Highlights from the International Congress for Ataxia Research and Ataxia Global Initiative

Between November 1st – 4th 2022, Ataxia UK, the National Ataxia Foundation (NAF), and the Friedreich's Ataxia Research Alliance (FARA) co-hosted the International Congress for Ataxia Research (ICAR) in Dallas, Texas, USA. The conference was a hugely successful event. Over 450 people attended from 19 different countries, making this one of the largest gatherings of ataxia researchers to date. This included researchers, pharmaceutical companies, and patient group representatives. The conference programme included sessions on many different aspects of ataxia, including gene discovery, disease mechanisms, biomarkers, and emerging therapeutics. Here are some of the highlights relevant to the TREAT-ARCA project.

COQ8A-ataxia presentation

Lead researcher on the TREAT-ARCA project, Dr Hélène Puccio from Université de Lyon, gave a presentation on COQ8A-ataxia. COQ8A-ataxia is caused by mutations to the COQ8A gene. Dr Puccio presented data on a mouse model which does not have the COQ8A gene, and therefore shows some features similar to COQ8A-ataxia. This mouse model could be used to test potential treatments for COQ8A-ataxia, including in the TREAT-ARCA project (for a summary of the aims of the TREAT-ARCA project, click here).

ARSACS presentation

Researchers from the groups of Dr Francesca Maltecca and Dr Bernard Brais, both researchers on the TREAT-ARCA project, gave a presentation on the underlying cause of ARSACS. ARSACS is caused by mutations to the SACS gene, which results in a loss-of-function of the SACS protein. However, the mechanism by which this causes the neurodegeneration seen in ARSACS is not clear. They looked at brain cells taken from mice that don't have the SACS gene in order to better understand how mutations in this gene cause neurodegeneration. They also treated these mice with a repurposed drug called Ceftriaxone, which is neuroprotective. Ceftriaxone treatment significantly improved motor performance in the mice. Testing repurposed drugs in mouse models of ARSACS is an aim of the TREAT-ARCA project, and the results of this study are encouraging for the future of ARSACS treatment.

Dr Maltecca's group also gave a talk on the effect of different mutations to the SACS gene that cause ARSACS, of which more than 200 have been identified worldwide. There is variability in the symptoms of ARSACS, which has not been explained by the different mutations. By analysing skin cells from people with ARSACS, they saw that the sacsin protein (produced from the SACS gene) was almost absent in people with ARSACS, regardless of the mutation they have in their SACS gene. They showed that when mutations are present in the SACS gene, the sacsin protein is degraded. As well as providing a possible explanation for the lack of correlation

between genetic mutation and symptoms, these results could also provide a fast way to diagnose ARSACS – by looking at the level of sacsin in blood samples.

Conference feedback

92% of ICAR attendees thought the conference was extremely useful or very useful for advancing their work, and 96% made new contacts whilst attending. International conferences are a fantastic opportunity for ataxia researchers to present their work, connect with other researchers, and are important for inspiring and progressing further ataxia research. Click here to read scientific summaries of all talks and posters in the abstract book.

Ataxia Global Initiative

The Ataxia Global Initiative (AGI) Conference was held immediately after ICAR 2022, On November 4th – 5th. The AGI is a worldwide research platform, with the goal of facilitating development of therapies for the ataxias. It involves researchers, pharmaceutical representatives and patient groups. The AGI conference included presentations on a range of ataxias, focusing on clinical trial-readiness.

Dr Matthis Synofzik, one of the researchers on the TREAT-ARCA project, is co-lead of the AGI. He gave a presentation on the ARCA cohort. ARCA stands for Autosomal Recessive Cerebellar Ataxias, and includes ataxias such as ARSACS and COQ8A-ataxia – the focus of the TREAT-ARCA project. Dr Synofzik described a trial-readiness study, in which 1006 people with ARCA/early-onset ataxia have been followed in the clinic over multiple visits, in more than 15 different countries. This is an extremely powerful resource for research, and has enabled researchers to capture the progression of ataxias such as ARSACS and COQ8A-ataxia. Due to the very rare nature of these ataxias, without the ARCA cohort combining the data over multiple countries, capturing the progression of these ataxias wouldn't be possible. This study improves the trial-readiness of researchers when treatments for these ataxias, such as those studied in the TREAT-ARCA project, become available for clinical trials.

DHAG Deutsche Heredo-Ataxie-Gesellschaft

euro ataxia

ATAXIA

Ataxia Charlevoix-Saguenay Foundation

Research that heals