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[The clinical features of Parkinson's disease in patients with mutations and polymorphic variants of GBA gene].

[Article in Russian; Abstract available in Russian from the publisher]

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Abstract in **English**, Russian

BACKGROUND: Mutations in the glucocerebrosidase gene (**GBA**) increase the risk of **Parkinson's disease** (PD) by 6-10 times in all populations and are associated with the early-onset of PD, development of cognitive impairment and presence of psychotic disorders. At the same time, polymorphic variants associated with the twofold increase in the risk of PD were also described in the **GBA** gene.

AIM: To estimate the clinical features of PD in patients with mutations and polymorphic variants of the **GBA** gene.

MATERIAL AND METHODS: Evaluation of motor, cognitive, emotional, psychotic and autonomic dysfunctions in patients with mutations (N370S, L444P) and polymorphic variants (E326K, T369M) in the **GBA** gene was performed using clinical scales.

RESULTS: Patients with mutations (mGBA-PD), and with polymorphic variants (pGBA-PD) in the **GBA** gene were compared with the group of patients with sporadic PD (sPD). Compared to sPD, affective disorders (depression and anxiety) were more expressed in the mGBA-PD group (p=0.001) and the general **GBA**-PD group (p=0.001) assessed with Sheehan anxiety rating scale, in the pGBA-PD group (p=0.012) and the general **GBA**-PD group (p=0.05) assessed with the NPI, in the mGBA-PD (p=0.003), pGBA-PD (p=0.022), and general **GBA**-PD groups (p=0.001) assessed with the Hospital Anxiety and Depression scale (HADS 'A'), and in the pGBA-PD group (p=0.005) assessed with the HADS 'D'. Non-motor symptoms assessed with the PD-NMS were more expressed in the pGBA-PD patients (p=0.007) and in the total group with **GBA**-PD (p=0,014) compared to sPD. Cognitive impairment measured with MMSE was more marked in mGBA-PD patients (p=0.022). Differences in motor and non-motor clinical symptoms between pGBA-PD and mGBA-PD groups were not found.

CONCLUSION: Thus, clinical features of non-motor symptoms were described both in carriers of **GBA** mutations and polymorphisms. Identification of the specific clinical phenotype of PD in carriers of **GBA** polymorphic variants is important due to their relatively high prevalence in PD patients.

KEYWORDS: GBA; **Parkinson**'s **disease**; glucocerebrosidase; non-motor symptoms

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