Abstract

Huntington´s chorea is a dominantly inherited disease caused by trinucleotide (Cytosine-Adenine -Guanine) expansion in a gene coding huntingtin protein. Carriers of these mutation show symptoms associated with motor impairment, a cognitive and psychiatric disturbance, which is called Huntington´s disease (HD). The major sign of HD is striatal atrophy in the middle age of life. Since it is known that huntingtin protein participates in a lot of cellular processes, such as transcriptional regulation and metabolism, these processes change by its mutation. One of the features observed in HD pathogenesis is the presence of oxidative stress. The aim of the work was to monitor the molecular changes preceding the HD manifestation in the knock-in minipig model. As a material for monitoring molecular changes leading to this condition, primary fibroblasts were used. Whereas, the oxidative stress arises from an imbalance between oxidants and antioxidants, level of reactive species and lipid peroxidation together with expression of antioxidant response associated genes was measured. At the same time, expression of metabolic and DNA repair related genes was monitored. Although the differences in oxidative stress level or the expression of antioxidative response genes were not detected, the changes in the expression of metabolic and DNA repair related genes were observed in fibroblasts from knock-in minipigs compared to their wild-type siblings.