Dr Bryony Thompson helps doctors and genetic counsellors bring a little more certainty to patients on the receiving end of genetic testing ...

Dr Bryony Thompson is an adult genomics curation scientist, and she’s loving every minute of it.

Based in the Department of Pathology at the University of Melbourne and working with Professor Ingrid Winship’s Adult Clinical Genetics group, Dr Thompson plays daily in the wonderland that is variants of uncertain significance.

“It’s my dream job,” she tells the MJA.

When a patient is tested for genetic causes of their particular cancer, a raft of results come back from the lab. Sometimes it is clear that a particular genetic variant is the cause of the disease. Sometimes it is clear that it is not.

And then there are the variants of uncertain significance (VUS).

“They are the bane of doctors’ and genetic counsellors’ existence,” says Dr Thompson, “because they have to explain to patients what has been found.

“Some doctors don’t quite understand VUS and then treat it as a significant result when it should not be.”

And that’s where Dr Thompson’s job begins.

“Part of my job is to pull information from all these different databases, and my own expertise, to try and make more sense of that variance,” she says. “Luckily there are computer programs that pull all the information together.”

Sometimes, she says, results can come back from laboratories across the country or around the world.

“At the moment they just send it to different labs around the country or around the world. “The [Royal Melbourne] wants to start doing whole exome sequencing for some cases that are a bit difficult to figure out and look like they may have a genetic cause for their disorders or disease,” says Dr Thompson.

“At the moment they just send it out to different labs around the country or around the world.

“Ingrid found some money so now part of that process is trying to curate the genomes to figure out what we can find in the exome sequencing that may be causing that particular phenotype.”

It was another example of being in the right place at the right time, she says.

“I dropped my fellowship because this is my dream job. I’d love to do this for a long time.

“I had wanted to get out of academia and get into the more diagnostic, lab-based work, so the timing was perfect. I’m really excited about this opportunity.”

How would she recommend this job to upcoming students?

“It’s a job where you’ll have a lot of variety,” says Dr Thompson.

“Because the cases that will go up for whole exome sequencing are the difficult cases where there is possibly no defined set of genes [causing phenotypes] you really have to look at all of them.

“There is so much variety, and it’s all challenging, but it’s going to be exciting.”

Cate Swannell

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“Earlier this year Dr Thompson came back from Utah to complete her fellowship at the University of Melbourne. Three days later Professor Winship invited her for a cup of coffee.

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