COGNITIVE IMPAIRMENTS IN FABRY’S DISEASE

N. Longato¹, C. Kleitz¹, L. Di Bitonto¹, B. Jung¹, E. Noel², F. Blanc¹

¹Neurology, ²Medecine Interne, CHU Strasbourg, Strasbourg, France

Fabry’s Disease (FD) is a X-linked genetic disease which enter on early childhood. Renal failure, cardiac insufficiency and cerebral strokes are the most frequent manifestations of this pathology. Only few studies have investigate cognitive dysfunctions.

In our study we aimed to explore cognitive functions in 10 patients with F.D. We also investigate correlations between cognitive dysfunctions and the white matter lesions rate (according to Scheltens et al., 1992) and/or global brain atrophy.

Nine subjects (7 women, 2 men) mean age 51.4 years treated by enzymotherapy, were assessed with a neuropsychological evaluation of memory, executive, attention and visuo-constructive abilities. Anxiety was appreciated with Hamilton’s scale and depression with Beck’s depression inventory. All patients had brain MRI (T1, T2, FLAIR).

Eight patients (89%) present a dysexecutive profile with attentional deficits and cognitive slowing. MRI objective cerebral lesions and global brain atrophy.

This study demonstrates that cognitive impairments exist in F.D. Correlations between white matter lesions, atrophy and cognitive impairments will be discussed after brain MRI results.