Meta Analysis: Prion Diseases

Daniela Calzada
Mentored by Dr. Denise Signorelli, PhD. College of Southern Nevada Biology Department

Introduction: Prions are a mutation of the PRNP polymorphic 129 codon. It is a misfolded protein that takes an abnormal shape that will infect other variants of the same protein. It affects different areas of the brain, however, no recent research has been found as to why these variants do so. This project explores the different variants of prion diseases. They are Familial Fatal Insomnia, Sporadic Creutzfeldt Jacob, and Gerstmann-Sträussler-Scheinker disease.

Overview/Methods: For each of the three diseases mentioned they have their own testing to determined the presence of each prion disease. Such as RT-QuIC (Real-time quaking-induced conversion), Polysomnography, and genetic testing.

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Figure 1

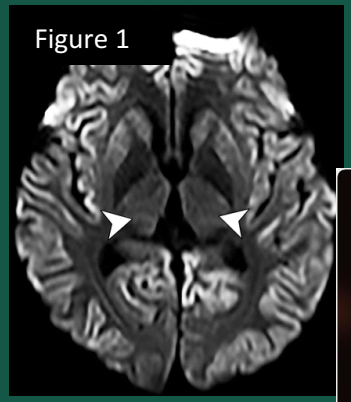

Fig 5 Courtesy of Dr. Bose, *AZO Life Sciences*. Following image displays proteins structure of a normal prion protein (left) and an abnormal prion protein (right).

Figure 2

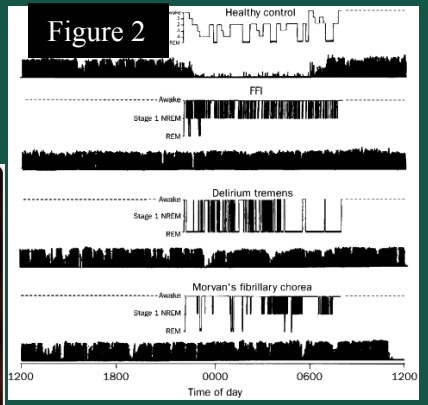

Figure 3

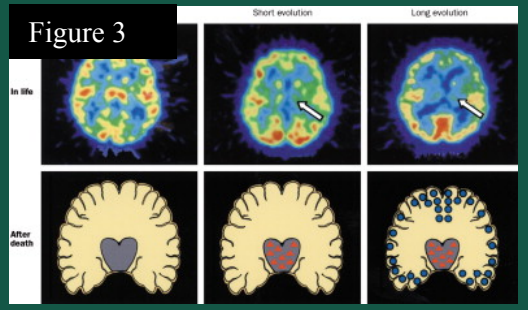

Fig 1. Courtesy of Fragoso et al, *RSNA*. The following image is of a woman in her early 30s with a sCJD subtype with valine homozygosity.
Fig 2. Courtesy of Montagna et al, *The Lancet Neurology*. Sleep histogram of a healthy control and a patient with FFI. Deep sleep stages as well as slow EEG are lost.

Figure 4

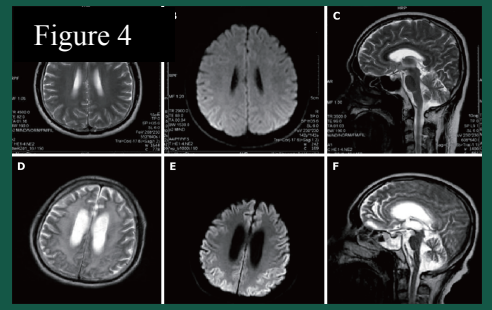

Fig 3. Courtesy of Montagna et al, *The Lancet Neurology*. Top panel shows patients with long and short durations of the disease. Bottom panel indicate the triangles as neuronal loss and circles as spongiosis.
Fig 4. Courtesy of Zhao et al, *Baishideng Publishing Group*. Top panel obtained after 2.5 years symptoms onset with no obvious abnormal signs. Bottom panel obtained after 5.5 years symptoms onset with visible abnormalities.

Sporadic Creutzfeldt Jakob Disease
Sporadic prion mutations appear with no prior genetic history. Spontaneous mutations in the prion gene make aberrant proteins which cause the misfoldling of normal proteins. These mutations are not inherited and the protein has not been transmitted from another infected individual. . Out of the three categories, sporadic proteins are the ones that researchers have no idea how they come to be or have any indicators that show if or when or why they'll appear. They affect the cerebral cortex RT-QuIC, particularly CSF RT-QuIC (Cerebral Spinal Fluid Real-Time Quaking-Induced Conversion), is able to detect small amounts of PrPSC ability to bind to fluorescent dye, thioflavin T.

Fatal Familial Insomnia:
As the name suggests, the recipient of the disease will have an inability to fall or stay asleep, short term memory loss, difficulty concentrating, difficulty coordinating movement, and sweat excessively. It primarily affects the thalamus which is responsible for the sleep wake cycle. Polysomnography of REM and NREM sleep can suggest the FFI diagnosis, which can be confirmed by sequencing the genetic lesion. This inherited mutation is 100% fatal.

Gerstmann-Sträussler-Scheinker Disease:
This disease affects the cerebellum. Like FFI, it is also inherited. It is a rare neurodegenerative disease with symptoms such as ataxia, difficulty walking, dysarthria, and nystagmus. Similar to sCJD testing, CSF RT-QuIC can be used along with MRI imaging and genetic testing.

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