion that a test result might solve their psychological or family problems. Those professionals who have much experience with general counselling and predictive testing know that alternative ways of coping with personal risks and, subsequently, life decisions might be preferable in some cases.

Genetic counselling involves a process of consultation by which information is imparted to individuals or families affected by or at risk for a genetic disorder. It includes information on the nature of the disorder; the size and extent of genetic risks; the options, including genetic testing, that may help clarify the risks; the available preventive and therapeutic measures, and the provision of psychological, social and practical support. In the context of genetic testing it may include responding to the concerns of individuals referred and their families, discussing the consequences of a test, and enabling them to choose the optimal decision for themselves, but not determining a particular course of action (American Society of Human Genetics 1975). The definition emphasises the two-way nature of the interaction between the test candidate and the counsellor. Moreover, counselling is considered as a process, taking place over a period of time. This process allows the assimilation of the potentially distressing information regarding diagnosis, prognosis, risk, emotional reactions, family dynamics etc. The counselling process allows attention for the autonomous decisions taken by the test candidate. The appropriateness of the decisions can be discussed and weighed extensively. This all requires ‘appropriately trained persons’ which implies special knowledge and skills distinct from those needed in other medical and counselling interactions (Platt-Walker 1998).

Individuals at risk for HD often come for genetic counselling to discuss aspects of the disorder they find difficult to deal with. Exploring with them their experiences, emotional responses, goals, cultural and religious beliefs, financial and social resources, family and interpersonal dynamics, and coping styles has become an integral part of the counselling process. Many individuals at risk with life long experience with a specific hereditary disease have no full awareness of how the disorder has influenced their psychological make up. An experienced counsellor must be able to recognise

and bring forth these responses. He or she can identify normal and maladjusted responses, reassure candidates that their reactions are normal, prepare them for the near future, new issues and emotions that may come up, and help them to mobilise the resources needed to encourage coping and adjustment.

A central assumption of genetic counselling has been the non-directive approach. This assumption is often misunderstood in a way that non-directiveness does not mean that the counsellor should by no means express their personal views, opinions or feelings (Kessler, Kessler et al. 1984; Djurdjinovic 1998). An individual at-risk can expect that the counsellor is willing to provide some guidance when needed to enable him or her to proceed in his own process of consideration. Yet, it requires from the counsellor a level of introspection and awareness of his or her personal feelings and interests in order not to be coercive. The lack of treatment options and future perspectives may facilitate the psychological defences of professional persons such as denial and displacement of responsibility. Families can be threatening to those professionals who have difficulties in working with conditions that cannot be cured. Although the defences protect professionals from the difficult and unsettling task of providing genetic counselling to healthy relatives at risk, they may prevent caregivers from establishing a relationship that is characterised by confidentiality, respect for autonomy and empathy (Martindale 1987). Permanent education and increase in awareness of the psychodynamics involved may lead to creative and constructive thinking about the current deficiencies in care and counselling services provided for families with a hereditary condition.

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Breaking the news

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One of the most challenging tasks for the genetic counsellor is to disclose results of genetic testing. Individuals may be waiting for disclosure of results of predictive or susceptibility testing, or waiting for test results for their children, or waiting for the outcome of prenatal testing. Any test outcome will evoke strong emotions. When disclosing test results counsellors may balance between facing reality and providing hope. Anticipating untoward, uncontrolled reactions in counselees may result in delay of disclosure by the counsellor. We will discuss the emotional and behavioral responses in both counselees and counsellors, and the prerequisites of breaking the news in genetic counselling.

Monday, May 2

Family dynamics: theory
Family dynamics: awareness

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The last decades, most attention has been given to individual psychological responses to genetic counseling and testing. Yet, an increasing awareness of the psychosocial challenges of genetics for patients, their partners and their core and extended families can be noticed. Genetics is in all senses a family affair. The diagnosis of an inherited condition affects not only biological family members who may themselves be at risk, but also family more generally. For disorders in which carrier, predictive, or confirmative testing is available, core time phases with salient developmental challenges of all family members involved need to be addressed, both pre- and post-testing, including a long-term adaptation phase. Professionals in clinical genetics have scarce training in family dynamics, but recognize the need for more knowledge and skills on these issues to improve clinical practice. In these two lessons students will learn some important Family communication theories and the Family Life Cycle model to let them become aware about how to work with family.

Genetic screening and testing in children

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The necessity of screening and testing children at risk brings along its own sensitivities. Parents naturally wish to ensure the safe and normal development of their offspring. However, in families at risk of genetic diseases, the future of a child can be overshadowed by the chance that life may be shortened or adversely affected by the condition. Families who seek genetic counselling frequently wish to discuss the issue of telling their child about the condition in the family, and informing the child that they are at personal risk. This issue arises whether or not testing is available. The decision to tell may not be clear-cut, as the desire to inform the individual may be juxtaposed with reluctance to cause anxiety in the child.

The general opinion among professionals is that testing for serious adult-onset disorders holds potential for harm.Testing may only justified if onset is expected in childhood or adolescence, and if treatment options are available. Testing removes the individual's future right to make own decisions as an autonomous adult, it removes the confidentiality, expected for any adult undergoing the same test, and it may alter the upbringing and the pattern of relationships within the family ands with peers, with the inclusion of stigmatisation and discrimination. Hence, DNA tests for adult-

onset diseases on asymptomatic children - at parental request - is generally not performed in most genetic centres.
This opinion is reflected in professional guidelines and has led to much debate amongst the professional and patient/family community. However the situation regarding carrier testing or testing for diseases with onset during childhood is more nuanced and most genetic counselling services would aim to work with parents to reach a decision in the best interests of the child and family as regards genetic counselling.
A family life overshadowed by the risk of a hereditary disease will obviously influence the way parents perform their parental tasks. An important task regarding their children is the establishment of a stable and safe environment for the family, which may become difficult if the parents fear the disease. They also have a task in explaining facts and circumstances of the grandparent's disease and their personal risks, which requires openness and courage to discuss these issues with their children. Parents must be able to understand their children's' developmental capacities for coping with their risk and a disease and they must be able to express this understanding. They must assist in tolerating and expressing uncertainty and anxiety, and facilitate the change to new relationships and responsibilities.
Having considered the tasks of parents and children, the tasks of the counsellor can be made more explicit. The counsellor can increase the awareness of how a hereditary disease has specifically affected every member of the family. He or she can help to further discuss the family stories and coping strategies in the family regarding the disease. The counsellor can help to explore the underlying motives of the test request and consider this in the light of the developmental and parental tasks. Genetic counsellors can aid the family in communication of the information in an age and developmentally appropriate way Such work might increase the cohesion in the family and lead to new, constructive, and creative ways to deal with the disease.
Test requests should be considered against the background of the specific age and role-related tasks that each member in a family with a hereditary disease has. The achievement of these tasks may have been extremely burdened by the occurrence of a specific disorder in the family. The test applicant's motives should be explored to enable him or her to make an informed decision. The decision should be made against the personal and family history and future. The decision must be understood as part of the entire family and individual coping mechanisms regarding the risks and the disease.

BBC Radio 4 Inside the ethics committee Genetic testing in children
http://www.bbc.co.uk/programmes/b038hhs7

BSHG Report on the genetic testing of Children
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Grief and loss issues – theory and personal awareness

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Loss is one of the most common experiences of families affected by genetic conditions and so is a key topic in genetic counselling. The loss can take many different forms besides death of family members. People may experience loss of confidence, relationships, work, social life or reproductive futures, to name a few.

Worden (2000) wrote of four tasks of mourning:

Task 1: to accept the reality of loss
Task 2: to work through the pain of grief
Task 3: to adjust to a new environment
Task 4: to emotionally relocate the loss and move on with life.
These tasks follow a cyclical pathway, rather than a longitudinal one and individuals may go backed forth between tasks.
In this session, we will look at grief, the ways in which grief manifests itself and the tasks of mourning. We will discuss how we can support individuals and families in the grieving process and how our own losses affect us as professionals.

Psychological issues in antenatal screening and testing

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Advances in technology increasingly facilitate parental choice with regard to prenatal diagnosis (PND); however, there are many ethical, legal, and social and psychological issues related to the clinical offer of prenatal screening and testing that require consideration. As with other medical procedures, enabling the parents to make an informed choice is integral to good clinical care; however, this can be challenging because of the understandable reluctance of parents to anticipate an abnormality in the fetus and the unpredictable nature of their reaction to the results. Beliefs and values relate to cultural norms, and women from dissimilar cultural backgrounds may show varied psychological responses and attitudes towards information and prenatal counselling. Considering the complexity of the matter, psychological issues will be examined to guarantee adequate counselling.

Cross cultural perspectives; a shared experience from Oman

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Culture is a general term that can describe beliefs, values, custom, social interaction, life interactions and geography shared by a group of people. It reflects the ethnic, historic and linguistic categorization. In genetic counselling, psychosocial, beliefs and morals behind a decision making is usually addressed. It is essential that the counsellor has a cultural awareness to create a cross cultural competence in counselling.

Genetic counselling practice in a highly inbreeding society with consanguineous marriage preference should have an understanding about the cultural beliefs and motives towards consanguinity.

Sultanate of Oman is one of the Middle Eastern countries that has a high rate of consanguinity. The service of genetic counselling is fairly new and is provided with two governmental genetic centers available in the country. To adopt the service within context of culturally driven marriage preferences for cousins, premarital genetic counselling service is provided. It serves a role in enhancing the families' awareness towards prevention of recurrence of a familial genetic disorder. However, challenges exist due to the heterogeneity of the culture and limitations of resources. Creating self-awareness about ethnocultural of the counsellee empowers the counsellor to increase interactive competence for different cultures.

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Tuesday, May 3

The counsellor end: self-awareness tools, occupational stress and burnout syndrome

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Self-awareness is an individual's tendency to pay attention and become aware to his or her own emotions, attitudes, and behaviour in response to specific situations. In the case of counsellors, self-awareness is their insight into how their emotional makeup influences patient care. Conceivably, such insight may improve counsellor' professional performance and counsellor – patient relationship. Specific approaches will be adopted to enhance counsellors self-awareness and to avoid occupational stress and burn-out syndrome. In fact, poor self-awareness could lead to under or overestimate own coping strategy with occupational stress. During the class we will explore both with frontal lesson and with exercise: empathy, burn-out symptom (how to recognise and how to prevent), self-awareness tools.

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