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Genetic case in Creutzfeldt-Jakob disease

Arantxa Oliver Ramírez, A Del Ángel Arroyo Sebastián, MJ Crespo Tudela, MJ Sánchez Romano, G Céspedes
Family Doctors in Alicante, Spain

Corresponding author: Dr Arantxa Oliver Ramírez, Doctor, Family Doctor, Alicante, Spain.
E-mail: arantxaoliver@yahoo.es

Creutzfeldt-Jakob disease (CJD) is a rare, degenerative, invariably fatal brain disorder. It affects about one person in every one million people per year worldwide. CJD usually appears in later life and runs a rapid course. Typically, onset of symptoms occurs about age 60, and about 90 percent of individuals die within 1 year. In the early stages of disease, people may have failing memory, behavioral changes, lack of coordination and visual disturbances. As the illness progresses, mental deterioration becomes pronounced and involuntary movements, blindness, weakness of extremities, and coma may occur.
There are three major categories of CJD:

- In sporadic CJD, the disease appears even though the person has no known risk factors for the disease. This is by far the most common type of CJD and accounts for at least 85 percent of cases.
- In hereditary CJD, the person has a family history of the disease and/or tests positive for a genetic mutation associated with CJD.
- In acquired CJD, the disease is transmitted by exposure to brain or nervous system tissue, usually through certain medical procedures.

There is no evidence that CJD is contagious through casual contact with a CJD patient. Since CJD was first described in 1920, fewer than 1 percent of cases have been acquired CJD. About 5 to 10 percent of all CJD cases are inherited. These cases arise from a mutation, or change, in the gene that controls formation of the normal prion protein. While prions themselves do not contain genetic information and do not require genes to reproduce themselves, infectious prions can arise if a mutation occurs in the gene for the body's normal prion protein. If the prion protein gene is altered in a person's sperm or egg cells, the mutation can be transmitted to the person's offspring. All mutations in the prion protein gene are inherited as dominant traits. Therefore, family history is helpful in considering the diagnosis. Several different mutations in the prion gene have been identified. However, not all people with mutations in the prion protein gene develop CJD.