Researchers reveal genetic links underlying neuroendocrine pancreatic cancer

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Credit: Professor Sean Grimmond

An Australian-led international research effort has revealed that genetic changes normally linked to breast, colon and ovarian cancers could also drive a rare form of pancreatic cancer.

Researchers found as many as one in five patients with pancreatic neuroendocrine tumours (PanNETs) had a clear genetic predisposition for their cancers, even though they had no family history of the disease.

The findings, published today in the journal *Nature*, offer the prospect of one day being able to identify people at risk of these cancers, as well as aggressive forms of the disease, and who might respond to current or new targeted therapies.

The breakthrough is the work of researchers at the University of Melbourne, Garvan Institute of Medical Research, QIMR Berghofer Medical Research Institute, The University of Queensland and the Children's Medical Research Institute, as well as European and US teams.

They carried out whole genome sequencing of tumours from 100 Australian patients recruited through the Australian Pancreatic Cancer Genome Initiative (APGI).

Study co-leader Professor Sean Grimmond, the University of Melbourne's Bertalli Chair of Cancer Medicine, said researchers were surprised to find striking similarities between the genetic drivers of PanNETs and other cancers.

"We found that the MUTYH and BRCA2 gene mutations, normally associated with colon and breast cancers, also appear to play an important role in PanNETs," Professor Grimmond said.

"This raises exciting possibilities for how we treat this disease in the future,"

Study co-leader Dr Nicola Waddell, of QIMR Berghofer, said people without a family history of cancer could still carry a faulty gene that increases their risk of developing this tumour.

"The genetic variants we've identified may help to predict how aggressive each patient's tumour is and what sort of treatment they're

likely to benefit from," she said.

"In the future, patients at risk of this rare pancreatic cancer could be identifiable through genetic screening."

PanNETs account for about two per cent of the 3,000 cases of pancreatic cancer diagnosed in Australia each year.

Amber Johns of the APGI at the Garvan Institute said although patients often have a better prognosis than those with more common pancreatic cancers, this group of tumours is highly unpredictable.

"Doctors currently face the challenge of being unable to tell apart patients who would benefit from early aggressive therapy from those who might be spared harsh treatment for less invasive cancers," Ms Johns said.

She said the study data would be freely available to cancer researchers and clinicians, to build on this work.

More information: Whole genome landscape of Pancreatic Neuroendocrine Tumours, *Nature*, nature.com/articles/doi:10.1038/nature21063

Provided by University of Melbourne