P142

Onset of parkinsonism in a patient with Fabry'sdisease

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Introduction: Fabry disease (FD) is an X-linked lysosomal storage disorder caused by the deficient activity of alpha-galactosidase A. There is a progressive and diffuse lysosomal accumulation of glycosphingolipids in vascular endothelium, kidneys, heart, brain, skin, cornea and other tissues leading to multiorgan damage. Clinically, patients suffer of transient ischemic attacks, strokes, acroparesthesia. Brain white matter lesions are common in MR examinations. We describe the case of a patient with Fabry syndrome with the onset of extrapyramidal signs and cognitive decline.

Case description: A 62 years old male affected by Fabry disease with involvement of kidney, eyes, hear and heart, come to our neurological department for the onset of mask-like face, mild rigidity, gait instability. Family members described difficulty in recent memory, recall, spazial orientation. Neurological examination revealed postural and gait retropulsion, hyperreflexia and positive Babinski's sign and rigidity on both sides. Brain MRI disclosed multiple high signal intensities in the basal ganglia and deep white matter regions and mild upper cortical atrophy. [123I]-FP-CIT confirmed parkinsonism with a reduction of striatal caption especially in the left side. Cognitive evaluation confirmed deficit in memory, spatial orientation and recall.

Conclusion: A lot of recent reports describe the occurrence of parkinsonian signs in patients with Fabry disease [1]. Wise et al. performed an online survey and family history questionnaire to determine the prevalence of PD in 90 FD patients. Gago et al. studied the prevalence of PD in a large cohort of 229 FD patients [2]. An italian MRI-study of Russo C. et al. [3], demonstrated a nigrostriatal involvement in FD patients. All these reports support the hypothesis that there may be an increased risk of developing PD in individuals with *GLA* gene mutations. The presence of cognitive decline in our patient suggests that the brain involvement is more widespread.

References:

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