

ABSTRACT

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Title of diploma thesis: Human prion disease – CREUTZFELDT-JAKOB DISEASE

Objective: Theoretical part of the thesis includes a comprehensive theory not only about prions, but also about Creutzfeldt-Jakob disease in general. The aim of the experimental part was to evaluate and quantify the cerebrospinal fluid and brain tissue of eight individuals suspected from prion disease, Creutzfeldt-Jakob disease specifically.

Methods: To determine the presence of 14-3-3 protein in cerebrospinal fluid and prion protein in the native brain tissue an analytical method called Western blotting was used. Results were expressed only qualitatively. For better differential diagnosis, immunological examination of so-called CSF triplet (p-TAU, h-TAU, β -amyloid) was completed. The quantitative determination was performed by enzyme immunoassay (ELISA).

Results: All eight subjects suspected from the prion disease were confirmed with the diagnosis of Creutzfeldt-Jakob disease. Determination of the prion protein *post mortem* has a great benefit for final confirmation of the disease. Detection of presence of 14-3-3 β -subunit is not specific as it is positive for all fast decays of brain tissue. During the examination of CSF triplet the β -amyloid concentrations were estimated from normal to slightly reduced values whereas the total h-TAU protein concentrations were significantly increased. These findings are typical for Creutzfeldt-Jakob disease. Measuring the concentrations of the phosphorylated form of TAU (p-TAU) protein gave values within the physiological range or slightly elevated.

Conclusion: Our results of eight samples determined for differential diagnosis of Creutzfeldt-Jakob disease correspond to the values stated in the current literature

and therefore the methods can be advantageously used for more precise diagnosis of this disease.

Keywords: prion protein, prion disease, Creutzfeldt-Jakob disease, 14-3-3 protein, h-TAU, p-TAU, β -amyloid