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More people could have vCJD than previously thought

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More people may be incubating variant CJD, the human version of so-called "mad cow disease", than was previously thought, according to scientists who today report an unusual case of the disease. All those tested worldwide since 1994 when the first cases were identified have been MM homozygous.

However, a 30-year-old man who died of vCJD in January this year was found to have a different genetic makeup from the rest of the 200 or so people diagnosed around the world. Six months before the man was diagnosed with the disease, he had been admitted to hospital with personality changes, unsteadiness in walking that became progressively worse and intellectual decline. He told doctors he had severe leg pain and memory problems. Two months later, he developed visual hallucinations. The symptoms got progressively worse and an MRI scan confirmed vCJD. The symptoms and the course of the illness were not unusual for vCJD, but the man had a different genetic makeup from the rest of the 200 or so people diagnosed around the world to date.

Variant CJD is caused by prions, infectious agents which are made up mainly of proteins. The same prions cause vCJD and also BSE - bovine spongiform encephalopathy - which was dubbed "mad cow disease" because cattle who contracted it staggered when they tried to walk. Prion diseases affect the structure of the brain or other neural tissue and are currently untreatable and fatal.

Doctors from the MRC Prion Unit and National Prion Clinic at the UCL Institute of Neurology and National Hospital for Neurology and Neurosurgery in London, report the unusual case in today's Lancet medical journal. Tests showed that the man had a particular form of the human prion protein gene. All those tested world-wide since 1994 when the first cases were identified have been MM homozygous. However, this patient was MV heterozygous.

The observation could be of concern. In some other human prion diseases, such as kuru - thought to be linked to cannibalism in Papua New Guinea - people who are MV heterozygous have incubated the disease for longer than those who are MM homozygous before symptoms have shown. Some MV heterozygous patients are reported to have incubated kuru for over 50 years.

It is possible, doctors say, that vCJD takes longer to develop in people who are MV heterozygous than in MV homozygous people.

"The majority of the UK population have potentially been exposed to BSE prions but the extent of clinically silent infection remains unclear," say the authors of the paper. About a third of the population have the MM homozygous genotype - and until now all the cases came from this group. If individuals with other genotypes are similarly susceptible to developing prion disease after exposure to BSE, further cases would be expected, they

say. However, they add, it is possible that susceptibility to vCJD and incubation period may be influenced by other genetic factors which have not yet been identified.