Prion disease – The last diagnosis

Hakim Ghani and Ana Ivancheva
Mayo University Hospital, Ireland

Abstract

Case Presentation

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.

Investigations and Results

First geriatrician:

Routine blood investigation	Normal
CT brain, carotid angiogram, thorax abdominal and pelvis(TAP)	Normal
Echocardiogram Holter	Normal

Second geriatrician:

EMG	Polyradiculopathy
MRI brain	Vasculopathic changes

Neurologist:

MRI brain	Diffusion restriction
DaTscan	Normal
EEG	Frontotemporal slowing
Serum protein electrophoresis	Normal
Hepatitis screen, syphilis, HIV	Negative
ANA, PR3, MP0 NMDA, Yo, Hu, Ri, Ma, Ta, amphiphysin, CV2/CRMP5, VGCC/VGKC, GAD, Purkinje cell, glycine receptor antibodies	Negative
14-3-3 S100B RT-QuIC	Positive

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 MUHl. She is an author in a medical textbook in Bulgaria and is currently pursuing her interest in academic writing.

anaivancheva@gmail.com