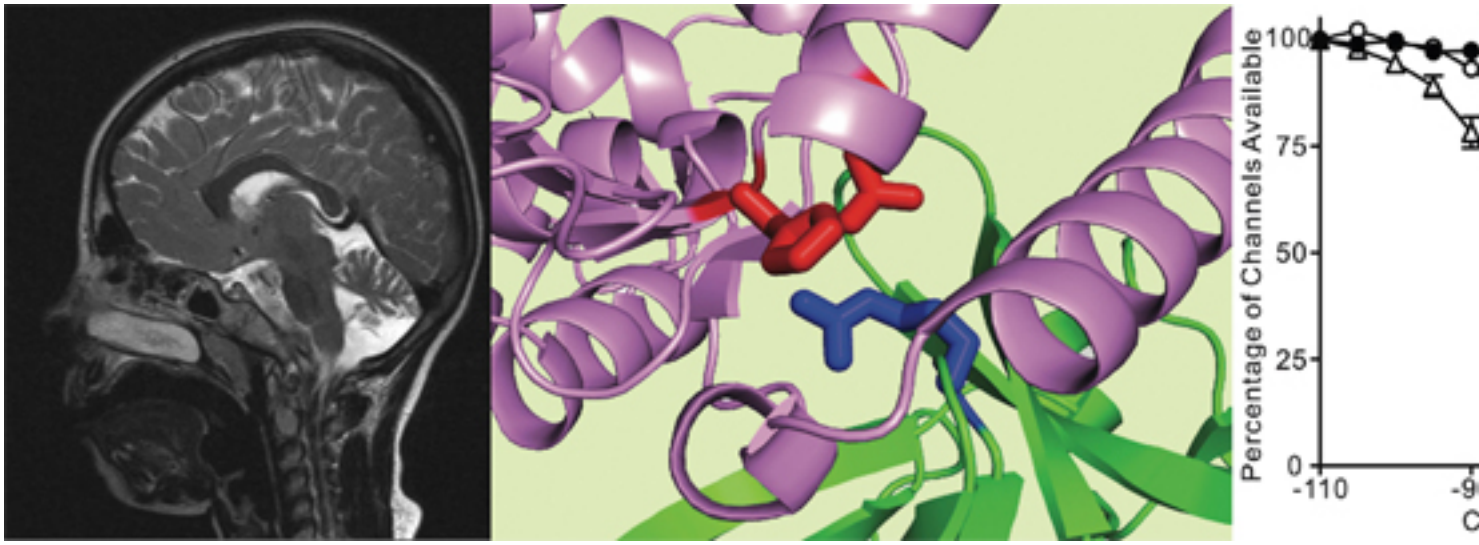

Genetic Cause of Severe Epilepsy Identified

Severe infantile onset epilepsy is a highly debilitating and generally fatal brain hyperexcitability disorder with limited responsiveness to known anti-epileptic drugs. Mitch Goldfarb's lab at Hunter College, collaborating with Gunnar Buyse and Peter de Witte's his research teams at the University of Leuven in Belgium, has characterized a single genetic mutation responsible for this disorder in two children. The mutation, in a gene called FHF1, affects the interaction of FHF1 protein with nerve cell sodium channels in a manner that enhances channel activity to drive greater excitability of nerve cells. The discovery of a protein-protein binding site underlying severe epilepsy offers a potential target for future drug design and treatment. These studies are featured in the 2016 June 7th issue of the journal Neurology.