Michigan Department Of Community Health

MDCH

Creutzfeldt-Jakob Disease (CJD) Fact Sheet for Families

What is Creutzfeldt-Jakob disease?

CJD is a type of prion disease. Prion diseases are rare, progressive, and fatal diseases of the nervous system that can affect both humans and animals. Prion disease results from the production of an abnormal form of a protein found in the brain. This abnormal prion protein self-replicates and accumulates in the brain causing illness. Physicians first recognized and described CJD in the 1920's. CJD is the most common type of human prion disease.

What are the different types of CJD?

Sporadic (or Classical) CJD- accounts for approximately 85-90% of CJD cases. The disease results from the spontaneous change of the normal brain prion protein into the disease-causing abnormal form of prion protein. This abnormal form of protein can not be broken down and removed by the body.

Familial CJD- is an inherited form of CJD caused by a genetic mutation and accounts for 5-15% of cases. This gene mutation makes it more likely that the prion protein will convert to the abnormal disease-causing form.

Iatrogenic CJD- is transmitted by direct exposure to abnormal prion proteins from an outside source during a medical procedure. In rare situations, CJD has been spread by the re-use of contaminated surgical instruments or the transplantation of certain high-risk tissues from a CJD-infected donor.

Variant CJD (vCJD)- is the human form of bovine spongiform encephalopathy (BSE) or "Mad Cow Disease." Variant CJD is linked to the consumption of meat from an animal infected with BSE or by blood transfusion from a donor infected with variant CJD. Variant CJD is a separate disease and is caused by a different prion than the sporadic or familial forms of CJD.

Additional types of human prion diseases that are not classified as CJD

Gerstmann-Straussler-Scheinker disease (GSS)- is an extremely rare prion disease. It is almost always inherited and is found in only a few families around the world. Onset of the disease generally occurs between the ages of 35 and 55.

Fatal Familial Insomnia (FFI)- is an extremely rare prion disease that affects the thalamus and interferes with the ability to sleep. The disease can occur spontaneously or as a result of an inherited genetic mutation. Onset of the disease generally occurs between the ages of 40 and 60.

Are sporadic CJD and variant CJD (vCJD) the same?

No, variant CJD is the human form of BSE or "mad cow disease" and is caused by a different prion protein from the sporadic or familial forms of CJD. Individuals diagnosed with vCJD are generally younger, <55 years old, have a longer course of illness, and a different progression of symptoms than sporadic cases. Beyond the differences in clinical signs and symptoms, the examination of brain tissue allows for differentiation between variant CJD and other forms of human prion disease. To date, three cases of variant CJD have been diagnosed in the United States. All of these cases were foreign-born and grew up outside of the U.S. To date, no cases of variant CJD acquired in the U.S. have been found.

What are the symptoms of CJD?

CJD has a very long incubation (the time from infection to the onset of symptoms) ranging from 15 months to 30 years. Initial symptoms of CJD generally include progressive dementia, behavioral changes, and muscle incoordination. Other symptoms may include depression, insomnia, and problems with vision and speech. Once clinical symptoms begin, the progression of the disease is rapid; nearly all individuals diagnosed with CJD will succumb to the disease within one year.

How many cases of sporadic CJD are seen in the US?

Current surveillance suggests that one to two cases of CJD occur per million people per year. However, the risk of CJD increases with age; the rate of disease is 3.4 cases per million people in individuals over the age of 50. According to recent surveillance data, less than 300 cases of CJD are reported per year in the U.S.

How is CJD diagnosed?

The use of computed tomography (CT) scans, magnetic resonance imaging (MRI), electroencephalogram (EEG), specific tests for the "14-3-3" and "tau" proteins in cerebrospinal fluid, and clinical signs can help provide evidence for the diagnosis of CJD. At this time, a confirmed diagnosis of CJD can only be made by a brain autopsy or sometimes by a brain biopsy. Direct examination of brain tissue is needed in order to determine the presence and type of prion disease.

Where can I find prion disease testing for a family member?

Prion disease testing including 14-3-3 and tau protein testing, genetic testing, brain biopsy and autopsy services are available through the National Prion Disease Pathology Surveillance Center (NPDPSC) at Case Western Reserve University. The NPDPSC works in collaboration with the Centers for Disease Control and Prevention (CDC). This service is free for families of patients with suspected prion disease. For help with the coordination of an autopsy or other testing protocols, family members can directly contact the NPDPSC at (216) 368-0587. Information can also be found on the center's website http://www.cjdsurveillance.com. The NPDPSC can work with the attending physician to arrange testing.

Are there tests for familial forms of prion disease?

Yes, genetic blood tests are available to determine whether a person carries the genes for certain inheritable prion diseases. Familial prion diseases are inherited in an autosomal dominant fashion, meaning if one parent has the genetic mutation there is a 50% chance that each child will have the gene mutation. Only one copy of the mutated gene is needed to cause disease. Carrying the gene mutation, however, does not mean the person will definitely develop prion disease, only that it is more likey the person's body will produce the abnormal prion protein that causes disease. It is strongly recommended that a genetic counselor be consulted when deciding whether to proceed with testing.

How long will it take to get test results?

14-3-3 and tau protein testing on cerebrospinal fluid (CSF) can generally be completed within a few weeks. Confirmatory testing on tissue from a brain biopsy or autopsy results will be sent to the ordering physician in approximately 5-6 weeks. Genetic blood testing can take up to 12 weeks to complete.

Will an autopsy change funeral arrangements?

Because only a brain autopsy is needed to diagnosis CJD or other prion disease, it should not change or delay funeral arrangements. Embalming is still possible after an autopsy and a viewing can take place if that is what is desired by the family. The CDC does recommend that if a body has been autopsied that the family avoid superficial contact, such as touching or kissing the patient's face, for safety. There are no special burial or cremation requirements for patients with CJD.

Does Chronic Wasting Disease (CWD) cause disease in humans?

CWD is fatal prion disease found in cervids (deer, elk and moose). To date, CWD is not known to cause or be associated with disease in humans. No increase in human prion disease has been observed in areas of the western United States where CWD has been found in cervid populations for decades. However, because much is still unknown about prion diseases, the Centers for Disease Control and Prevention and the World Health Organization advise that humans do not consume animals that are known to be infected with CWD. In general, people should not handle or consume wild animals that appear sick or act abnormally, regardless of the cause. The risk of acquiring a prion disease from an animal source is extremely remote. Research suggests that the biological differences between humans and animals prevent animal prion diseases from being easily transmitted to people. Scrapie is an animal prion disease in sheep that has been recognized since the 18th century and has never been shown to be transmitted to or cause disease in humans.

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