IARC 2015 Lay Summary Session 1: New Genes & Developments in the Diagnosis of the ataxias

Session 1 mainly focused on the identification of new ataxia associated genes and genetic disease mechanisms by making use of advanced sequencing technologies.

Firstly, Prof Andrea Nemeth (University of Oxford, UK) reviewed the current landscape in the field of genes and diagnosis for the ataxias. She explained how the genetic cause of disease has always been a big obstacle for clinicians due to the high variability of the ataxias as well as technical limitations. She went on to describe how less than ten years ago a revolutionary technique transformed traditional genetic research: next generation sequencing, particularly exome sequencing. The latter consists of sequencing certain fractions of the genes: the DNA that encodes for proteins, also known as exons (a small amount of DNA making up just 1% of the whole human genome), that seems to have the most important role in the onset of human genetic disorders. Now that next generation sequencing has entered into the field of diagnostics, clinical practice has been able to progress rapidly. The use of advanced sequencing technologies was echoed at the conference as several new genes associated with the ataxias were discovered using this technology.

Dr Matthis Synofzik (Hertie-Institute for Clinical Brain Research, Germany) presented his important research demonstrating his use of new sequencing technologies in finding four novel genes and genetic mechanisms associated with early onset ataxia (EOA). The research, which involved the collaboration

of multiple institutions, reiterates how important partnerships are in identifying genes within a short time frame.

Prof Patrick Chinnery (Newcastle University, UK), also using an exome sequencing approach, identified a mutation in the SPG7 gene (already known as the causative gene of Spastic Paraplegia Type 7), that seems likely to explain approximately 18% of patients with unexplained ataxia. This was well received following Prof Marios Hadjivassilious talk on his research (Royal Hallamshire Hospital, Sheffield, UK), which found that 22% of patients with progressive ataxia at his specialist centre had no clear cause to explain their diagnosis.

With new genetic sequencing techniques comes a vast amount of data with unknown significance, as it takes a massive amount of time to analyse all of the data produced. To try and make use of this data and cut down on time, Dr Schule (Miller School of Medicine, University of Miami, USA) involved in the development of the Genomes Management Application (GEM.app), pointed out the importance of exome data sharing to reduce interpretation time and resolve the puzzle of rare diseases. The aim of GEM.app is to create a collaborative network between researchers that are performing next generation sequencing and to share all data in a common database, in order to facilitate the identification of new disease causing genes. This promising approach will help us to determine the clinical signs and the course of the disease, by identify more families affected by similar disorders and in unravelling the mechanism of the diseases.

In conclusion, it was very impressive to see how many people are working and collaborating in the same field (in particular on Spinocerebellar ataxia type 3 and Friedreich's ataxia) with different approaches, in order to develop novel therapies that might become effective against the ataxias.

By Dr Cecilia Mancini, University of Torino & Dr Jana Schmidt, University of Tübingen