

CLINICAL AND GENELOGICAL FEATURES OF PARKINSON'S DISEASE

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Objective: The initial purpose of the work is to study the clinical and genealogical features of Parkinson's disease.

Background: Currently, clinical genetics has a number of methods for obtaining information to identify patterns inherent in hereditary diseases. The genealogical analysis is one of the affordable and cost-effective methods, which often plays an important role in the prediction, diagnosis, and adequate therapy of diseases.

Method: The study included 213 patients with PD, 90 women, 123 men, whose average age was 46.17 ± 0.63 years, mainly of Uzbek nationality. A control group of 20 healthy people of Uzbek nationality of the same age without signs of PD. PD patients were selected in accordance with the international criteria of the British Brain Bank "Parkinson's Disease Society Brain Bank". To exclude other causes of Parkinson's syndrome, patients underwent CT or MRI of the brain. In PD patients, the form of the disease (akinetic-rigid, mixed, or tremulous) and the functional stage of the disease (according to the Hoehn-Yahr scale) were assessed.

Results: Due to the supposed differences in the pathogenesis of early and late forms of PD, the main group was into subgroups based on the age of onset of the disease. The subgroup of early parkinsonism consisted of 79 patients in whom the symptoms of the disease manifested before the age of 45, and 76 patients with a burdened family history (it is in these categories of patients that the probability of detecting mutations is maximal), the subgroup of late parkinsonism accounted for 58 patients with an age of onset of primary parkinsonism > 45 years. A detailed pedigree was compiled, which included information about diseases in 2-3 generations of the family. A total of 1741 people were analyzed in the model population. A burdened family history of PD was observed in 76 cases, 44 men, 32 women. In families of probands, PD in generations in relation to the total number of patients with each concentration is III - 16; II- 23; I- 37. The burden in the population of practically healthy people was observed much less: III - 0%; II - 0%; I- 0%.

Conclusion: It turned out that PD is more often affected by relatives of the I degree of kinship 37, which in relation to the total number of patients with this concentration is 35.68%, and men accounted for 44 cases. In one case, there was a burdened family history of PD, with early-onset at 45 years of age, and two-family cases with PD in a cousin and a sister.

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