

Huntington's disease-case report

ŠEJMA BEGANOVIĆ¹, BABAJIĆ ERMIN², GRAPKIĆ JASMINA³

¹UNIVERSITY OF TUZLA, FACULTY OF MEDICINE, TUZLA, BOSNIA AND HERZEGOVINA

²UNIVERSITY OF TUZLA, FACULTY OF MEDICINE, DEPARTMENT OF NEUROLOGY, TUZLA, BOSNIA AND HERZEGOVINA

³UNIVERSITY HOSPITAL CENTRE TUZLA, CLINIC FOR NEUROLOGY, TUZLA, BOSNIA AND HERZEGOVINA

sheyma10@hotmail.com

Huntington's disease (HD) is autosomal dominant neurodegenerative disease that never skips generations. It starts between 30 and 50 years of age and ends after 15-20 years with death. It is a disease of CAG triplet repeats and is characterized by poliglutamine repeats. The number of CAG trinucleotide repeats correlates with the age of the onset of the first symptoms, as well the clinical picture. The selective therapy for Huntington's chorea still does not exist and for symptomatic treatment the blockers of dopamine have turned out to be the most useful. Its prevalence in the world is 8-10 per 100000 inhabitants and in Bosnia and Herzegovina 4,46 per 100000 inhabitants.

We report a case of a 50-year old patient with HD. The symptoms started 2 years before hospital admission when the patient noticed "muscle flutter" on the left side of his face and left shoulder and occasionally on the upper leg. The patient has been hospitalized in the Neurology Clinic in the period of 02-14.08.2014. when he was released under the dg, Distonia. Laesio n. ischiadici sin post vulnum explosivum aa XX. Applied therapy didn't bring any results, patient is less talkative, sometimes can't control his sphincters and has risen amplitude of choreas. The patient is sent on Institute for genetical engineering and biotechnology on University of Sarajevo. When genetical analysis is done suspicion between HD and WD is solved. Sequestering egsones of genotype showed abnormal genotype for HD and normal genotype for WD. "Patient is carrier of one allele from 27 CAG repeatings and one allele from 48 CAG repeatings in the structure of HD gene".

Genetical consulting is important for presenting the results to healthy family members for future life. Approach needs to be multidisciplinary whether the results are good or bad.

Keywords: HD, genetics, chorea.

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