## WORKSHOP ON THE GENETICS OF NEUROCOGNITIVE DISORERS

# March 7, 2014 --- 12pm-2pm Re(act) 2014 Basel

Many disorders affecting higher brain functions show a remarkable overlap both at a clinical as well as a pathophysiological level. Conversely, the identical disorder may show a high variability between different patients. Since recently, progress in genetics is contributing significantly to our understanding of what causes variation in neurocognitive disease. This workshop will highlight some of the insights gained on the genetics and pathophysiology of selected neurodevelopmental and neurocognitive disorders. The event is initiated and organized by NPSuisse, the Swiss Association for Niemann-Pick Diseases.

### PROGRAM

- 12:00-12:10 Snack (Sandwiches)
- 12:10-12:15 Christoph Poincilit (NPSuisse) / Heiko Runz Introduction
- 12:15-12:40 Nicolas Charlet-Berguerand (IGBMC Strasbourg) RNA and protein gain of function in Fragile X Tremor Ataxia Syndrome (FXTAS)
- 12:40-13:05 **Sven Cichon** (University of Basel) **Common and rare genetic risk factors in neuropsychiatric disorders**
- 13.05-13:25 Michel Koenig (University of Montpellier) Ataxia by partial loss of function: a common theme
- 13:25-13:50 Heiko Runz (Univ. of Heidelberg and Massachusetts General Hospital) Genetic and phenotypic complexities in the rare Mendelian disorder Niemann-Pick type C (NP-C)

13:50-14:00 Coffee

**NPSUISSE** (<u>www.npsuisse.ch</u>) is the Association of Niemann-Pick disease (NPD) families in Switzerland, is managed by parents, relatives and friends of patients with NPD types A,B or C. NPSuisse aims to inform and support researchers interested in NPD and similar disorders. The association has initiated the Loire Valley Meeting on NP-C as a unique forum for therapy-related NP-C research and participates in the International Niemann Pick Disease Alliance (INPDA).

**Niemann-Pick type C disease (NP-C)** is a rare, progressive lysosomal lipid storage disorder affecting the brain and visceral organs. Thanks to fundamental research, knowledge on this disease has improved considerably during the recent years, but development of curative therapies remain a challenge (<u>www.inpda.org</u>).

Actelion Pharmaceuticals Inc. (Basel) is acknowledged for kindly supporting this workshop.

## SPEAKERS

## Dr. Nicolas Charlet-Berguerand, (IGBMC Strasbourg, France)



Since 2007, Dr. Charlet-Berguerand heads an independent research group at IGBMC Strasbourg. Followig a PhD at University of Paris on RNA metabolism in cancer, his current research adresses two distinct areas: First, studying the biogenesis and roles of non-coding RNAs and RNA binding proteins and their implications in human genetic diseases. And second, how expanded non-coding RNA repeats cause RNA gain-of-function diseases such as Fragile X-associated tremor/ataxia syndrome (FXTAS), myotonic dystrophies or spinocerebellar ataxias. Dr. Charlet-Bergerand's goal is to elucidate the molecular causes of these diseases and to identify drugs able to restore a normal function in patient models.

### Dr. Sven Cichon, (University of Basel, Switzerland)



In 2013, Dr. Cichon was appointed Director of the Division of Medical Genetics at Basel University. After a PhD and postdoc on CNS-receptor/transporter genes and their impact on neuropsychiatric disorders at the Institute of Human Genetics in Bonn. Dr. Cichon conducted research at Millennium Pharmaceuticals Inc. (Cambridge, USA) and University of Antwerp (Belgium). He returned to Bonn in 2004 as Head of Molecular Genetics at the Life&Brain centre of excellence in translational biomedicine and a group leader for Genomic Imaging at Research Center Juelich and University of Bonn. His ongoing research aims to identify genetic factors influencing complex neuropsychiatric disorders as well as structural and functional variability of the human brain.

## Dr. Michel Koenig, (University of Montpellier, France)



Dr. Koenig is a newly-appointed group leader at the Institute of Human Genetics at University of Montpellier. He received his MD/PhD at University of Strasbourg, followed by a postdoc in the Department of Pediatrics and Genetics at Harvard Medical School (Boston, USA). Until 2013, Dr. Koenig was an independent Investigator at IGBMC and Professor of Human Genetics at University of Strasbourg. Since 1990, Dr. Koenig has been dedicated to the unraveling of the autosomal recessive ataxias and has made break-through discoveries in this field.

### Dr. Heiko Runz, (University of Heidelberg, DE/Massachusetts General Hospital, USA)



Dr. Runz is a group leader at the Institute of Human Genetics, University of Heidelberg and the MMPU, a translational research unit between Heidelberg University and the European Molecular Biological Laboratories (EMBL). Since 2012, he also conducts research at the Mass. General Hospital in Boston, USA. Following a postdoc at EMBL and clinical training as a specialist in medical genetics, Dr. Runz' research centers around the mechanisms that cause variability in human genetic disease and how such variability may lead to novel therapies. His research bridges the genetics of common complex conditions with that of rare neurocognitive syndromes such as Niemann-Pick type C disease (NP-C).