

Editorial on Creutzfeldt-Jakob Disease

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Editorial

Creutzfeldt-Jakob disease (CJD) is an extremely rare degenerative brain disorder that usually occurs later in life. Patients may experience failing memory, behavior changes, impaired coordination, and vision problems in the early stages. As the disease progresses, mental deterioration becomes severe, and they can have uncontrolled movements, blindness, weakness, and go into a coma.

This condition often leads to death within a few weeks or months after symptoms begin. In approximately 90 percent of cases, CJD appears to occur randomly for no apparent reason (sporadically). About 10 percent of affected individuals may have a hereditary predisposition for the disorder. It occurs in approximately one in every one million people worldwide.

Symptoms

The primary symptoms includes subtle signs of confusion, depression, forgetfulness, sleeping difficulties (insomnia), and/or behavioral changes, impaired vision, abnormal physical sensations, and/or difficulties with voluntary coordination. Abnormal sensations (dyesthesia) or pain in the face, arms, and legs, cerebellar ataxia, hypotonia, halting speech, chorea and dementia often occurred.

Causes

Researchers believe that a transmissible agent causes Creutzfeldt-Jakob disease (CJD). The agent is called a prion, and it is thought to transform normal protein molecules into infectious ones. These proteins occur in both a normal form, which is a harmless protein found in the body's cells, and in an infectious form, which causes disease.

CJD may develop from direct contamination (transmission) with abnormal prion protein in infected brain tissue or from changes (mutations) in the gene* that regulates (encodes for) the production of the human prion protein. Abnormal changes in the prion protein are thought to play some role in causing deterioration in certain areas of the brain, appearing as sponge-like holes and gaps. The gene that regulates the production of the human prion protein, known as prion-related protein or PRNP.

Diagnosis and Treatment

According to the medical literature, CJD should be considered in adults who experience a sudden onset of rapidly progressive dementia and there is currently no single diagnostic test for Creutzfeldt-Jakob disease (CJD). Computerized tomography and Magnetic resonance imaging (MRI) are often used that can help in diagnosing CJD. Brain biopsy or autopsy is an only way to confirm a diagnosis of CJD. The treatment of CJD is symptomatic and supportive. Affected individuals should be carefully monitored to help guard against infections. Researchers have tested many drugs, including amantadine, steroids, interferon, acyclovir, antiviral agents, and antibiotics. The treatment for CJD is aimed at alleviating symptoms and making the patient as comfortable as possible.

References

1. <https://rarediseases.org/rare-diseases/creutzfeldt-jakob-disease/>
2. <https://rarediseases.info.nih.gov/diseases/6956/creutzfeldt-jakob-disease>

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