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## Brain parenchyma sonography findings in patients affected by Parkinson's disease associated with GBA gene mutations

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*Introduction:* Parkinson's disease associated with GBA mutations (PD-GBA) is a common cause of early onset PD, with a peculiar phenotype characterized by prominent non-motor features and even in the early stages of the disease. Single-photon emission computed tomography (SPECT) with DaT has high specificity and sensitivity, but in early stage of disease can be negative [1]. Brain Parenchyma Sonography (BPS) have been shown to be a useful tool in detecting early stage of PD and bilateral substantia nigra (SN) hyperechogenicity was detected in PD and already in the premotor stage [2]. Previous studies have found that BPS findings in PD-GBA is similar to those of patients with sporadic PD [3].

*Objective:* The aim of this study is to describe the BPS finding in a series of PD-GBA patients which could be consider as a biomarker both in early stages and in advanced PD-GBA.

*Methods:* Case series.

*Results:* Five patients affected by PD-GBA were investigated by BPS. Three patient carried the N370S mutation, one patient carried the E365K mutation and one patient was found to be carrier of a new mutation c.1312C>T. The age at onset was 48.5 ±9.9 years (range 40-59). The disease duration was 9.3 ±11.0 years (range 1-25). A DAT SPECT showed bilateral dopaminergic denervation in 4 patients and unilateral in one; a BPS revealed bilateral and symmetric hyperechogenicity of SN in 3 patients, asymmetrical hyperechogenicity, contralateral to the site of motor onset, in the others two.

*Conclusions:* GBA mutation carriers with PD have greater hyperechogenicity similar to sporadic PD. The mechanism underlying parkinsonism in these patients is still debated. The presence of this sonographic finding is consistent with iron deposition. Larger simple size is needed to explore possible differences in nigral echogenicity in PD patients with different genotipes.

**References:**

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