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An Unusual Case of Variant CJD

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A Case Report published in this week's The Lancet , written by Professor John Collinge, MRC Prion Unit and National Prion Clinic, UCL Institute of Neurology and National Hospital for Neurology and Neurosurgery, London, reports the particular genetic make-up of a 30-year old man who has died of variant Creutzfeldt–Jakob disease (vCJD). The case report suggests that there could be other people with the condition who at the moment have no symptoms.

vCJD is caused by infectious agents called prions, which are made primarily of protein. The prions which cause vCJD are the same as those that cause bovine spongiform encephalopathy (BSE, also known as mad cow disease) in cows. Prion diseases affect the structure of the brain or other neural tissue, and all are currently untreatable and eventually fatal. Disease-causing prions are thought to consist of abnormally folded proteins that spread by encouraging the normal healthy prion protein found on the surface of most cells in the body to change shape. Prion diseases share similar disease mechanisms with Alzheimer's, Parkinson's, and other neurodegenerative brain diseases.

The 30-year-old man was admitted to hospital in June, 2008, with a 13-month history of personality change, progressive unsteadiness, and intellectual decline. He complained of severe leg pain and poor memory. Two months later he developed visual hallucinations. His symptoms worsened over the next three months. An MRI scan and other tests led to a diagnosis of vCJD. The man died in January 2009.

The case is unusual because tests showed the man had a particular genotype at his human prion protein gene (*PRNP* 129 codon), which can code for the amino acids valine (V) or methionine (M). People can be VV (homozygous), MM (again homozygous), or MV (heterozygous). Since 1994, around 200 cases of vCJD have been identified worldwide, and all those tested have been MM homozygous. However, the man in this Case Report was heterozygous.

Other prion diseases such as kuru or CJD associated with the use of pituitary hormones tend to have longer incubation periods in people who are *PRNP* heterozygous than those who are MM homozygous. The authors have recently reported some heterozygous patients with kuru had been incubating the disease over 50 years. Thus the authors believe there could be other cases like this one in which people are infected with vCJD but experiencing a long incubation period.

The authors say:

> *"The majority of the UK population have potentially been exposed to BSE prions but the extent of clinically silent infection remains unclear. About a third of the UK population are PRNP codon 129 methionine homozygous. If individuals with other genotypes are similarly susceptible to developing prion disease after BSE prion exposure, but with longer incubation periods, further cases, which may or may not meet diagnostic criteria for vCJD, would be expected in these PRNP codon 129 genotypes."*

They conclude:

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"However, prion disease susceptibility and incubation periods are also affected by other genetic loci, and the possibility remains that cases of vCJD to date may have unusual combinations of genotypes at these loci, yet to be fully characterised."

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