Capacity Building for Rare Genomics

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Infrequent occurrence of rare diseases (RD) has led to prolonged time needed to make an accurate diagnosis. Research and development of new treatments has been insufficient due to the narrow market for affected patients. Fascinating progress and accumulation of knowledge in the field of genomics provided for improvement of diagnosis and follow-up of numerous RD. It also led to the design of molecular-targeted therapeutics and more effective treatment of RD.

Building up a centre for genomics of rare diseases comprises strengthening of several components in an institution already involved in molecular genetics and biomedical research, such as, infrastructure, human resources, local and international networking and dissemination and outreach activities.

Regarding infrastructure, facilities for Next-generation sequencing (NGS) and array-based comparative genomic hybridization (aCGH) are necessary to perform high-throughput genome-wide screening. Establishment of the bioinformatics unit is also very important. At the heart of genomics of RD is biological samples storage and database facilities. Biobanks are a critical resource in the field of genomics of RD. Additionally, strong connections with local medical institutions have to be made.

Reinforcement of human potential, especially through recruitment of experts in bioinformatics, is indispensable contribution to the implementation of new equipment and high-throughput methodologies. Trainings of researchers are continually in focus of an expert centre for genomics of RD.

Integration of the centre into European networks in order to intensify the transfer of knowledge is one of the cornerstones for its capacity building. The data and biospecimens that are distributed to all researchers, are absolute requirements in the pathway to develop modern science, diagnosis and cures for RD.

Finally, dissemination and outreach activities are needed to ensure increased visibility of the centre within local and international scientific community and stakeholders and social entities interested in exploiting the results of the centre.

The Challenge of Living with Rare Disorders: Considerations for Genetic Counseling

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Genetic counselors play an important role in providing risk assessment, education, psychosocial support and care coordination to patients and families with rare genetic disorders. Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following: Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence. Education about inheritance, testing, management, prevention, resources and research. Counseling to promote informed choices and adaptation to the risk or condition. This process is particularly salient for patients and their family members who face multiple challenges whether or not a diagnosis is found. Individuals with rare disease face isolation, delay in diagnosis and treatment, significant physical and/or intellectual disability and health care providers who are unprepared to care for individuals with rare conditions. The possibilities for improved diagnostic accuracy in rare disease with the advent of genomic sequencing calls for improved methods to support and educate patients and their families as well as the providers who care for them. Genetic Counselors are medical providers who are specially trained to provide patient-centered care that is focused on those aspects of care that relate specifically to the rare and inherited nature of genetic conditions. This talk will address novel approaches by genetic counselors to support patients and their families with rare disease.

The Genomics of Rare Diseases: Improving the Health Economics Evidence Base

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Next generation sequencing (NGS) technologies could provide both health and non-health benefits to individuals with rare diseases. These benefits may include improved disease stratification, individually tailored therapies, and the provision of prognostic or diagnostic information that improves patient understanding, allows patients and relatives to plan and make lifestyle modifications, and reduces the frequency of diagnostic odysseys. However, these technologies have had a limited impact on clinical practice to date due to a lack of evidence, particularly economic evidence. Economic evaluations of NGS technologies pose a number of methodological challenges for health economists, consequently little empirical evidence on the costs, benefits and cost-effectiveness of these technologies has been generated to date. This prevents decision-makers from making fully informed healthcare resource allocation decisions with respect to genomic technologies and may lead to the misallocation of scarce healthcare resources, reducing population health.

In this lecture, James will begin by summarising the challenges that health economists face when undertaking economic evaluations of NGS technologies for rare diseases. He will then outline how the health economic evidence base for these technologies has recently improved, presenting the results of several health economic studies of the use of NGS technologies to diagnose, manage and treat rare diseases. This evidence will include new data on the costs of these technologies, quantitative estimates of the preferences of key stakeholders for genomic technologies, and some preliminary results from the health economic analyses that are being undertaken as part of the 100,000 Genomes Project in England. James will conclude by outlining the key priorities for researchers who hope to continue to improve the health economic evidence base for NGS technologies in the coming years.