Beate Winner, Zacharias Kohl, Carol Marchetto, Fred H. Gage, Jürgen Winkler
Laboratory of Genetics, Salk Institute of Biological Studies, La Jolla, USA
Division of Molecular Neurology, University Hospital Erlangen, Erlangen, Germany

Investigating neurons from HSP patients

With support of the Tom Wahlig Advanced Scholarship for the research into Hereditary Spastic Paraplegia we investigate patient derived neurons from patients with hereditary spastic paraplegia (SPG 4 and SPG 11). We reprogram HSP patient's fibriblasts and fibroblasts from controls into induced pluripotent stem (iPS) cells. These iPS cells, generated from human skin cells are then differentiate into human neurons. Here different differentiation protocols are used to generate cortical neurons as well as motor neurons. The aim of our research is to establish phenotypic iPS cells from patients with HSP skin samples and differentiate those into motor neurons. We will investigate axonal pathology of those human patient derived motor neurons. We will focus on genetically defined SPG4 (most frequent dominant form of HSP) and SPG11 (most frequent recessive form of HSP) patients. We will investigate cellular and molecular abnormalities that may underlie the disturbance in axonal transport and maintenance in these patient's motor neurons and test the individual potential of drug candidates and targets for future pharmacological intervention.