

## Skou Professor Seminar on ADHD and autism – from genomics to biology and precision medicine

Tuesday 8 October 2019 at 10.00 – 11.10



### Biological Psychiatry – moving from limitations to opportunities

#### Professor Barbara Franke

Department of Human Genetics and Psychiatry,  
Donders Institute for Brain, Cognition and Behaviour,  
Radboud University Medical Center, Nijmegen, The Netherlands.

**Champion: Ditte Demontis**

### Abstract

Through technological innovations and international collaboration, we are finally succeeding in identifying genes that affect risk for psychiatric disorders. Our increasing possibilities also help us understand the genetic architecture of brain disorders. Having found ways to eliminate bottlenecks in gene-finding, our field is running into new ones where we are trying to understand, how the identified risk factors contribute to disease risk. Mapping of the biological pathways from gene to a psychiatric disorder requires interdisciplinary ‘Convergence Neuroscience’ approaches, most efficiently achieved through collaboration. In my lab, we have developed a pipeline including bioinformatics, cell-based and small animal (*Drosophila*) models, as well as neuroimaging genetics studies, which enables us to probe the underlying processes leading from gene to neurodevelopmental psychiatric disorders. A thorough understanding of the biological basis of psychiatric disorders seems indispensable, where we aim at improving the life quality of patients with psychiatric disorders through better diagnostic services and improved treatment options for the future.

### Biosketch

Barbara Franke holds the Chair of Molecular Psychiatry at Radboud University in Nijmegen, The Netherlands. She is an elected member of the Royal Netherlands Academy of Arts and Sciences, the Royal Holland Society of Sciences and Humanities, and of Academia Europaea.

Her research is focused on understanding the genetic contribution to neurodevelopmental psychiatric disorders, especially ADHD and its comorbidities. Beyond gene-finding, she uses complementary approaches (bioinformatics, i-neurons, small animal models, neuroimaging genetics) to map biological pathways from gene to disease.

She coordinated the EU-FP7 Aggressotype consortium on ADHD and aggression and leads work packages in two EU-funded consortia (CoCA on ADHD comorbidity, Eat2beNICE on impulsive behaviour and nutrition). She founded the International Multicentre persistent ADHD Collaboration (IMpACT) and the ECNP Network ‘ADHD across the Lifespan’, is a co-founder of ENIGMA, and chair of the Psychiatric Genomics Consortium’s ADHD Working Group. In 2018, she was elected to the Board of Directors of the International Society for Psychiatric Genetics. Barbara Franke has published over 400 peer reviewed publications and is on Clavirate’s list of the 1% most highly cited researchers worldwide.



## From gene discovery to precision medicine in autism

### Professor Joseph Buxbaum

Department of Psychiatry, Icahn School of Medicine,  
Mount Sinai, New York, USA.

### Champion: Anders Børglum

### Abstract

Autism and associated disorders have a strong genetic component, with rare and spontaneous mutations having a major impact on individual risk. With the advent of inexpensive, high-throughput sequencing to analyze virtually all of our 20,000 genes, several hundred genes have been identified for autism and associated disorders. When mutated, these genes confer high risk for autism and provide an entrée into understanding the biology of autism. Moreover, they present an opportunity to consider precision medicine in autism, with the ultimate hope of developing therapies that are optimized for individuals, based on their biological and genetic profile. In this talk, the role of rare and spontaneous mutations in autism will be described. In addition, a path to precision medicine in carriers of rare mutations will be outlined.

### Biosketch

Dr. Buxbaum is a Professor and Vice-Chair for Research, Department of Psychiatry at the Icahn School of Medicine at Mount Sinai. He is the Director of the Seaver Autism Center, with 15 faculty using preclinical and clinical approaches to understand and treat autism. Dr. Buxbaum is the co-founder and co-lead investigator of the Autism Sequencing Consortium, an international collaboration to discover genes for autism, working with tens of thousands of individual DNA samples. His laboratory has identified common and rare genetic variants that underlie autism spectrum disorders and has developed cell and animal model systems for functional follow up. Dr. Buxbaum has published over 300 articles and is the co-editor-in-chief (with Simon Baron-Cohen) of the journal *Molecular Autism*. In 2015 he was elected to the National Academy of Medicine and in 2019 was elected as a Fellow of the International Society for Autism Research

### Programme

10.00 – 10.05 Welcome by Anders Børglum

10.05 – 10.35 Lecture by Barbara Franke

10.40 – 11.10 Lecture by Joseph Buxbaum

### Venue:

Merete Barker Auditory, The Lakeside Theatres, Aarhus University, Bartholins Allé 3, 8000 Aarhus C.

All are welcome!