Description for the general public (in English):

Microcephaly is a medical condition in which the circumference of the head is smaller than normal. It has been estimated that this problem affects 0.1% of population. Abnormal head size may reflect abnormal brain development and/or brain growth arrest. Microcephaly may be caused by genetic factors that interfere with the growth of cerebral cortex during early months of fetal development. It may results from exposure to environmental factors including intrauterine viral infections, perinatal asphyxia, or teratogenic agents. The recent developments in the field of human genetics and especially introduction of whole exome sequencing accelerated identification of novel genes associated with human hereditary disorders. We propose to collect a cohort of patient with microcephaly and interrogate their genomes with whole exome sequencing. We expect to identify mutations in known microcephaly-associated genes in some patients however our goal is to find novel candidate genes associated with abnormal brain development. We propose to investigate these candidate genes further and model detected mutations using animal models including fruit fly and zebrafish. We believe the results of this study will contribute to our understanding of mechanism controlling brain development and development of novel therapies.