Ana Ivancheva et al., J Alzheimers Dis Parkinsonism 2017, 7:4(Suppl)
DOI: 10.4172/2161-0460-C1-028

Prion disease – The last diagnosis

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A 73-year-old lady was referred to a neurologist after two geriatric consults due to deteriorating cognitive function, ataxia and weakness. She was completely dependent in slightly over one year. From walking with a frame, she became wheelchair bound. Her cognition continued to decline and could not feed herself due to apraxia. When assessed she was found to have rigidity, myoclonus and left-right disorientation. Overall impression was that she had prominent extrapyramidal and cortical disease.

Diagnosis: Due to rapid symptoms progression, typical MRI brain findings and highly specific CSF 14-3-3, S100B and RT-QuIC, she was diagnosed with probable sporadic Creutzfeldt-Jakob disease(sCJD). Brain biopsy post-mortem would confirm the diagnosis.

Discussion: SCJD has unclear mechanism of initial conformational change of prion protein. CJD has worldwide distribution, where 85% are sCJD and occurs at rate of 1 in 1,000,000 population per year. Mortality rate is less than 1 per million depending on country. Researchers mention difficulties in ascertaining accurate incidence, prevalence and mortality rate due to under-reporting, under-awareness and poor diagnostic capability. Possibly CJD is higher than thought due to misdiagnosis

Biography

Ana Ivancheva graduated from Medical University of Sofia in 2016. She has completed her internship in Sofia, Bulgaria. She is currently completing BST programme in internal medicine at MUHI. She is an author in a medical textbook in Bulgaria and is currently pursuing her interest in academic writing.

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