Identification of nuclear genes of mitochondrial diseases with neurological involvement

Mitochondrial disorders represent a heterogeneous group of genetic diseases that should be considered as a large and heterogeneous group of different orphan diseases. The aim of our project is to identify novel genes of mitochondrial disorders with neurological involvement. We have selected a group of 7 patients with syndromic or non-syndromic neurological involvement of mitochondrial origin and enzymatic respiratory chain defect. The exome sequencing will be performed only for the probands as we observed in our previous exome projects that the comparison of exome data of patients and parents is not essential as we use a filter that select genes encoding mitochondrial proteins. The functional validation of the mutations will be done by various approaches depending on the function of the gene mutations we will identify. Our expertise in the field of mitochondrial disorders has allowed us to already set up several techniques for studying various mitochondrial functions such as protein synthesis, mitochondrial RNA maturation, respiratory chain assembly that could be used for functional validation of the mutations.