DEPARTMENT OF MOLECULAR BIOLOGY AND HUMAN GENETICS

22. GENETIC ASPECTS OF THE HUNTINGTON DISEASE

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Background. Huntington disease (HD) is an incurable, adult-onset, autosomal dominant inherited disorder associated with cell loss within a specific subset of neurons in the basal ganglia and cortex. HD is named after George Huntington, the physician who described it as hereditary chorea in 1872. Characteristic features of HD include involuntary movements, dementia, and behavioral changes.
Case report. Purpose and objectives: This study focuses on the variability of the HTT gene expression and its correlation with the onset of the disease. It also outlines the genetic aspects of the disease: types of inheritance, anticipation and the frequency of new mutations in the population. Etiology: The selective neuronal dysfunction and subsequent loss of neurons in the striatum, cerebral cortex, and other parts of the brain can explain the clinical picture seen in cases of HD. Several mechanisms of neuronal cell death have been proposed for HD, including excitotoxicity, oxidative stress, impaired energy metabolism, and apoptosis. Treatment: In a study published recently in the New England Journal of Medicine, researchers from UBC and their colleagues have demonstrated for the first time that the drug IONIS-HTTRX (now known as RO7234292) successfully lowered the levels of the mutant huntingtin protein - the toxic protein that causes Huntington disease -- in the central nervous system of patients. The treatment is designed to silence the gene. On the trial, 46 patients had the drug injected into cerebrospinal fluid. The first in-human trial showed the drug was safe, well tolerated by patients and crucially reduced the levels of huntingtin in the brain. Experts say it could be the biggest breakthrough in neurodegenerative diseases for 50 years.
Conclusion. Taking into consideration the above description of the new trial treatment of the disease, the studies made in this field could be crucial for the next generations.
Key words: Huntington's disease, anticipation, penetration, treatment, mutation

23. RECURRENT AORTIC DISSECTION: A PECULIAR COMPLICATION OF MARFAN SYNDROME

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Background. Marfan syndrome (MFS) is an autosomal dominant disorder caused by a mutation in FBN1 gene which involves abnormal connective tissue. MFS affects different parts of the body such as bones, joints and eyes, but the most serious complication involves cardiovascular system. Acute aortic dissection (AD) is a life-threatening condition caused by a tear in the intimal layer of the aorta or bleeding within the aortic wall, resulting in the separation of the layers of the aortic wall. Even though AD is a characteristic complication of the MFS, recurrent aortic dissection (RAD) is a rare phenomenon where MFS is a strong independent