BEAUMONT HOSPITAL

Frequently Asked Questions Creutzfeldt-Jakob disease (CJD)

INCJDSU
Irish National Creutzfeldt-Jacob Disease Surveillance Unit

What Is CJD?

Creutzfeldt - Jakob disease (CJD) is a rare illness and is one of a group of diseases called prion diseases, which affect humans and animals. Prion diseases exist in different forms, all of which are progressive, currently untreatable and ultimately fatal.

They are caused by the presence of an abnormal protein in the brain tissue, called prion protein (PrP^{Sc}), and are believed to result from a change in the shape, of a normal cellular protein (PrP^{C}) which is present in the brain. As the amount of abnormal prion protein grows, it becomes hard to break down, causing brain degeneration and neurologic disease.

Prion Diseases are extremely rare reportedly affecting one to two people per million of the population each year worldwide; equating to ~ 4-10 cases annually in Ireland.

How did my loved one get it?

This depends on the type of CJD. Acquired types of CJD, such as the highly publicised Variant CJD (vCJD), are incredibly rare. Variant CJD resulted from eating meat from diseased cattle. There have been a total of 229 cases of vCJD identified internationally since it was first reported in 1996; with no new cases since 2013. Genetic CJD or inherited forms of Prion Disease exist which can be passed on from generation to generation.

However, the most prevalent human prion disease is Sporadic CJD, which accounts for ~85% of all those identified. The cause of Sporadic CJD is not known, but it is thought to be due to spontaneous or “chance” misfolding of the normal prion protein (PrPc) which is present in all of us.

Is there any treatment or cure for CJD?

At present, there is no cure for CJD; the disease is ultimately fatal. Treatments for CJD patients focus on easing their symptoms and discomfort; including drugs to control distress, myoclonus and hallucinations, catheters to collect urine, intravenous fluids and frequent repositioning of the patient to avoid bedsores.

Is it contagious? Can you catch CJD from someone?

Prion diseases are not infectious in the usual way. For example, they are not spread by airborne droplets like colds and flu, or by body fluids or sexual contact like HIV. The overall evidence suggests that there is no increased risk of developing CJD from contact with a person suffering from the condition. Special precautions are not required by anyone coming into contact with a CJD patient, however, it is sensible for anyone who might be exposed to the blood of a CJD patient to wear gloves and take standard precautions.

Am I at increased risk of contracting CJD by caring for my loved one?

No, multiple studies have demonstrated that there is no evidence that healthcare professionals or caregivers are at any increased risk of prion disease compared to the general population.

How can we be sure that the clinical diagnosis of CJD is correct?

Each individual case of CJD can be assigned to one of three forms: sporadic, genetic, or acquired (variant or iatrogenic). The diagnostic methods may vary depending on the type.

In sporadic CJD, the spinal fluid test (RT-QuIC) has improved the diagnostic accuracy while the patient is alive (sensitivity 92% and specificity 100%), and it is now included as one of the diagnostic criteria along with the electroencephalogram (EEG) and MRI. MRI images, when reviewed by a neuroradiologist trained to detect CJD, can also be very accurate indicators of CJD.

In genetic CJD, the diagnosis depends on development of particular neurological symptoms and the identification of a PrP gene (*PRNP*) mutation by genetic analysis.

In acquired CJD, iatrogenic CJD is diagnosed on the basis of symptoms developing in someone with a relevant exposure. In vCJD, diagnosis is very difficult while the patient is alive, unless the diagnosis of vCJD is suspected and tonsil biopsy is carried out. An MRI scan may prove to be useful; however, a definitive diagnosis depends on examination of brain tissue or lymphoreticular tissue such as the tonsils.

Ultimately, examination of brain tissue is the only known method of detecting CJD with 100% certainty. While brain biopsy is an option, it is not recommended for CJD patients unless used to rule out other treatable conditions. Analysis of the brain at post mortem is recommended to confirm any prion disease diagnosis.

Is the CJD patient in pain?

Clinical observation of people in the later stages of CJD indicates that they lose awareness of their condition as the disease progresses. Obviously this saves them, but not their families, from suffering. However, in the early stages, patients with CJD can develop marked fear which can be very distressing and is probably associated with visual hallucinations. They may feel discomfort, and some of the symptoms of the disease such as myoclonus are distressing to caregivers. Neurologists believe there is no pain associated with the disease itself. For example, there is no rise in pressure in the head which could cause headache or any other obvious cause of pain.

How long do you live with CJD?

Again this is dependent of the type of CJD; most people die within 4 months to 2 years.

Is an autopsy necessary and what should we expect?

CJD is a notifiable disease in Ireland and all suspected CJD cases must be notified to the coroner at the time of death. Referral of a death to the coroner is independent of the family's wishes and family permission is not required to carry out a post mortem.

However, it is important to remember that an autopsy helps the family to know the definite cause of death. Furthermore, autopsy examination to confirm the diagnosis of CJD helps protect public health.

Please download the INCJDSU CJD Autopsy Information Booklet should you require further information.