Creutzfeldt Jakob Disease (CJD)

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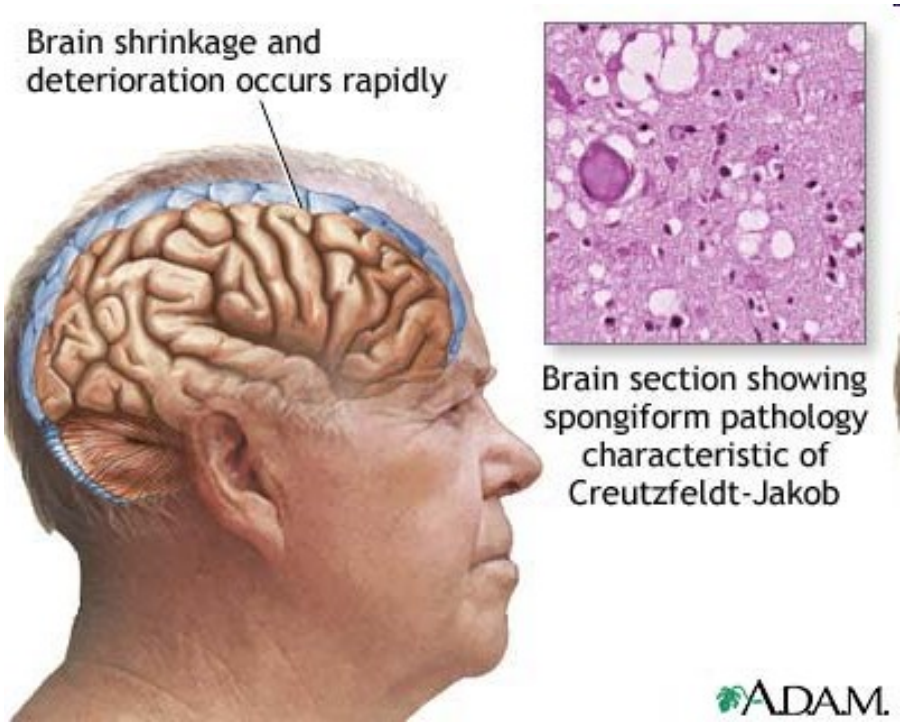
Overview of Creutzfeldt-Jakob Disease

- Rare, fatal, neurodegenerative disease
- Transmissible Spongiform Encephalopathy (TSE)
- Transmission: sporadic, hereditary or acquired
- Cause: defective prion protein that infects tissue
- Average age onset: 60
- Death usually from infection

Symptoms of Classical CJD Diagnosis



- Cognitive
- Muscular
- Neurological

Diagnostics of Creutzfeldt-Jakob Disease

- Only way to confirm a diagnosis of CJD is brain biopsy
- CJD in neuropathologic form shows spongiform degeneration and astroglioses
- Most diagnostics are used to support a conjecture of the disease
  - Cerebrospinal Fluid (CSF) Analysis
  - Brain imaging
  - Electroencephalograms (EEG)
  - Olfactory biopsy

The Gene and Novel Diagnostics

- The gene associated with CJD: a prion protein (PRNP) located at 20pterp12
- New diagnostic: molecular genetic testing and PRNP targeted mutation analysis
- No definitive sequencing mutation
  - 1-9 additional actapeptide repeats *(Pro-His-Gly-Gly-Gly-Trp-Gly-Gln)*
  - glu200-lys variation
- The specific mutation within the gene varies – families with CJD often have different mutation than others with CJD

Treatment of Creutzfeldt-Jakob Disease

- Because scientists are still unsure exactly of the material within prions, they cannot create an effective treatment or medication
- Treatment aimed at severe symptoms:
  - Sodium valproate and clonazepam for myoclonus
  - Antiepileptic drugs (diphenylhydration or carbamazepine) for seizures
  - Feeding tube for dysphagia
  - Quinacrine?

References

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