Contemporary study of the difficulties of late diagnostics in patients with Huntington's disease in Uzbekistan

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Objective: To identify the causes that lead to late diagnosis of Huntington's disease.

Background: Huntington's disease is a rare, progressive, and fatal autosomal dominant neurodegenerative disorder, typically of adult-onset.

Method: To this study was included 34 patients with a confirmed molecular genetic diagnosis, followed up with us from 2021 to the present.

Results: According to our observations, the disease often debuts with mental disorders (5 families), in connection with which these patients are observed in psychiatric hospitals and examined by neurologists and geneticists out of time or do not receive consultations from these specialists at all. We have described a patient with Huntington's disease, in whom the disease debuted with cranial dystonia. In 2 families, the onset of the disease is observed in the son earlier (at the age of 35) and more severe than in the father (after the age of 70) – the phenomenon of anticipation. An allele with incomplete penetrance (36–38 repeats) was identified in 2 families.

Conclusion: The main reasons leading to late diagnosis of Huntington's disease are identified: 1) The possibility of disease manifestation with cranial dystonia or other atypical neurological symptoms; 2) The presence in the population of alleles with incomplete penetrance, which complicates the early diagnosis of the disease due to the frequent absence of family history, in some cases – the minimum severity of choreic hyperkinesis and intact intelligence; 3) The phenomenon of anticipation, leading to diagnostic errors since the disease debuts later in parents than in children, and has an erased clinic; 4) Manifestation of the disease with mental disorders and frequent cases of suicide in the family before the start of a typical clinic, which leads to long-term observation by psychiatrists.

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