CREUTZFIELD – JACOB DISEASE (CJD)

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Introduction

Creutzfeldt-Jakob Disease (CJD) causes a type of dementia that progresses unusually fast. More common causes of dementia, such as Alzheimer's dementia with Lewy bodies and frontotemporal dementia, typically progress more slowly.

Prion diseases, such as Creutzfeldt-Jakob disease, occur when prion protein, which is found throughout the body but whose normal function isn't yet known, begins folding into an abnormal three-dimensional shape. This shape change gradually triggers prion protein in the brain to fold into the same abnormal shape.

Through a process scientists don't yet understand; misfolded prion protein destroys brain cells. Resulting damage leads to rapid decline in thinking and reasoning as well as involuntary muscle movements, confusion, difficulty walking and mood changes. CJD is rare, occurring in about one in 1 million people annually worldwide.

Experts generally recognise the following main types of CJD:

- **Sporadic CJD** develops spontaneously for no known reason. It accounts for 85 percent of cases. On average, sporadic CJD first appears between ages 60 and 65.
- **Familial CJD** is a heredity form caused by certain changes in the prion protein gene. These genetic changes are "dominant," meaning that anyone who inherits a CJD gene from an affected parent will also develop the disorder. Familial CJD accounts for about 10 to 15 percent of cases.
- **Acquired CJD** results from exposure to an external source of abnormal prion protein. These sources estimated to account for about 1 percent of CJD cases.

Diagnosis

Rapid symptom progression is one of the most important clues that a person may have Creutzfeldt-Jakob disease. There is no single test — or any combination of tests that can conclusively diagnose sporadic CJD in a living person, however the following tests may help determine whether an individual has CJD:

- **Electroencephalogram (EEG)** measures the brain's patterns of electrical activity similar to the way an electrocardiogram (ECG) measures the heart's electrical activity.
- **Brain magnetic resonance imaging (MRI)** can detect certain brain changes consistent with CJD.
- **Lumbar puncture (spinal tap)** tests spinal fluid for the presence of certain proteins.

Sporadic Creutzfeldt-Jakob disease has no known cause. Most scientists believe the disease begins when prion protein somewhere in the brain spontaneously misfolds, triggering a "domino effect" that misfolds prion protein throughout the brain. Genetic variation in the prion protein gene may affect risk of this spontaneous misfolding.

Mutations in the prion protein gene also may play a yet-to-be-determined role in making people susceptible to acquired CJD from external sources. Scientists don't yet know why acquired CJD seems to be transmitted through such a limited number of external sources. Researchers have found no evidence that the abnormal protein is commonly transmitted through sexual activity or blood transfusions.

Familial CJD is caused by variations in the prion protein gene that guarantee an individual will develop CJD. Researchers have identified more than 50 prion protein mutations in those with

inherited CJD. Genetic testing can determine whether family members at risk have inherited a CJD-causing mutation. Experts strongly recommend professional genetic counselling both before and after genetic testing for hereditary CJD.

Procedure

1. **Symptoms**

Specific Creutzfeldt-Jakob disease symptoms experienced by an individual and the order in which they appear can differ significantly. Symptoms include:

- depression
- agitation, apathy and mood swings
- rapidly worsening confusion, disorientation, and problems with memory, thinking, planning and judgment
- ataxia and difficulty walking
- muscle stiffness, twitches and involuntary jerky movements
- bowel incontinence
- dysphagia
- loss of vision
- pyrexia due to undiagnosed infections or the effect of the disease on the central nervous system

2. **Treatment and Outcome**

There is no treatment that can slow or stop the underlying brain cell destruction caused by Creutzfeldt-Jakob disease and other prion diseases. Various drugs have been tested but have not shown any benefit. Clinical studies of potential CJD treatments are complicated by the rarity of the disease and its rapid progression.

Current therapies focus on treating symptoms and on supporting individuals and families coping with CJD such as analgesia to treat pain and muscle relaxant or anti-seizure medication to treat muscle stiffness or muscle twitching. In the later stages of the disease, individuals with CJD become completely dependent on others for their daily needs and comfort.

CJD progresses rapidly. Those affected lose their ability to move or speak and require full-time care to meet their daily needs. An estimated 90 percent of those diagnosed with sporadic CJD die within one year. Those affected by familial CJD tend to develop the disorder at an earlier age and survive somewhat longer than those with the sporadic form, as do those diagnosed with CJD. Scientists have not yet learned the reason for these differences in survival.

3. **Advance Directive**

Many people with CJD draw up an advance directive (also known as an advance decision) and our organisation will support the individual and family to get this in place if they so wish.

An advance directive is where a person makes their treatment preferences known in advance, in case they can't communicate their decisions later because they're too ill. Issues that can be covered by an advance directive include:

- whether a person with CJD wants to be treated at home, in a hospice or in a hospital once they reach the final stages of the condition
- what type of medications they would be willing to take in certain circumstances
- whether they would be willing to use a feeding tube if they were no longer able to swallow food and liquid
- whether they're willing to donate any of their organs for research after they die (the brains of people with CJD are particularly important for ongoing research)
- if they have respiratory failure (loss of lung function), whether they would be willing to be resuscitated by artificial means – for example, by having a breathing tube inserted into their neck
- Normal social and clinical contact, and non-invasive clinical investigations (e.g. x-ray imaging procedures) with CJD individuals do not present a risk to healthcare workers, relatives, or the community. There is no reason to defer, deny, or in any way discourage

the admission of a person with a CJD into any healthcare setting. Based on current knowledge, isolation of individuals is not necessary; they can be nursed using Standard Precautions. As the disease is usually rapidly progressive, the individual will develop high dependency needs and require ongoing assessment.

- It is essential to address the physical, nutritional, psychological, educational, and social needs of the individual and the associated needs of his or her family. Co-ordinated planning is vital in transferring care from one environment to another.

4. Prevention

- Sterilisation methods used to help prevent bacteria and viruses spreading are also not completely effective against the infectious protein (prion) that causes CJD. However, tightened guidelines on the reuse of surgical equipment mean that cases of CJD spread through medical treatment (iatrogenic CJD) are now very rare.
- There are also measures in place to prevent variant CJD spreading through the food supply or via the supply of blood used for blood transfusions.
- To reduce the already very low risk of CJD transmission from one person to another, people should never donate blood, tissues, or organs if they have suspected or confirmed CJD, or if they are at increased risk because of a family history of the disease, a dura mater graft, or other factor.

5. Preventing cross contamination

Follow universal infection prevention control procedures as contamination by body fluids poses no greater hazard than for any other person:

- Cover cuts and abrasions with waterproof dressings.
- Wear surgical gloves when handling individual's tissues and fluids or dressing the individual's wounds.
- Avoid cutting or sticking with instruments contaminated by the individual's blood or other tissues.
- Use face protection if there is a risk of splashing contaminated material such as blood or cerebrospinal fluid.
- No special precautions are required for feeding utensils, feeding tubes, suction tubes, bedding material or pressure ulcer care, follow standard infection control procedures
- Clinical waste to be disposed of following organisational procedures

6. Post-exposure management

No case of human CJD is known to have occurred through occupational accident or injury. A number of strategies to minimise the theoretical risk of infection following accidents have been proposed, but their usefulness is untested and unknown. For the present the following actions are recommended and medical advice is sought.

- Contamination of unbroken skin with internal body fluids or tissues: wash with detergent and abundant quantities of warm water (avoid scrubbing), rinse, and dry. Brief exposure (l minute, to 0.1N NaOH or a 1: 10 dilution of bleach) can be considered for maximum safety.
- Needle sticks or lacerations: gently encourage bleeding; wash (avoid scrubbing) with warm soapy water, rinse, dry and cover with a waterproof dressing. Further treatment (e.g., sutures) should be appropriate to the type of injury. Report the injury according to organisational procedures.
- Splashes into the eye or mouth: irrigate with either saline (eye) or tap water (mouth); report according to organisational procedures
- Health and safety guidelines mandate reporting of injuries and records should be kept for no less than 20 years.
- The organisational Infection Control, Waste Management and Decontamination policies should be read in conjunction with this procedure
- **The GP or medical professional must notify the National CJD Research & Surveillance Unit (NCJDRSU) at Edinburgh University** of any new CJD diagnoses

Further support and information

- *Creutzfeldt-Jakob Disease Foundation*
- *The Alzheimer's Association*
- *Social Security Administration (SSA) has a compassionate allowance programme*
- *Medline Plus: Creutzfeldt-Jakob disease*