P87

Clinical, genetic and neuroradiological characterization of an Italian PFBC cohort

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Introduction: Primary familial brain calcification (PFBC) is a neurodegenerative disorder caused by mutations in seven genes (SLC20A2, PDGFRB, PDGFB, XPR1, MYORG, JAM2, CMPK2).

Objective: To describe the clinical, neuroradiological and genetic analysis of a cohort of PFBC patients.

Methods: PFBC patients seen at the movement disorders clinic of Padova University (2018-2022) were included. Clinical phenotypes, neuropsychological profiles, CT scans (calculating total calcification score, TCS), DaTSCAN features, genetic analyses are presented.

Results: Among 51 patients (mean age at onset 52±14years) 45 were symptomatic (88%). 73% presented at least one movement disorder: parkinsonism (76% with a positive DaTSCAN), tremor (49%), dystonia (25%), cerebellar signs (20%). Anxiety, depression and headache were the most commonly reported non-motor symptoms. 42% had MCI and 9% dementia, with visuospatial functions, language and executive functions being the most involved domains. There was no difference between groups in age at onset, disease duration and clinical phenotype. 10 patients carried mutations in SCL20A2, 3 in PDGFB, 1 in PDGFRB, 1 in XPR1, 2 in JAM2 (monoallelic mutation), 6 in MYORG (6 biallallelic, 1 monoallelic), 21 tested negative.
We observed 3 symptomatic monoallelic MYORG and JAM2 mutation carriers. Bilateral lenticular calcification were observed in 100% of patients, followed by dentate nuclei (63%), thalami (51%), caudate nuclei (49%), white matter (WM, 41%). Symptomatic and asymptomatic subjects showed no difference in TCS, without correlation between TCS and age at onset, age at CT scan and disease duration, with the exception for age at CT scan and TCS in SLC20A2 patients.
MYORG and SLC20A2 patients showed higher TCS score than the indeterminate genetic group, whereas MYORG patients had a higher score at DN, thalami and WM than the indeterminate genetic group.

Conclusions: Our study confirms that no clear correlation between genotype and clinical-radiological phenotype exists in PFBC patients.