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VIC NEWS

Whole exome sequencing gives 40 per cent of patients useful data about disease for first time

In-depth genetic testing is offering hundreds of Victorians never-before-available information about their medical condition which can be life-changing, or life-saving in some cases.

Brigid O'Connell, Health reporter, Herald Sun

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New comprehensive genetic testing is giving hundreds of Victorians vital information about their medical condition, which is changing treatments or redefining diagnoses.

The first report card from the Melbourne Genomics Health Alliance shows that in its initial five years more than 3800 Victorians with one of 11 rare or serious health conditions were tested.

Almost 40 per cent or more than 1300 people received clinically useful information.

This included delivering a more accurate diagnosis, ruling out a disease or changing their management. This rate was 19 times higher than through standard testing.

Melbourne Genomics director Professor Clara Gaff said whole exome sequencing was proving life-changing or life-saving for conditions such as cancer, heart disease, bone marrow failure, superbug infection and neurological diseases.

"When we started there was a view that so-called rare diseases weren't going to be treatable. But we've found that's not the case," Prof Gaff said.

"It's allowing families to link in with others around the world with the same condition, doctors can have a better idea of the likely cause, and in some cases it's possible to offer treatment for the first time," she said. Prof Gaff told the alliance's symposium this week that they were now focusing on research to increase the strike rate of the test and pick up those who still cannot get a diagnosis.

An inherited kidney disease has seen Bree Faulkner, 30, hover at around 17 per cent kidney function for most of her life.

Bree Faulkner with her husband, Michael, and son Tyler, nine months. Picture: Sarah Matray

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Ms Faulkner was one of 204 kidney patients to undergo the new genetic testing, which changed or clarified the diagnosis for a third of those patients.

The new tests revealed Ms Faulkner had a recessive version of polycystic kidney disease, so any of her children — including nine-month-old son Tyler — will be carriers and not affected by the disease.

“It gave us information and also the courage to start our own family,” she said.

“The benefits go on. Now Tyler has that information going forward.”

brigid.oconnell@news.com.au