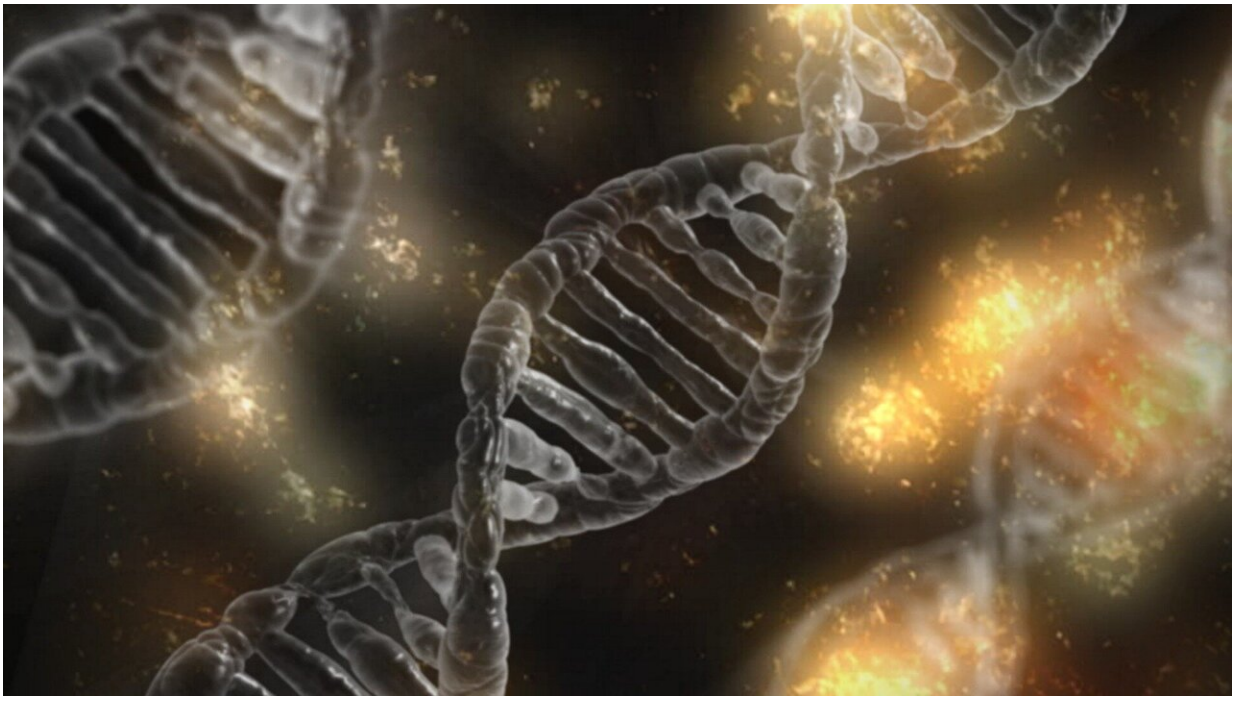
Researchers uncover new sarcoma gene

June 11 2019



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A team of WA researchers has uncovered new genes that could play an important role in the development of sarcoma, a group of rare bone, muscle and connective tissue cancers.

The study, undertaken by Ph.D. student Rachel Jones and led by Associate Professor Evan Ingley from the Harry Perkins Institute of Medical Research and Murdoch University, identified a new sarcoma

familial risk gene, which is inherited from parents and increases the risk of developing the potentially deadly disease.

Most cancers are caused by genetic errors in our cells which can be triggered spontaneously or by lifestyle or environmental factors, but in some cases cancer-causing genes can be passed down from our parents.

These familial genes are known as cancer susceptibility genes and are responsible for up to 10% of familial cancer risk, and while more than 100 of these genes have been identified, altogether they only account for a small percentage of familial cancer risk.

Associate Professor Ingley said the team used the International Sarcoma Kindred Study (ISKS), a cohort of families with sarcoma and other cancers, to sort through the catalogues of familial genes and identify any that could be associated with an increased risk of cancer.

"If we can identify more of these familial cancer risk genes, we will know more about which genes are involved in cancer development and this could assist us in identifying new ways of targeting cancer" Associate Professor Ingley said.

The study ultimately led to the team discovering two new genes not previously connected to sarcoma, one called ABCB5, which is associated with other cancers such as melanoma and leukaemia, and a previously undiscovered gene called C16orf96—named for its gene number (96) on chromosome 16, but with an unknown function.

"We are trying to find out what this new gene does and how it affects sarcoma risk, and then hopefully give it a more meaningful name."

In addition to this, the team is now planning to investigate how widespread the association of these two new sarcoma risk genes is in

other families with sarcoma and other cancers, and they are also looking at what are the functions of these two new genes in sarcoma.

"From these further studies we will know if these genes might be future candidates for developing targeted therapies."

"One very important thing we have learnt from this research is that although we know the DNA sequence identity of all of our genes, there are still many genes that we don't know their specific biological role or for what diseases they may be important. So there is still a lot we need to learn in terms of the function of our genes."

More information: Identification of novel sarcoma risk genes using a two-stage genome wide DNA sequencing strategy in cancer cluster families and population case and control cohorts, *BMC Medical Genetics* (2019). DOI: 10.1186/s12881-019-0808-9

Provided by Harry Perkins Institute of Medical Research