

My name is Ovidiu-Lucian Bajenaru, I am PhD student in Neurology at the University of Medicine and Pharmacy "Carol Davila" Bucharest, under the scientific coordination of Professor Bogdan Ovidiu Popescu MD, PhD, Head of the Department of Neurology in the Colentina Hospital, Bucharest.

The title of my PhD thesis is: "Analysis of correlations between the clinical profile and the genomic profile in patients with idiopathic dystonia with focal onset". The objectives of my thesis are:

1. Identification of possible new variants/genetic mutations for idiopathic dystonia with focal onset;
2. Analysis on the relationship between these potential new findings and the type of dystonia, demographic features, mood and neurocognitive disturbances and other associated neurological manifestations and the quality of life;
3. Family analysis of genetic linkage.

My work is to examine and determine the clinical profile of the patients included in the study according to the inclusion/exclusion criteria, both by direct clinical examination and evaluation by dystonia-specific clinical validated scales. From these patients we collect samples from the oral mucosa to be used for genetic analysis, to find potential genetic modifications after excluding those patients who have known genetic mutations (using a specific genetic panel for known genes); for these selected patients with not yet known genetic modifications, we apply the technique of WES (Whole Exome Sequencing) followed by confirmation with Sanger method for eventual positive results.

Based on these clinical and genetic results we create a data base for the bioinformatic analysis in order to confirm the specific outcomes proposed for this thesis.