My name is Iulia Popescu-Olaru and I am a neurologist. Since I started my training in neurology, I got interested in patients with movement disorders. Currently, I am a PhD student with my research project focusing in dystonia, a group of relatively rare diseases generating abnormal movements or postures therefore having a highly debilitating potential and a significant socio-economic impact.
The title of my PhD thesis is: ***Management of focal idiopathic dystonia – correlating therapeutic response with genetic, epidemiological profile and comorbidities***, following the next **objectives**:
1. Describing the potential correlations between therapeutic response and genetic, epidemiological profile and comorbidities in a group of adult-onset idiopathic isolated focal dystonia patients
2. Identifying genetic profiles in order to replicate previously published results or to uncover gene/variants causing-disease
3. Validating UDRS (Unified Dystonia Rating Scale) in Romanian language

In order to attain these objectives, I will include and examine peoople with adult-onset isolated focal dystonia between November 2017 – December 2019 or until 120 eligible subjects are enrolled. Each subject will be well characterized from clinical point of view using the five descriptors recommended by the most recent classification consensus: age at onset, body distribution (focal, segmental, multifocal, generalized, and hemidystonia), temporal pattern (persistent, action specific, with diurnal fluctuations, and paroxysmal, each of these being either static or progressive), coexistence of other movement disorders (isolated versus combined), and coexistence other neurological or systemic manifestations. A full family history will be obtained.
I will further collect the oral swab samples required for the DNA testing of the patients. My colleagues from the Genetic Department have already designed a panel based on the genes found to be associated with adult-onset isolated focal dystonia and other movement disorders: DYT1 (TOR1A), DYT4 (TUBB4A), DYT6 (THAP1), DYT23 (CIZ1), DYT24 (ANO3), DYT 25 (GNAL), PARK2, PNKD etc. We will use this panel to screen the samples and then perform a confirmatory Sanger sequencing analysis. The genetic screening will be performed based on a targeted NGS protocol using a panel consisting of 30 genes previously found to be associated with adult-onset isolated focal dystonia. Using these clinical and genetic data we will create a data base and further perform **statistical and multivariate analysis** in order to confirm the specific proposed objectives.

Summarizing, my study comprises: ***a descriptive study*** regarding the clinical response to treatment and ***an analytic case-control study*** regarding the above mentioned genes.