BBC

Family help Cardiff Uni find motor neurone disease gene

A Gwent family with a history of motor neurone disease (MND) has helped scientists from Cardiff University find a "new gene" linked to the disease.

An international scientific team studied the family and a large group of patients in Finland.

They discovered a common gene which will now lead to new blood tests for families with a history of the disease.

One of the Gwent family, which has lost eight people to MND, said they felt "relieved" the gene had been found.

The findings of the international team, which also included scientists from Manchester University and University College London, have been published in the Neuron journal.

Funded by the Motor Neurone Disease Association, the research was conducted by taking blood and skin samples and conducting post-mortem examinations.

**Genetic link**

Researchers found that the Welsh family and patients in Finland shared "a changed genetic segment on the short arm of chromosome 9".

The family, who did not want to be identified, traced their link to the disease back to the 1940s.

Over the years they have lost many relatives to early onset motor neurone disease and also to a neurodegenerative illness known as Pick's disease.

Motor neurone disease is currently incurable and fatal, with an average survival of between two and five years.

Dr Huw Morris, based at the Centre for Neuropsychiatric Genetics at Cardiff University and the Royal Gwent Hospital in Newport, has worked with the family for the past 10 years.

He said that for most patients with motor neurone disease (MND) there is no risk to other family members, but some families are affected by "familial MND".

Dr Morris said the research was aimed at "helping future generations".

**Breakthrough made**

"Six weeks ago we knew very little about the cause of this condition and now we know the primary cause of the disease within this family and similar families in other parts of the world," he said.

"To our surprise this is very common in familial MND so that a third of families around the world where many people have been affected carry this gene change."

He added the breakthrough was "the end of our long hunt for this gene", but also marked "the beginning of our search for therapies based on this discovery that can stop this brutal disease in its tracks".

A member of the family said they "have been waiting for years and years for this to be found".

"I can't really describe how I felt when I heard that the cause of motor neuron disease in our family had been discovered," he said.

"I was thrown at first, then I felt relieved that it had been found.

"I appreciate all of the hard work and support that we have had from Dr Morris and the other doctors and scientists involved with this."