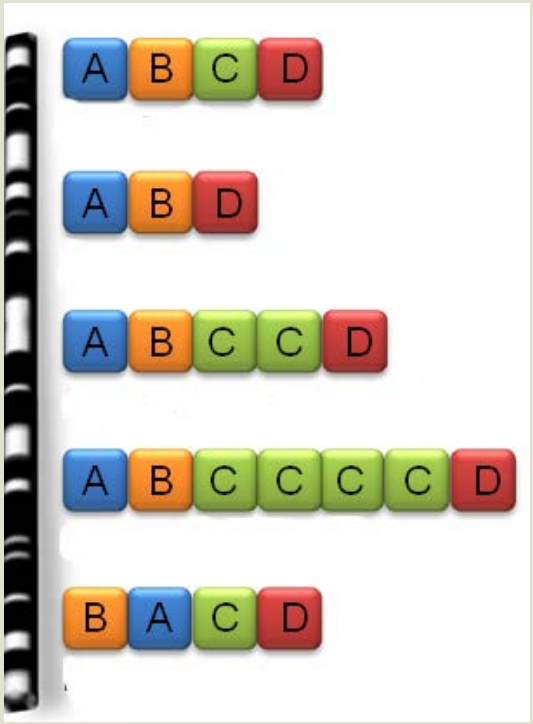

The Jacobs Center's Series on Social Science and Genetics

Dr. Katrin Männik

Center for Integrative Genomics, University of Lausanne

RARE DNA COPY-NUMBER VARIANTS AND COMPLEX DEVELOPMENTAL PHENOTYPES IN THE GENERAL POPULATION

Although individual differences in cognition and education achievement are known to be highly heritable, studies have only now begun to identify contributing genetic factors. For example, our recent study showed that structural changes in the human genome called DNA copy-number variants (CNVs) contribute to common health and developmental problem. During the past decade, rare CNVs have been established as important contributors to the origins of rare genetic diseases, especially intellectual disability syndromes. However, their association with disorders has almost exclusively been investigated in clinically ascertained patients, leaving it unclear how these variants affect health in the general population. To investigate the health burden of rare CNVs in unselected populations, we analyzed 14,778 presumptively healthy biobank participants from four different European countries. Our results showed that about 1 individual in 118 carries a CNV causative for a complex set of developmental and health-related traits for academic success, about 1 in 40 people appears to be negatively affected by rare CNVs. We also found that the cumulative set of genes encompassed by rare deletions is significantly enriched for biological processes directly involved in neurodevelopment, behavior, learning, memory and cognition. Thus, rare CNVs might account for a substantial portion of the population variance in education attainment.

During my talk, I intend to give to the audience an overview of our studies in this field. The "genome-wide approach" will be complemented by a representative example of CNVs in the chromosome interval 16p11.2 to show how multiple layers of molecular, clinical and neuropsychological information can be used to better understand the effect of CNVs. The 16p11.2 CNVs are known genetic causes of neurodevelopmental problems and pose an excellent model for studying complex heritability of cognitive and behavioral traits, inter-individual variability, pleiotropy and functional interactions between genes.

Friday, March 3, 2017, 10:00 – 10:45 h

At the Jacobs Center for Productive Youth Development
Andreasstrasse 15, 4th floor, AND 4.19, 8050 Zürich

Individual meetings with Dr. K. Männik are available,
if interested please contact Maria Schönholzer maria.schoenholzer@jacobscenter.uzh.ch