**Intellectual disability and developmental disorders**

The intellectual disability research group focuses on the identification of new candidate genes for intellectual disabilities and related disorders. Even though the majority of cases of intellectual disability is believed to be of genetic cause, a diagnosis can only be made in about 40-60% of cases. Our research group aims at the identification and characterization of novel candidate genes using Next Generation Sequencing (NGS) Technologies.

For sporadic cases we sequence parent-child-trios and filter for de novo variants. For familiar cases we sequence one individual, filter for variants in concordance with the inheritance pattern and check for segregation. The most promising new candidate genes are further characterized with functional analysis.

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**Filtering strategies for Whole Exome Sequencing (WES) data**

- **WES of both parents and affected individual (Trio Analysis)**
- **WES of one affected individual**

**Inheritance Cases solved with NGS**

- **Solved 25%**
- **Under investigation 14%**
- **Good candidate 15%**
- **Under investigation 11%**
- **Autosomal recessive (ARID) in Jordan**

In a bilateral project between Germany and Jordan, we examined 28 consanguineous Jordanian families with intellectual disability (ID) or related neurodegenerative disorders. Whole exome sequencing revealed deleterious mutations in 10 families and good candidate variants in further 2 families. Interestingly, 25% of the identified mutations were de novo variants.