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CJD and Prion Disease

Charity no. 1097173

Prion diseases are a group of rare and fatal brain disorders that occur in humans and some animals. While they have been recognized for hundreds of years, they first came to the attention of the wider public in the 1980 with the appearance of BSE* (Bovine spongiform encephalopathy) in UK cattle and subsequently in 1996 with the identification of human variant CJD.

These prion diseases cause the brain to look sponge-like when looked at under a microscope, and for that reason they have also been called 'spongiform encephalopathies' (as an 'encephalopathy' in medical terms means 'disease of the brain').

Human prion diseases can be divided into three categories – sporadic (of uncertain cause), genetic (inherited) and acquired (transmitted as infections).

The table below lists the main recognised prion diseases in humans.

Human Prion Disease	
Genetic	Genetic CJD
	GSS (Gerstmann-Sträussler-Schenker Disease)
	FFI (Fatal Familal Insomnia)
Sporadic	Sporadic CJD
	VPSPr (Variably Protease Sensitive Prionopathy)
Acquired	Variant CJD (originally due to BSE)
	Iatrogenic CJD

The Prion Protein

Proteins are important molecules which have structural and functional roles in the body. There are many different types of proteins, and they are all coded for by genes specific to them. The genes act as instructions which cells use to make the various proteins. Proteins are constantly being made by cells, serving their purpose, then being broken down.

One such protein is the prion protein. This is present in everyone, although the precise function of this protein is not yet known. The gene that corresponds to the prion protein is called the Prion Protein Gene (PRNP).

Prion disease is caused by the prion protein being assembled incorrectly by the cells, causing it to work abnormally. The abnormal form of the protein is much more difficult to break down than the normal form, meaning that it builds up in body tissues. Another crucial difference is that this incorrect form can convert the normal form into a copy of itself, leading to even more of a build-up of the abnormal form of the protein.

*In the past, some media have used the term 'mad cow disease', an unfortunate name that is offensive to many affected families and the CJDSN feels it is not a term that should remain in use,

So how does the abnormal form get created in the first place? There are different ways, depending on the form of prion disease:

1. An abnormality of the gene causes it to instruct the cell incorrectly, as in genetic prion disease.

2. The abnormal form of the protein is introduced through the diet or medical procedures, as in acquired prion disease.

3. A random error occurs in the making of the protein, causing an abnormal protein to be made. This is the likely explanation for sporadic prion disease, although that has not been conclusively proven.

The disease process

Prion diseases affect the central nervous system, which is made up of the brain and the spinal cord. They cause loss of brain cells, called neurons. These neurons connect to each other and create and transmit messages throughout the body, such as messages telling our arms to move, or our skin to feel. They are responsible for all of the functions carried out by the brain. This means that all of these functions can be affected by prion disease, and the disease can look different in different people. Some common features are:

- Impairment of memory, thinking, speech and behaviour
- Problems with balance and co-ordination
- Involuntary jerking movements

At the moment, how the abnormal protein building up in brain tissue is related to brain cells being destroyed is not known. The disease is progressive, often rapidly, and sadly there is no treatment or cure, meaning that they are invariably fatal.

Diagnosis

One way to diagnose prion disease is to take a biopsy of the brain and examine it under a microscope. This is an invasive test and in most cases is not done. A sample may be taken post-mortem to confirm the suspected diagnosis.

A clinical diagnosis in life is usually possible and typically with a high degree of confidence. A clinical diagnosis is based on the symptoms, the way the illness progresses, the findings on examination of the patient and the results of various tests. Because the presenting symptoms are similar to those seen in other disorders of the brain, several other possible diagnoses may need to be considered before getting to the correct one. There are tests which help to reach this diagnosis, and to rule out other possibilities. These include MRI scans of the brain and taking samples of spinal fluid for analysis. These tests can take time, which due to the rapid progression of prion disease often leads to a fairly late diagnosis. It is important to do these tests, as treatable causes of brain dysfunction need to be ruled out.

I'd like more information... If you would like to know more about CJD, the CJD Support Network would be happy to hear from you. Contact support@cjdsupport.co.uk or call 0800 774 7317 (8am-6pm, Tues and Fri).