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Researchers find genetic key to preventing spine tumours

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Genetic medicine experts from Manchester Biomedical Research Centre at Saint Mary's Hospital and The University of Manchester have identified a new gene responsible for causing an inherited form of tumour, known as spinal meningioma.

Meningiomas are the commonest form of tumour affecting the brain and spine. Usually meningiomas can be removed by surgery and do not recur. Occasionally people can develop more than one meningioma or many members of the same family can be affected.

A team led by Dr Miriam Smith, Professor Gareth Evans and Dr Bill Newman worked with families with a history of meningiomas affecting the spinal cord. Using a powerful new genetic sequencing technique called next generation sequencing, they were able to check all the genes of three individuals with multiple spinal meningiomas. This lead to the identification that changes in a gene called SMARCE1 lead to spinal meningiomas in some families.

In December 2012 the government announced a focus on genetic sequencing with an aim of sequencing the genomes (a person's DNA) of 100,000 Britons with cancer and rare diseases in UK centres. The voluntary sequencing of patients will lead to better testing, better drugs and above all better care for patients. Manchester is already using this technology in their well established Genetics department at Saint Mary's and it is enabling doctors to ensure patients have access to the right drugs and personalised care quicker than ever before.

In the past year 10 genes have been discovered using the new next generation sequencing technology in Manchester including genes for developmental problems, deafness, short stature and bladder problems that lead to kidney failure.

"With our new DNA sequencing machines, we have been able to show that changes in the SMARCE1 gene are responsible for multiple spinal meningioma disease," said Dr Smith. "Before our work, doctors did not know that inherited spinal meningiomas have a completely different cause to other tumours affecting the brain and spine.

"The next step is to develop a screening programme to assess the risk of developing spinal tumours for individuals in affected families, and to investigate possible treatments to prevent the spinal tumours from growing."

Professor Richard Marias, Director of Cancer Research UK's Paterson Institute at The University of Manchester, said "This research highlights the complexity of tumour diagnosis. Such detailed molecular characterisation underpins current thoughts about how meningioma and cancer will be managed in the future and is at the heart of the personalised medicine approach."
Just over two people in every 100,000 develop meningiomas in the head and spine, with twice as many women as men diagnosed with the condition.

The team's pioneering work was funded by The Children's Tumor Foundation, a US-based charity supporting neurofibromatosis research, and the Association for International Cancer Research, a global cancer research charity.

The research study findings have recently been published in the prestigious international journal *Nature Genetics*.

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More information: www.nature.com/doifinder/10.1038/ng.2552

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