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Genetic brain disease discovered

Scientists have traced a new neurodegenerative disease

By BBC News Online's Jane Elliott

Scientists have discovered a new neurodegenerative disease, which has lain undiscovered among the branches of one Cumbrian family for over 200 years.

For generations the family, which has not been named, thought they had an inherited risk of either Parkinson's or Huntington's disease as their limb movements became harder to control.

One family member even killed herself as her body started to show symptoms of what she thought was the potentially fatal Huntington's.

It was her death and subsequent post-mortem that gave Newcastle scientists the break-through they desperately needed.

Iron build-up

"You don't get dementia, but I don't know whether that is a blessing or a curse

Professor John Burn

Professor John Burn and his team from the Institute of Human Genetics at the University of Newcastle, had been studying the West Cumbrian family for over 15 years.

And they were mystified by the symptoms, which so clearly mimicked either Parkinson's or Huntington's.

Professor Burn said: "I became convinced that it looked like Huntington's, but it wasn't.

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"One woman had been suffering from this for 30 years, and although her movements were bad she could still beat the rest of the family at the TV quiz show Countdown.

"You don't get dementia, but I don't know whether that is a blessing or a curse."

Now hopes are high that the discovery of "Neuorferritinopathy", which is caused by a build-up of iron in the brain, can shed new light on the causes of Parkinson's and Huntington's.

It may in the future lead the way to finding a cure for both diseases.

Prof Burn said that although the disease has so far been confined to members of the same family, a screening test of 100 patients with similar brain disorder revealed another six linked families.

Close monitoring

So far all the cases have been based in the North of England, but as the affected family are linked to Fletcher Christian, leader of the Mutiny on the Bounty, the disease could well have spread via the sea-faring West Cumbrian family to other more remote parts of the globe, where it is still being wrongly diagnosed.

The patients had limb movement problems, but showed a remarkable brain clarity and none of the degeneration associated with Huntington's and Parkinson's.

By studying the family, Prof Burn's team found an error in the ferritin light chain.

Ferritin stores essential iron inside cells and prevents the iron causing damage.

The error leads to large amounts of iron and ferritin collecting in the cells and causing problems with body movements.

Prof Burn said: "What was happening in the family was that the ferritin was not working properly and the cells in the basal ganglia were filling up.

"The iron was building up in the brain like a pile of bin-liners when the bin man hasn't come to collect them. And the build up eventually stops the cells from working.

"That leads to movement problems."

In the past it has always been difficult to say whether iron levels have a significant impact on neurological diseases, because the levels do tend to increase with age.

Prof Burn said: "We are now looking at the exact cause of the build up of iron and to see if we can remove it.

"The findings in this rare family make it more likely that problems with iron metabolism can cause similar disease like Parkinson's disease."

The study was published in Nature Genetics.

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