



Society News

The DGNG held its 18th annual meeting, which was jointly organized by Prof. Georg Auburger, Frankfurt, and Prof. Ulrich Müller, Giessen, from Oct. 25 to 27 2012 in Frankfurt. Main topics of the meeting included psychiatric disorders, neuromuscular diseases, Parkinson's disease and dystonias, but also more basic science-oriented sessions on the role of RNA in normal and abnormal brain function, RNA-translation and protein degradation. A particular highlight was undoubtedly the keynote lecture, held this year by Prof. Daniel Geschwind, Depts. Of Neurology and Psychiatry, University of California, Los Angeles, on "Autism: Integrating genetic and neurobiological understanding".

This program again showed many exciting advances in neurogenetics and provided a broad, yet detailed image of the current state of the art.

As the timing of the Annual Meeting of the DGNG in the fall was felt to be increasingly problematic for organizers and attendants given the many competing meetings and conferences at this time of the year, the board of the society decided to change the schedule, and move the meeting to the spring, effective of **2015**, when the 19th Annual Workshop of the German Society for Neurogenetics will be held on **March 25 to 27, in Bonn**. As every year, there will be invited lectures and poster-presentations, the best three

posters can expect a poster award. A preliminary program will be available soon. At this meeting in 2015, the DGNG will again present an „**Junior Research Award**“ of 2.000 Euro to a young investigator (below 35 years) for a paper published or submitted within one year of the **deadline** of submission which will be **Oct 31, 2014**.

Submissions should be sent as a single pdf-file including a cover letter, the full paper and a brief CV of the applicant to the secretariat of DGNG, Isolde.Marterer@med.uni-tuebingen.de.

In **2014**, the Society will support and participate in the organization of the Annual Meeting of the Society for Genetics (Gesellschaft für Genetik, GfG), which will be held on **Oct. 1 – 3 in Luxemburg**. Organized by Prof. Rudi Balling, this year's meeting will be focusing on Neurogenetics. Further information can be found on the homepage of the GfG (<http://www.gfgenetik.de/tagungen/>) and on <http://neuroconference2014.uni.lu/>

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NeurOmics: -omics research for diagnosis and therapy in rare neuromuscular and neurodegenerative diseases – an EU funded FP7 project

This article will give a brief introduction into the structure of a large EU funded research project on rare neuromuscular and neurodegenerative diseases, which was initiated in January 2013 for a five year funding period. Moreover, first results after 18 months of collaborative work illustrate the progress and present highlights of research activities so far.

NeurOmics is an EU-funded translational research project which has the primary aim of improving the understanding and therapy of neuromuscular and neurodegenerative diseases. This means that NeurOmics research will contribute significantly to the International Rare Diseases Research Consortium's (IRDiRC) objectives of a genetic diagnosis for most rare disease patients and the development of 200 new therapies by 2020. The project focusses on 10 rare, genetic neuromuscular and neurodegenerative disease groups: Huntington's disease (HD), fronto-temporal lobar degeneration (FTLD), hereditary spastic paraplegia (HSP), ataxia, spinal muscular atrophy (SMA) / lower motor neuron disease (LMND),

hereditary motor neuropathy (HMN), congenital myasthenic syndrome (CSM), congenital muscular dystrophies (CMD) and myopathies, muscular dystrophies (MD) and muscular channelopathies. All disease entities had already demonstrated fruitful European collaborative activities in the past. Especially managing expertise for rare disorders (treat NMD, in Newcastle, UK; ataxia study group, in Tübingen, GER) merged their expertise, in order to further strengthen visibility, communication, and research integration (further information can be found online: www.rd-neuromics.eu).

Consequently and in accordance with the IRDiRC goals, the main objective is to **establish new therapies** for patients with these rare diseases. In order to take first steps in this process, deep **phenotyping** in several patient cohorts will provide quantitative measures for disease progression, which will serve as outcome markers in therapeutic trials. Since clinical observations are only one potential outcome measure, a second workpackage deals with the identification of **biomarkers** in clinical samples of these patients making use of large transcriptomic and proteomic facilities. Moreover, a third research group is dedicated to **identifying new disease genes** in as yet unresolved families. Gene panel sequencing as a diagnostic tool and whole exome sequencing in 1.100 individuals as a research tool are the core activity of

this workpackage. Since data integration and interpretation has become a major challenge in OMICs research, a second large EU-funded consortium – **RD-Connect** – coordinated in Newcastle, UK by Hans Lochmüller (www.rd-connect.eu) has started building up analysis pipelines and dedicated databases in parallel. This infrastructure is indispensable in order to guarantee the public availability of datasets that inform further research in the disease entities presently studied and beyond.

18 months into the project, clinical structures are established, and sample collection for the biomarker studies are ongoing. The fastest progress was achieved in the gene discovery projects, since most of the patient DNAs were immediately at hands. Roughly 600 of a total of 1.100 whole exome analyses have already been accomplished and have led to the identification of 33 new disease genes.

Whilst single gene publications are still under review, a collaborative landmark publication has recently been published in Science (Novarino et al., 2014). As a proof of concept, a global research team with strong contribution of NeurOmics partners pooled exome sequencing data and network analysis in order to identify and validate 18 putative HSP genes. This approach will not identify novel disease mechanisms as it is using established disease-associated pathways to identify additional mutated genes in these

pathways. Nonetheless, through this work, common models of these affected pathways immediately become much more robust and inform about redundancy and vulnerability in the physiological maintenance of cellular transports, nucleotide metabolism, and synapse and axon development in hereditary spastic paraplegia.

In hereditary spastic paraplegia, especially disturbances in lipid metabolism are a primary target for preclinical and clinical intervention in NeurOmics.

Since the network has established and is going to establish many collaborative partnerships that open up central resources for non-members, clinicians and researchers interested to contribute to the projects objectives are invited to contact the project management.

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