Genomics provide hope for those with 'one in a million' cancer diagnosis

November 13 2018

New research has shown that many Australians with rare cancers can benefit from genomic profiling. The findings of the patient-driven trial are being presented today at the Clinical Oncology Society of Australia Annual Scientific Meeting and could result in dramatic changes to the way those with rare cancers are diagnosed and treated.

The initial data from the pilot study for Nominator Trial is being presented by Professor Clare Scott from the Walter and Eliza Hall Institute of Medical Research and Peter MacCallum Cancer Centre, and was funded in part by Rare Cancers Australia.

The data shows that genomic profiling provides meaningful information that influences diagnosis and treatment in approximately 50 percent of people with rare cancers. 20 percent of those tested got a new treatment plan as a result and 6 percent of participants were given a new diagnosis.

The aim of the national initiative is to trial the use of genomic testing to match rare cancers to cancer treatments. Testing is used to identify molecular features of the cancer or genetic mutations that can be targeted with existing treatments used in other cancer types with the same characteristics.

While genomic testing is becoming increasingly used in other cancer types, this is one of the first Australian studies of its kind to look at the potential benefits for those with rare cancers, which have very low survival rates.

Professor Clare Scott says the initial pilot data is exciting and proves that there is a current unmet need.

"The treatment options for Australians with rare cancers are currently extremely limited and this ultimately leads to poor survival rates. Research has also typically been restricted because of the challenges of finding enough of each type of cancer patient to design appropriate clinical trials.

"Australians in this trial came to us after they had exhausted all their options. The cancers they had are extremely rare—the chances of being diagnosed with these cancer types are often around one in a million.

"Using genomic profiling we were able to uncover new information that gave many patients new treatment options—and ultimately, new hope."

Professor Scott will share examples during her presentation of those who have benefited.

"In one case we were able to identify that a rare heart tumour actually had a genetic profile most closely resembling a melanoma. Using that information we were able to get access to the latest treatments that are benefiting melanoma patients—which we hope will provide better outcomes for this patient."

The Nominator Pilot Study results released today included 36 patients. The two-year study will eventually include 100 patients and will lay the groundwork for other national initiatives looking into genomic profiling across a range of cancer types.

Professor Phyllis Butow, President, Clinical Oncology Society of Australia said one of the impressive things about the study was that it was driven by Australians directly affected by rare cancers.

"Around 52,000 Australians are diagnosed with rare or less common cancers each year. Those directly affected by the disease, led by Kate and Richard Vines from Rare Cancers Australia, helped call for and fund this research, so it's great to see these initial promising results being presented to cancer experts from across the country."

Provided by Walter and Eliza Hall Institute