Summary / Zusammenfassung
Autism spectrum disorders (ASDs) are a lifelong and complex neurodevelopmental disorder including early infantile Autism, Asperger’s disorder and atypical Autism. They are collectively characterized by impaired social interactions and communication skills as well as repetitive restrictive behaviors/restricted interests. The incidence of ASDs is now estimated as 1 in 100. ASDs show strong evidence for genetic and multifactorial susceptibility. Heritability estimates from family and twin studies suggest that about 80-90% of variance is attributable to genetic factors, making this disorder considered to be among the most heritable neuropsychiatric disorders. The clinical definition still relies on observation of behavior and cognitive phenotypes. Consequently, understanding the functional mechanism of the genetic alterations would be important for improved strategies for early and proper identification and to establish biological markers. There are several recent studies providing a remarkable advance in the knowledge of the genetic causes. It was proposed that sporadic ASD mainly results from spontaneous mutation with high penetrance in males, but low penetrance in females. These parents transmit the mutation in a dominant manner to their offspring, leading to high-risk families. Previous findings supported this hypothesis and showed that patients with sporadic ASD have more often de-novo copy number variations (CNVs) than affected persons from high-risk families or controls. Recent work has shown unbalanced genomic alterations that are likely pathogenetic in more than 10% of cases, demonstrating an important role for CNVs in the genetic aetiology of ASD. Since both genetic and environmental factors are currently implicated in the aetiology of ASDs, studying epigenetic mechanisms like the methylation status may provide additional novel insights into the pathogenesis of ASDs. The aim of this study is to identify CNVs and to analyze a potential link between them and the methylation variations in genes known to be involved in ASDs.

Keywords / Suchbegriffe
Autism spectrum disorder (ASD), CNV-analysis

Project Leadership and Contacts / Projektleitung und Kontakte
Prof. Dr. Susanne Walitza (Project Leader) susanne.walitza@kjpdzh.ch
PD Dr. Edna Grünblatt (Project Leader) edna.grünblatt@kjpdzh.ch
Dr. Ronnie Gundelfinger (Project Leader) ronnie.gundelfinger@kjpdzh.ch
Dr. Anna Werling (Project Leader) anna.werling@kjpdzh.ch

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In Collaboration with / In Zusammenarbeit mit
Prof. Dr. Anita Rauch
University Zurich
anita.rauch@medgen.uzh.ch
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