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News Article

Webcast courses in Medical Genetics and Next Generation Sequencing

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The European School of Genetic Medicine organised the 26th Course in Medical Genetics and the 2nd Course in Next Generation Sequencing, between the 12th and 20th May 2013. Both courses were webcast live from the Bologna University Residential Centre, Bertinoro, Italy. Participants in Malta attended these courses at the University's Medical School.

The course in Medical Genetics covered various aspects of this rapidly developing field of Medicine. The different methodologies used in human genome analysis, an introduction to Next Generation Sequencing (NGS), approaches to clinical and molecular genetics, complex genetic disorders, therapy and gene regulation, were covered.

The second course provided a comprehensive insight into NGS technologies, from the basics to the new world of disease gene identification by hand-held devices. It also covered insights into bioinformatics challenges, sample preservation and trans-omic studies, and new frontiers including the investigation of single cells and of the non-coding genome.

Medical Genetics or Genetic Medicine as it is increasingly being referred to nowadays, is a constantly changing clinical specialty. In the past 15 years there has been a massive increase in referrals of conditions regarded as common complex disorders such as breast and bowel cancer, and some cardiac diseases. An increase in the range of medical genetic services has benefited patients with genetic disorders and their families. "Careful clinical observation is at the heart of medical genetic practice" (Donnai xxxx). "The new technologies enabling targeted capture and massively parallel sequencing of individual genomes/exomes, have resulted in major discoveries on small clinically well characterised patients" (Donnai xxxx). Through the identification of novel genes, new developmental pathways have been discovered; many disorders with overlapping clinical features shown to be due to mutations in functionally related genes, might be responsive to treatment by similar molecules (Donnai xxxx).

Prof van Duijn delivered an interesting presentation entitled 'Complex Disease Genetics: GWAS and beyond'. Whole genome association studies using Affymetrix®/ Illumina®arrays, have revealed increased number of variants involved in complex disease. Sometimes it is difficult to predict the genetic risk for conditions such as dyslipidaemia and Alzheimer's disease. Genome tests are currently limited with respect to their predictive ability (van Duijn xxxx). "In the near future, personal genome tests would very likely be based on whole genome sequencing, but will these technological advances increase the utility of personal genome testing? The utility of testing depends on the predictive ability of the test, the likelihood of actionable test results, and the options available for the reduction of risks" (van Duijn xxxx). The identification of new variants in monogenic diseases could present a challenge due to the number of rare disease variants. The complexity of disease aetiology and disease heritability will affect the prediction of genetic risk for complex diseases. New omic technologies will aid in the discovery of biomarkers which could be used to improve predictions (van Duijn xxxx).

Next generation sequencing is a technological revolution in genomics which will have a major impact

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Figure 1

on the entire field of medicine. "All genomic variation that can be linked to disease is detectable in a single experiment!" (Veltman xxxx). In the near future it will soon be possible to sequence an entire genome at an affordable price which will lead to personalised medicine. Important characteristics of NGS platforms include: accuracy, ease of mapping / assembly sequence, sequence throughput / coverage, application, robustness, ease of interpretation and costs (Veltman xxxx). "The unbiased sequencing of the exome has become an integral part of the toolkit for genetics researchers" (Gilissen xxxx). Exome sequencing of trios (mother, father, child) are being offered to families of individuals with unknown disorders. Patients with specific disorders such as retinal dystrophy, cataract and epilepsy are being offered targeted testing using large panels of genes (Donnai xxxx). Preliminary results of diagnostic applications of NGS show that there is a much wider phenotypic spectrum associated with mutations in many genes than was previously thought. There have been concerns about the ethical aspects of NGS but as experience increases most centres are finding ways of addressing these in conjunction with patient groups (Donnai xxxx).

Handheld diagnostics on nanowires was one of the fascinating presentations during the NGS course. A novel, cheap, portable, handheld DNA sequencing device, is being developed by QuantumMDx for infectious disease applications at point of care testing, as an alternative to slow and relatively expensive capillary electrophoresis DNA sequencing (O'Halloran xxxx).

The presentation entitled 'Sample Preservation and Trans-omic Studies to Accelarate Scientific Research' delved into the use, requirements and benefits of biobanking. "Fast development of sequencing technology with decreasing cost has substantially promoted large cohort study, population health screening, and epidemiological prevention study on a genomic level" (Cheng xxxx). The 100 million US dollar China National Genebank (Shenzhen), operated by the Beijing Genomics Institute (BGI), aims at combining large bio-specimen collection and transcriptomic data production to accelerate scientific discovery and translation to clinical use (Cheng xxxx). "The Genebank has accumulated genetic variation information of more than 2000 monogenetic diseases by collecting suspected

pedigree samples countrywide and worldwide" (Cheng xxxx). It also established databases of global human genetic variation, cancer genetic variation and human inhabited microbial composition which are essential for understanding human disease and in the development of personalised medicine.

The faculty members for both webcast courses were internationally renowned experts in the field of Medical Genetics. The cutting edge lectures delivered during both courses, provided a golden opportunity for post-graduate research students studying at the University of Malta, to widen and update their knowledge on the latest developments in Medical Genetics, and also on the technologies used in analysing genetic data.

The courses were organised by Dr Isabella Borg, Director of the European School of Genetic Medicine, Malta Remote Training Centre, based at the University of Malta Medical School, with the assistance of Ms Joanna Vella from the Malta BioBank. Funding was provided by the Malta BioBank, University of Malta, as part of the training initiative.

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