Science

Epilepsy is one of the most common neurological disorders worldwide and is characterized by recurrent and spontaneous seizures. The clinical manifestation of epilepsy differs greatly in onset age, seizure type(s) and frequency, drug-response, and additional features affecting the cognitive, physical, psychological, and social wellbeing of the patient. Despite the existence of different symptomatic treatments, multiple therapy is often needed to gain seizure control. Side-effects are also frequent, and 30% of the patients remain refractory. New insights in the precise underlying pathomechanism of brain hyperexcitability are required to elucidate the true basis of epileptogenesis and locate potential therapeutic targets.

In 70% of the epilepsy patients genetic factors are thought to play a role. Gene-identification studies using traditional genetic approaches have led to the discovery of a dozen epilepsy causing genes and many more associated loci. The last decade, NGS technologies have introduced a less biased and wider applicable gene-identification approach. Despite the increasing amount of disease associated gene defects and partially because of unexpected genotype-phenotype relationships, many patients remain without a molecular diagnosis. Gene-identification studies remain of importance to establish a framework for better diagnostic strategies. Additionally, they lay the groundwork for studying disease mechanisms and provide input for generating disease models.

As a starting point for the conducted molecular genetic studies, my research focuses on patients and families exhibiting a Mendelian inheritance pattern. By applying different NGS strategies in large scale investigations I aim to further broaden our understanding on heritable epilepsy syndromes.
Selected Publications


All publications