

# The 17<sup>th</sup> National Congress of the Romanian Society of Neurology PROCEEDINGS

## ORAL PRESENTATION – ABSTRACTS

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### Huntington disease – case presentation and differential diagnosis algorithm

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**Introduction.** Huntington disease is a neurodegenerative disorder, easily recognized due to its specific hyperkinetic movement manifestations, but also due to the accompanying neuropsychiatric symptoms. Misdiagnosis of Huntington disease frequently arise in patients having severe psychiatric symptoms but without evident motor symptoms.

**Materials and methods.** Case presentations of 2 patients diagnosed with Huntington disease, having severe neuro-psychiatric symptoms, but with scarce motor manifestations. The clinical cases will be accompanied by the description of an algorithm that can be used for the differential diagnosis of Huntington disease. Moreover, we will present the most recent guidelines focused on behavioral symptoms of Huntington disease.

**Results.** The clinical cases describe two female patients with similar ages (47 and 63 years-old), admitted to the Neurology department for severe behavioral symptoms with onset at 40, respectively 50 years of age, followed by progressive motor manifestations, consisting in mild athetosis of the upper limbs. Both patients had significant familial history of similar manifestations with death at young age. None of the affected relatives received a proper diagnosis. Both patients were evaluated by cerebral MRI, which revealed atrophy of the head of caudate nucleus and genetic testing which confirmed the diagnosis of Huntington disease, one of the patient having 43 CAG repeats in the Huntington gene and the other 45.

**Conclusions.** Despite broad availability of genetic testing and modern cerebral imaging techniques nowadays, patients with Huntington disease are still misdiagnosed. This is probably due a low prevalence of this disease, but also due to the predominance of behavioral symptoms in some cases. The treatment of Huntington

disease is exclusively symptomatic. According to the current guidelines, treatment behavioral symptoms can significantly improve the quality of life of patients with Huntington disease.

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### Vascular microdecompression in facial hemispasm

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Facial hemispasm is a rare disorder which might be a result of a conflict between a branch of the vertebral artery and facial nerve at its apparent origin from brain stem. Very rarely the conflict is given by direct contact of vertebral artery itself and the facial nerve. The current presentation refers to clinical aspects, current treatment methods and intraoperative surgical aspects in facial hemispasm. By the use of advanced technology – microscope integrated neuronavigation and electrophysiology – minimally invasive neurosurgical interventions are possible, making surgery an important treatment option, with low risk and solid and rapid effects for the patient.

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### Neuropsychiatric disorders in patients with Parkinson’s disease – descriptive study

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**Introduction.** Despite the fact nonmotor manifestations of Parkinson’s disease (PD) are intensively studied

worldwide, few data are available regarding neuropsychiatric abnormalities in patients diagnosed with PD in Romania. Therefore, the aim of the present observational prospective study is to establish possible correlations between neuropsychiatric symptoms and certain features of the neurodegenerative disease on a population from North-Eastern Romania.

**Materials and methods.** Subjects were recruited from patients diagnosed with PD which were admitted to First Neurology Clinic of “Prof. Dr. N. Obłu” Emergency Clinical Hospital, Iasi within a 2 year period (1.02.2018 to 1.02.2019). A total of 112 patients were included. Each patient was assessed by a clinical psychologist which administered several neuropsychological tests: MoCA, HAM-D, QUIP-RS and GAF.

**Results.** Almost 51% of subjects have minor cognitive impairment, with the highest frequency among patients with an age at onset higher than 66 years. Also, the highest MoCA scores were obtained by patients treated with association between dopamine agonist and levo-dopa. No correlations were established between impulsive-control disorders and dopaminergic treatment. The level of depression influences GAF and MoCA scores inverse proportionally.

**Discussions and conclusions.** Data obtained after statistical analysis follows the main points present in medical literature. However, there are some differences especially regarding dopaminergic treatment: there is lack of correlation between dopamine agonists treatment and impulse-control disorders and dopamine agonists appear to be associated with less cognitive dysfunctions. Both results must be interpreted under the reserve of the short duration of the study. Further, more prolonged studies are required.

**Keywords:** Parkinson’s disease, neuropsychiatric disorders, dopamine agonists

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## Sleep disorders in Parkinson’s disease

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Sleep disorders are commonly encountered in Parkinson’s disease (PD), being reported by more than half of the patients. These symptoms are recognized as being clinically relevant by the PD patients and may significantly affect their quality of life. Sleep disorders in PD can be classified into: disturbances of sleep and disturbances of wakefulness. All types of sleep disorders may

be encountered in PD: insomnia, excessive daytime sleepiness (EDS), rapid eye movement sleep behavior disorders (RBD), restless legs syndrome and sleep-disordered breathing. Insomnia in PD patients has an estimated prevalence of 40% and it has important consequences, like sleepiness, fatigue, mood disorders. EDS is a frequent complaint among PD patients and it is characterized by difficulties in staying awake in passive or active situations. Episodes of sudden onset of sleep might also occur. RBD has a prevalence of 19-70% and constitutes one of the prodromal symptoms of PD, being manifest even before the occurrence of the motor symptoms.

The etiology of sleep disorders in PD is multifactorial and it might be related with the degeneration of the sleep-regulating structures. The severity of the motor features and of other non-motor symptoms might influence the quality of sleep. The assessment of the sleep disorders can be done with generic and specific scales, but also using sleep recording techniques, like actigraphy or polysomnography. This lecture will focus on reviewing the classification, definition and the characteristics of the most frequent sleep disturbances encountered in PD, along with their pathophysiology, assessment, and management.

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## Modified cognitive performance in patients treated by carotid artery stenting – a subdomain analysis correlated with the persistence of microembolic signals

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**Introduction.** The aim of this study was to assess the long term cognitive performance of patients undergoing carotid artery stenting (CAS), with an in-depth subdomain analysis. Also, seeing that microembolic signals (MES) presence in the cerebral circulation is a potential cause for cognitive decline, another objective was to establish if there is a correlation between the altering of different cognitive subdomains and the detection of MES.

**Methods.** 52 patients with significant symptomatic carotid stenosis were monitored for MES detection and

assessed using a battery of cognitive tests 24 hours before and one year after CAS. Both a screening test and tests aimed at various subdomains were used: executive function, work memory, attention, processing speed, language, visuo-spatial orientation, learning and memory. The patients were segregated according to Mini-Mental State Examination score decline: control group with 0/1 point decline and test group with 2/ more points decline.

**Results.** The test group showed significant decline in executive function, work memory, processing speed and attention scores ( $p < 0.05$ ), results that were strongly correlated with the detection of MES at one year after stenting – Rs ranging from 0.54 to 0.89 and OR ranging from 8.5 to 24 in different tests.

**Conclusion.** Patients treated by CAS that present with cognitive decline one year after the procedure have a pattern of cognitive deterioration consistent with the vascular cognitive decline type. In the affected subdomains there is a significant correlation with the persistence of MES detection one year after stenting.

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### A more sensitive-case blood pressure management may be linked to a better outcome in acute ischemic stroke

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**Introduction.** Blood pressure (BP) is elevated in more than 70% of patients with acute ischemic stroke, whether they were hypertensive or not, and is a major risk factor for poor outcome. Although more evidences of BP control were accumulated in the last decades, the optimal BP management during the acute stage still remains unclear.

**Objectives, method.** To analyze the determinants in the BP control in the first week of ischemic stroke and associated with good outcome.

A cohort of 462 patients over 18 months (65% female, mean age 71 y) with acute ischemic stroke due to large artery occlusion was retrospectively analyzed. Premorbid condition and comorbidities were considered, along with stroke complications, interventions for BP control and neurological status after the first week from onset and at discharge.

**Results.** Of the 462 patients with atherothrombotic and cardioembolic ischemic stroke, 406 (87.87%) have

survived, more than 2/3 with moderate and severe disability (mRankin 3-4). Systolic blood pressure at onset was at least 2 cmHg higher than premorbid values (more in previous hypertensive pts) and necessitated at least one antihypertensive agent, except 34 patients: 21 with congestive heart failure or concomitant myocardial infarction and 13 with severe dehydration of different causes. The most common factors that interfere with BP control: limited oral drugs administration (impaired level of conscience and swallowing), overtreatment and unexpected BP variations (cardiac arrhythmias, heart failure, hyperthermia, inadequate water balance).

**Conclusions.** Although with few patients and for a short follow-up period, this study supports the fact that BP control may be difficult in acute phase of stroke, particularly in patients with ischemic stroke due to large artery atherosclerosis or embolic heart diseases. Besides the fact that the moment and the level of BP lowering are still on debate, many individual factors may interfere with BP and require specific interventions.

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### Neurosyphilis and stroke: How, when and how often?

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**Introduction.** Untreated *Treponema pallidum* (TP) infection can be complicated by a vasculitic inflammatory process affecting mainly cerebral small vessels of middle cerebral artery (MCA) and basilar artery (BA) territory, leading to their occlusion.

**Objectives.** To analyze the stroke and CSF characteristics in patients with latent TP infection secondary diagnosed as meningovascular neurosyphilis.

**Methods and results.** Retrospective analysis of 26 HIV-negative patients diagnosed with neurosyphilis in ten years period. 14 patients were diagnosed with meningovascular neurosyphilis and middle cerebral artery stroke syndrome.

We observed an increased CSF protein level in the group of patients with vascular form of disease and pleocytosis in syphilitic meningitis.

Different forms of neurosyphilis can cause the same symptomatology. We noticed statistically significant correlation between vascular damage and cranial nerve palsies ( $p = 0.004$ ).

**Conclusion.** The meningovascular neurosyphilis involving MCA territory was the most common form in our group of patients. The cranial nerve palsies is probable caused by the same inflammatory mechanism (small vessel vasculitis).

The presence of an increased CSF protein levels it's due to the blood-brain barrier damage. Cerebrospinal fluid pleocytosis is most consistent with syphilitic meningitis and the severity of TP infection.

**Keywords:** neurosyphilis, stroke

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## Genetics of dystonia

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Genetic factors play an important role in isolated dystonia, considering that more than 10 percentages of affected persons have a positive familial history. Three causative genes have been associated with isolated dystonia and validated extensively in different published reports, TOR1A, THAP1 and GNAL. Other genes have been found recently associated to the pathogenesis of isolated dystonia, CIZ1, ANO3 and TUBB4 but due to inconclusive results these must be further assess in replication studies. Most of the early-onset idiopathic cases seem to have a monogenic basis with autosomal dominant or sometimes recessive inheritance. Those with late-onset seem to have a more complex pattern of aggregation suggesting genetic and epigenetic/ environmental causes. However, in more than 75% of the cases the responsible gene is not identified by using state of the art knowledge and methods (rendering them to the idiopathic group). Here I will present a review of genetics of isolated and combined dystonia and the way neurologists can approach this complicated field.

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## Gaucher disease and Parkinson's disease – Research Project for Romania

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**Introduction.** Recent research has focused on the association between GBA (glucocerebrosidase) mutations and Parkinson's disease (PD). The risk of developing PD is ten times higher for patients with Gaucher disease than for the general population. Moreover, studies have shown that almost 25% of first and second degree relatives of patients with Gaucher disease are diagnosed with PD. On the other hand, approximately one third of patients with PD are carriers of GBA mutations. These patients have an earlier age of onset, more associated cognitive changes and a poorer response to levo-dopa.

**Aim.** The main objective of our research project is to determine the correlation and association between the presence of GBA mutations and the phenotypic characteristics of PD.

**Methods.** We will test patients with PD from Romania for GBA mutations. After the selection of mutation carriers, we will compare them with a control group of non-carrier patients with PD in order to show if there are any differences in terms of age at disease onset, sex, levo-dopa response, motor and non-motor symptoms between the two groups.

**Conclusion.** Demonstrating an interdependence between GBA mutations and PD will contribute to a better understanding of the pathophysiology of both Gaucher and PD and could have important therapeutic implications for mutation carrier patients.

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## The spectrum of nervous system involvement in Behçet's disease

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Behçet's disease is an autoinflammatory disease with a variable phenotype that can include cutaneous,

ocular, vascular, gastrointestinal, articular and neurologic manifestations.

Neurologic involvement in Behçet's disease is rare but should be taken into consideration for the differential diagnosis of various neurologic diseases of infectious, demyelinating, vascular or neoplastic etiology. The spectrum of neurologic manifestations of this disease (named neuro- Behçet's disease by a recent consensus of experts) can include: 1) a parenchymal syndrome, manifesting with symptoms and signs of brainstem, cerebral hemispheres or spinal cord involvement; 2) optic neuropathy; 3) non-parenchymal syndromes, manifesting as cerebral venous thrombosis, intracranial hypertension (*pseudotumor cerebri*) or aseptic meningitis. Brain and spinal cord magnetic resonance imaging provides important clues for establishing a cor-

rect diagnosis in the majority of cases but interleukin-6 dosing in the cerebrospinal fluid has emerged during the past few years as a valuable additional element for both the definitive diagnosis of neuro- Behçet's disease and the assessment of disease severity. The serum inflammatory markers, the pathergy test and HLA-B51 are extremely useful for the diagnosis of Behçet's disease, but their utility in patients with exclusively or predominantly neurological involvement is limited.

The therapeutic options targeting the neurological manifestations of Behçet's disease have improved significantly over the past few years, so that several immunosuppressive drugs (including azathioprine, methotrexate, infliximab, etanercept and tocilizumab) are currently available for different stages and grades of disease severity.