Abstract

Huntington´s disease (HD) is an autosomal dominant inherited disorder with manifest of symptoms around the age of 40. This disorder is caused by an expansion of CAG repeats in huntingtin gene, Huntingtin (Htt) is a protein expressed in almost all tissues. HD is mainly characterized by neurodegeneration in the basal ganglia and cerebral cortex, but mutation in huntingtin have also serious influence on peripheral tissues. Many studies show serious heart dysfunction, weight loss, altered glucose homeostasis, impairment of energetic metabolism and muscular atrophy in HD patients and animal models. Till now, mechanism of these changes has not been sufficiently described and there is nor an adequate treatment yet.

Key words:

Huntington´s disease, mutated huntingtin, CAG repeat, peripheral tissue