

Abstract

Huntington's disease (HD) is an autosomal dominant inherited neuro-psychiatric disease with usual onset in the middle age. The mutation, located on the short shoulder of chromosome 4, is an expansion of a nucleotide triplet, containing cytosine, adenine, guanine (CAG), with critical limit of 40+ repetitions. The principal symptoms include motor symptoms (chorea, dystonia, disorders of voluntary movements), progressive cognitive deterioration and neuropsychiatric symptoms (behaviour disorders, affective symptoms and so on). The clinical diagnosis is confirmed by a genetic test, which may also be carried out presymptomatically in offsprings of the diseased person.

The objective of the 1st study consisted in the characterization of differences in psychiatric examination and neuropsychological testing among the people at risk (PAR), in whom it was recommended to delay the test, and people at risk, who were recommended to continue in the so-called predictive protocol. The total of 52 people have been examined (32 females, 20 males). In addition to the common psychiatric examination we have also administered the Eysenck Personality Questionnaire (EPQ-A), self-rating scale of general psychopathology (SCL-90), three short cognitive tests – Trail making test, test of Verbal fluency and Auditory verbal learning test and quality of life questionnaire (MANSA). PAR with the recommended test delay showed statistically significant lower rate of neuroticism and lower EPQ-A lie score, higher values on the phobia scale and the so-called "positive symptom distress index" in v SCL-90 and significantly lower quality of life in MANSA than PAR with the recommendation to continue in the test. The positively tested PAR showed statistically significant higher EPQ-A neuroticism rate and lower extroversion rate than the negatively tested PAR, however, no statistically significant differences in the total rate of psychopathology and in any cognitive tests have been found. The results indicate that the formalized psychological testing in the prediction of recommendation to perform a genetic test or to delay it does not bring significant information. In the process of indication or delay of the predictive genetic test, the clinical psychiatric examination remains the decisive factor. The motivation of applicants, which cannot be assessed by the application of scales, is considered one of the most important factors in the decision-making process.

The objective of the 2nd study consisted in the assessment of the most significant problems which are dealt by caregivers (CGs) of the patients with HD from the first symptoms up to the developed stage of the illness. The information from 21 CGs was obtained from the structured interview prepared in the form of a questionnaire. We have evaluated the following principal areas: a) problems with the obtaining of adequate information about HD in the first stages, b) main problems associated with diagnosing, c) cooperation and communication with doctors, d) clinical symptoms of HD which most affect the life of CGs, e) information about the supportive group and its usefulness for CGs. Fourteen out of the total of 21 CGs have had serious problems with the obtaining of adequate information on HD in the early stages of the disease, seven out of the total of 21 CGs have been informed adequately. The main problem of the diagnosing was the risk of HD for children of the patients. Fifteen out of the total of 21 CGs described the cooperation and communication with doctors as very problematic, six out of the total of 21 CGs had good experience with the doctors. The main points of dispute mentioned by the CGs included insufficient interest of doctors in family/social conditions and almost no contact with the partners. As the worst symptoms of HD, changes in behaviour and affective disorders were described by twelve out of the total of 21 CGs, dementia by ten out of the total of 21 CGs and speech disorders by seven out of the total of 21 CGs. The awareness of doctors and medical facilities about the existence of the supportive group was considered insufficient by sixteen out of the total of 21 CGs. In conclusion, we may say that the issues of CGs are neglected by the doctors – it is necessary to accept the CG as a client and to realize that many CGs need medical aid and social/psychotherapeutic support.

Key words: Huntington's disease, CAG triplet, genetic test, predictive genetic test, caregiver, people at risk