Machado- Joseph Disease - single center experience from Central Portugal

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We report on a series of 80 spinocerebellar ataxia type 3 (SCA 3) patients from 39 unrelated families followed in our clinic, focusing on novel clinical findings and phenotypical diversity.

Demographic, clinical (scale for the assessment and rating of ataxia (SARA), Montreal Cognitive Assessment, clinical Total Neuropathy Score-cTNS scores), and genetic features were recorded. Neurophysiological evaluation included nerve conduction studies, needle electromyography and cutaneous sympathetic skin response (SSR). Functional imaging using the radioactive tracer 123I-ioflupane was perfomed to assess dopamine deficiency, exclusively in parkinsonian patients. Neuro-ophthalmological assesment including strabismus evaluation, and the use of binocular video-oculography video-horizontal head impulse test (vHHIT) were further performed.

Results and Conclusion

Patients' median age was 53+/-14 years, mean duration of disease was 15 (± 8) (3 – 38) years, median SARA score was 15.4 +/-6, median number of CAG repeats was 71+/-8, and median MoCA score was 25 (18 -29). Three cases presented initially as a levodopa responsive parkinsonian syndrome, and one of them underwent successful deep brain stimulation. Peripheral neuropathy is frequent in our population (>50%), showing predominant sensory involvement. The presence of peripheral neuropathy positively correlates with age but not with triplet expansion size. Strabismus was a universal finding, particularly esotropia at near. Vestibular ocular reflex loss positively correlated with SARA score. A novel saccadic intrusion has been found. In our series we have identified potencial oculomotor biomarkers of disease.