CJD victim 'had different gene'

A 30-year-old man thought to have died in January from vCJD belonged to a genetic group that had not shown any signs of the disease, scientists say.

In the UK, 166 people have died of variant CJD, linked to eating BSE-infected beef, and all were thought to have shared a certain gene.

Writing in the Lancet, the scientists say Grant Goodwin, of Lanarkshire, had a different version of the gene.

They estimate that up to 350 people in this group could get vCJD.

Scientists have always thought that a second wave of vCJD cases would emerge some time after the first.

This is the first indication that this theory is being borne out with the identification of the first probable vCJD patient outside of the initial genetic group, BBC science correspondent Pallab Ghosh reports.

It is probable because the diagnosis is based on observations of the progression of the disease rather than post-mortem tests which would have provided absolute confirmation of the disease, he adds.

The case report written by Professor John Collinge, of the National Prion Clinic, and colleagues is a reminder that the disease has not gone away.

Many thousands of people may be carrying the infection and although they will never show any symptoms, they have the potential to infect others.

> " **The majority of the UK population have potentially been exposed to BSE prions but the extent of clinically silent infection remains unclear** "
> Professor John Collinge, National Prion Clinic

vCJD is caused by infectious agents called prions.

Prion diseases affect the structure of the brain or other neural tissue and are currently untreatable.

Disease-causing prions are thought to consist of abnormally folded proteins, which spread by encouraging the normal healthy prion protein found on the surface of most cells in the body to change shape.

Tests showed that Mr Goodwin had a heterozygous version of the gene which codes for the human prion amino acids valine (V) or methionine (M).

'Incubation periods'

People can be V V (homozygous), M M (homozygous) or M V (heterozygous).

Since 1994, around 200 cases of vCJD have been identified worldwide, and all those tested have been M M homozygous.

However, Mr Goodwin was M V heterozygous.

It is thought that 47% of the population have this version of the gene.

Professor Collinge said: "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 M M homozygous.

"If individuals with other genotypes are similarly susceptible to developing prion disease after BSE prion exposure, but with longer incubation periods, further cases would be expected."

The scientists have previously looked at another prion disease in New Guinea, called kuru, which is induced by eating infected human tissues.

The original cases were all M M but more recently M V have appeared.

They say this indicates that M V people can get prion diseases like kuru but have a much longer incubation period.

A Department of Health spokesperson said: ""The Spongiform Encephalopathy Advisory Committee (SEAC) have noted this finding, which confirms the need for ongoing vigilance and robust surveillance of CJD.

"We are continuing to provide resources for CJD surveillance and research, and the development of a test for vCJD remains a priority."

Story from BBC NEWS:
http://news.bbc.co.uk/go/pr/fr/-/2/hi/health/8419459.stm

Published: 2009/12/18 00:37:34 GMT