Creutzfeldt-Jakob Disease (CJD)

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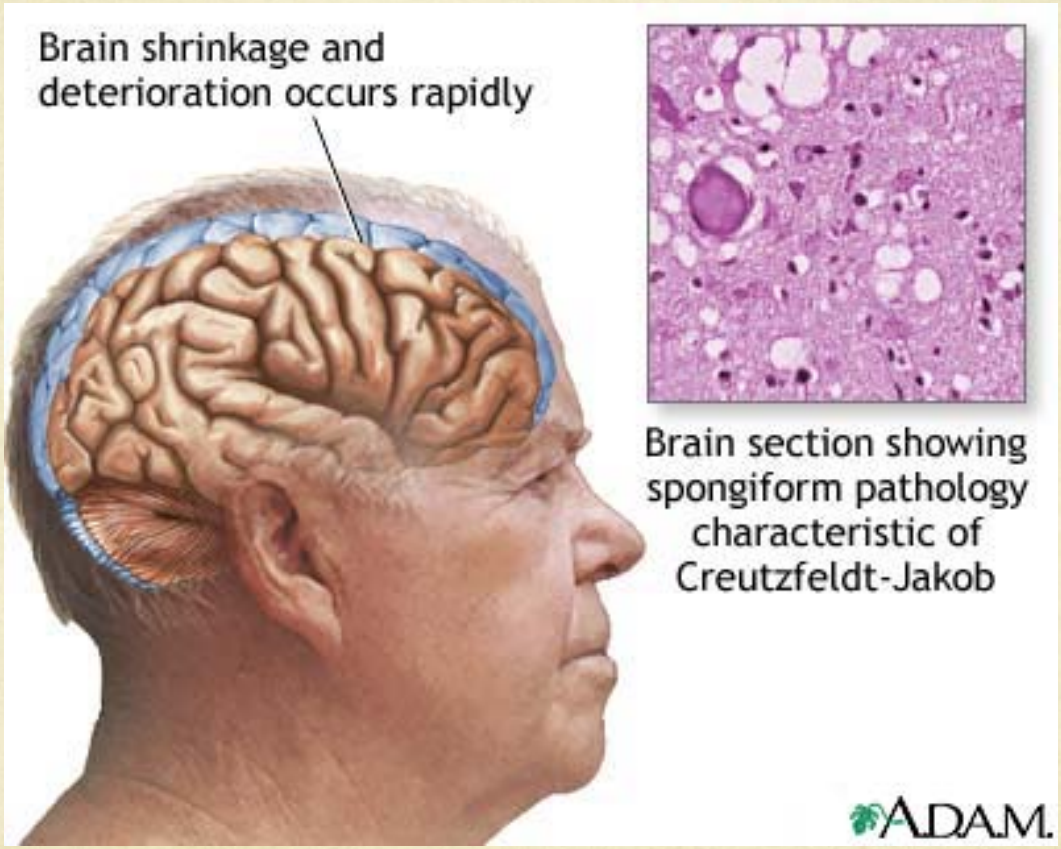
Creudtzfelt-Jakob Disease: An Overview

- A type of spongiform encephalopathy closely related to Gerstemann-Straussler-Scheinker disease, Fatal Familial Insomnia, Mad Cow Disease, Scrapie, and Kuru.
- Caused by a defective prion protein (PRNP; PRP;PRIP) that is able to infect the tissue around it.
- The disease is autosomal dominant, meaning that children have a 50% chance of inherited the mutation.
- Onset is between 30-90 years of age
- Can take its course anywhere from a few months to 5-7 years, varying widely
- Death is usually from infection such as pneumonia or urosepsis.

Pathology


Brain shrinkage and deterioration occurs rapidly
Brain section showing spongiform pathology characteristic of Creutzfeldt-Jakob
A.D.A.M.

Symptoms

- Cognitive difficulties
  - Dementia
  - Psychiatric symptoms
- Muscular difficulties
  - Myclonus
  - Ataxia
  - Weakness or spasticity
  - Chorea
- Neurological difficulties
  - Stroke-like episodes
  - Dysarthria

Diagnostics

- There is no single diagnostic used to identify CJD. Most diagnostics are used to support a conjecture of the disease.
- CDJ in neuropathologic form shows spongiform degeneration and astroglioses throughout the cortex and deep nuclei of the brain.
- Electroencephalograms
- Brain imaging
- Examination of cerebrospinal fluid (CSF)

The Gene and Novel Diagnostics

- The gene associated with CDJ is a prion protein known as PRNP (also PRP and PRIP) located at 20pter-p12.
- The location of the gene has allowed novel diagnostics in the form of sequencing and analysis. The new diagnostic is molecular genetic testing and PRNP targeted mutation analysis. The absence of PRNP mutation by the test does not necessarily rule out a genetic prion disease.
- Several sequencing mutations have been recorded and there appears to be no definitive one. There appears to be a duplication of 1-9 additional octapeptide repeats (Pro-His-Gly-Gly-Gly-Trp-Gly-Gln) whereas normal alleles have five.
- In addition, there appears to be an glu200-lys variation that has been strongly linked to CJD.
- Overall, the specific mutation within the gene varies-- families with CJD often have a different mutation than others with CJD.

Treatment

- There is no cure and virtually no treatment for CDJ as is the case for all prion diseases. The location of the gene has not added any novel treatment of the disease. Because scientists are still unsure exactly of the material within prions (they are proteins and therefore have no genetic makeup but are still able to convert other proteins), they cannot create an effective treatment or medication. Thus, treatment is aimed at some more severe symptoms.
- Medications:
- Antiepileptic drugs (diphenylhydratoin or carbamazepine) for seizures
- Clonazepam for myoclonus
- Occasionally, a permanent feeding tube is used for dysphagia

Works Cited

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