

University of Manchester news release

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## New cause of child brain tumour condition identified

Doctors and scientists from The University of Manchester have identified changes in a gene, which can increase the risk of developing brain tumours in children with a rare inherited condition called Gorlin syndrome.

Gorlin syndrome causes an increased risk of developing cancers of the skin and, rarely, in the brain. Around 1 in 30,000 people has the condition.

Most people with Gorlin syndrome have a change in a gene called *PTCH1*, but the new research has revealed that changes in a gene called *SUFU* also cause Gorlin syndrome and it is children with a change in *SUFU* that are 20 times more likely to develop a brain tumour.

Dr Miriam Smith, a lecturer in cancer genomics from the University's Institute of Human Development led the research, which was also carried out with The Christie NHS Foundation Trust and the Royal Manchester Children's Hospital. She said: "We have essentially found a new cause of Gorlin syndrome, but one that results in the specific outcome of a childhood brain tumour called a medulloblastoma in small children."

The researchers at Manchester identified mutations in the *SUFU* gene as a cause of Gorlin syndrome in families with at least one person affected by a medulloblastoma. They compared the risk of developing a medulloblastoma for people with *SUFU*-related Gorlin syndrome to the risk for people with a *PTCH1* mutation and found that those with the *PTCH1* changes had around a 2% risk of developing the brain tumours, but in those with the *SUFU* changes it was around 33%.

The findings, published in the *Journal of Clinical Oncology*, have major implications for the way in which children with Gorlin syndrome are treated and how frequently they require brain scans. Currently all children with Gorlin syndrome regardless of whether the changed gene is *SUFU* or *PTCH1* are scanned once a year up to the age of eight.

The research was funded by the **British Skin Foundation**.

The paper, '[Germline mutations in SUFU cause Gorlin syndrome-associated childhood medulloblastoma and redefine the risk associated with PTCH1 mutations](#)' was published in the Journal of Clinical Oncology.

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