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Fighting rare diseases

A multi-million Euro initiative is bringing together researchers from across the world to develop new diagnostic tools and new treatments for people with rare diseases and to connect research data in this area on a global scale.

Rare diseases – while individually uncommon – affect one person in every 17. 80% of rare diseases have a genetic component, and they include genetic kidney diseases like nephrotic syndrome and conditions like Huntington’s disease, ataxia and muscular dystrophy.

Today, the EU has announced 38 million Euro funding for research towards new treatments and for the development of a central global rare disease hub involving 70 institutions that will allow scientists to share data from their genomics research projects. This will lead to faster diagnosis and better treatments and improve the quality of life for patients with rare diseases.

The revolution in DNA sequencing, which means an entire human genome can now be sequenced within days and for less than 10,000 Euro, has brought the hope of personalized treatments for many of these diseases a step closer.

Professor Hanns Lochmüller of Newcastle University, UK, who is leading the new rare disease hub, said: “Being able to sequence a person’s entire genetic code is an important advance, particularly for people living with the many rare genetic disorders, but it has also shown us that sequencing is only the first part of the story. It doesn’t replace clinical expertise – in fact, being able to combine genetic data with clinical data is more important than ever.”

Dr Ségolène Aymé, Emeritus Director of Research at INSERM, the French Institute of Health and Medical Research, added: “Sequencing produces a vast amount of information, but in most cases it will find hundreds of genetic changes in each person. We now need to collate the data internationally to discover which change – or combination of changes – actually causes the disease.”

The International Rare Diseases Research Consortium (IRDiRC), under which these new grants have been awarded, aims to accelerate research into rare diseases. Professor Paul Lasko of McGill University in Montréal, Canada, Chair-Elect of the IRDiRC Executive Committee, explained: “IRDiRC’s goal is to reach 200 new rare disease therapies, and diagnoses for all rare diseases, by the year 2020. To this end, it is today launching three major projects which will combine international genetic data with clinical information and data on biomaterials to help interpret the vast amounts of data the genome yields. This will aid scientists in the search for genetic causes of diseases and help identify new ways to create targeted therapies”.

Professor Lochmüller said: “Already we have drugs being tested in clinical trials which can, in effect, patch up the faults in the genes for some rare diseases such as Duchenne muscular dystrophy. Drugs like this are at the vanguard of a new generation of therapies that change a person’s genes rather than just treating their symptoms, and they have the potential to make a real difference to the

quality of life of people with the condition. By sharing data and clinical expertise in this structured way across an international network, we hope to discover similar life-changing drugs for other rare diseases.”

A rare disease is defined by the European Union as one that affects fewer than five people in every 10,000 of the general population. There are between 6,000 and 8,000 known rare diseases, and approximately 30 million people across Europe are affected by a rare disease.

The four IRDiRC projects being launched in Barcelona today have received nearly 40 million EUR of funding for cutting-edge research and collaboration over the next six years. Funding focuses on international collaborations:

- Identifying the genetic and epigenetic causes of rare kidney disorders – EUREnOmics led by Heidelberg University Medical Centre, Germany
- Addressing rare neurodegenerative and neuromuscular disorders using next generation whole-exome sequencing – Neuromics led by the University of Tübingen, Germany
- Developing a global infrastructure to share the research of rare disease projects – RD-Connect led by Newcastle University, UK
- Supporting international rare disease collaboration through IRDiRC – SUPPORT-IRDiRC led by INSERM, France

The projects funded by the EU under the IRDiRC:

EUREnOmics (www.eurenomics.eu) focuses on rare kidney disorders and aims to identify novel genetic and epigenetic causes and modifiers of disease and their molecular pathways, develop innovative technologies allowing rapid diagnostic testing, discover and validate biomarkers of disease activity, prognosis and treatment responses, and develop in vitro and in vivo disease models to apply high-throughput drug candidate screening.

Coordinator: Professor Franz Schaefer MD, Heidelberg University Medical Center, Germany

Neuromics (www.rd-neuromics.eu) addresses rare neurodegenerative and neuromuscular disorders and will use next generation whole-exome sequencing (WES) to increase the number of known gene loci, increase patient cohorts through large scale genotyping by gene panel enrichment and next generation sequencing, develop biomarkers for clinical application with a strong emphasis on presymptomatic utility and cohort stratification, identify disease modifiers and develop targeted therapies using latest generation genetic approaches. Trend-setting for future EU projects, top scientists from the US, Canada, and Australia participate in the network.

Coordinator: Professor Olaf Riess MD, Institute of Medical Genetics and Applied Genomics, University of Tübingen, Germany. Co-coordinators Professor Brunhilde Wirth, Institute of Human Genetics, Cologne, Germany, and Professor Gert-Jan von Ommen, Institute of Human Genetics, Leiden, The Netherlands

RD-Connect (www.rd-connect.eu) will develop a global infrastructure for sharing the research outputs of these and other rare disease projects, enabling scientists and clinicians worldwide to access a single centralized repository for omics data, phenotypic and biomaterial information. Every IRDiRC research project will be entitled to share its own data and access related data from other projects under policies agreed at a global level.

Coordinator: Professor Hanns Lochmüller MD, Institute of Genetic Medicine, Newcastle University, UK

Support-IRDIRC (www.irdirc.org) provides the organisational support for the implementation of the International Rare Diseases Research Consortium in close collaboration with the European Commission, the NIH and research funding agencies from participating countries, as well as with relevant research projects supporting IRDiRC objectives.

Coordinator: Dr Ségolène Aymé, Emeritus Director of Research, INSERM, France

The International Rare Diseases Research Consortium (IRDIRC) aims to foster international collaboration in rare disease research, a highly challenging area of medical research that has the potential to benefit tremendously from the recent advances in genomics, proteomics and other omics technologies. IRDiRC has set itself the bold aims of delivering 200 new rare disease therapies and diagnosis for all rare diseases by the year 2020. Spearheaded by the European Union, the United States National Institutes of Health and the Canadian Institutes of Health Research, the IRDiRC now numbers 29 member funding institutions across the world. This global collaboration between major research funders will ensure greater harmonization of rare disease research activities and lay the foundations for future networking that is essential to accelerate progress in the field.

Rare diseases include:

Muscular Dystrophy

Spinal Muscular Atrophy

Cystic Fibrosis

Spinocerebellar Ataxia

Huntington's Disease

Osteogenesis imperfecta

Ehlers-Danlos Syndrome

Epidermolysis Bullosa

Rett Syndrome

Marfan Syndrome

Hemochromatosis

Thalassaemia

Mucopolysaccharidosis

Polycystic Kidney Disease

Nephrotic syndrome

Haemophilia

Progeria

Amyotrophic Lateral Sclerosis