Ten stories of ADPKD in Australia

**ANNA**
“I don’t define myself as a sick person or think that PKD should hold me back in any way.”

**FRASER**
“Those seem to be a good chance I will die of old age before I need to think about dialysis.”

**CHARMAINE**
“I’m still young, and I still look healthy. People just assume I am healthy rather than having issues to deal with.”

**LAURA**
“My mum was the strongest person I have ever seen. She never said, I can’t do this because of my disease.”
Autosomal Dominant Polycystic Kidney Disease (ADPKD) is a genetic, progressive disease characterised by the formation of cysts which increase kidney size and can ultimately lead to kidney failure. Approximately 10,000 people are living with ADPKD in Australia.*

Rare Humans is a project inspired by the courage and optimism of people living with ADPKD. This is a collection of unique stories about Australians who have chosen not to be defined by their diagnosis.

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www.rarehumans.com.au

ANNA  
RYDE, NSW  
43 years old  
Page 4

MITCHELL  
SYDNEY, NSW  
36 years old  
Page 8

FRASER  
TOORAK, VIC  
51 years old  
Page 12

ALICIA  
CENTRAL COAST, NSW  
28 years old  
Page 16

CHARMAINE  
COLLAROY, NSW  
29 years old  
Page 20

JODIE  
NARRAWONG, VIC  
47 years old  
Page 24

JULIE  
NORTH ADELAIDE, SA  
50 years old  
Page 28

LAURA  
OAKPARK, VIC  
29 years old  
Page 32

JODIE  
MELTON, VIC  
33 years old  
Page 36

MELISSA  
MONT ALBERT, VIC  
47 years old  
Page 40
The threat has done little to slow down the pace of her international career or life with her three children. A senior human resources manager with a leading management consultancy, Anna, now in her early 40s, has an overseas role as well as leading the company’s human resources services in Australia.

The family’s history suggests that Anna, too, may eventually need dialysis or transplantation. “I try not to be overwhelmed by that, because you could let it become all-consuming,” she says. “Sometimes when I look at the future I pull back and realise that there’s so much more in life. This is just one element that I have to live with.”

**PKD in the family**

As a child Anna visited her grandmother who was having dialysis at home. She became one of Australia’s longest-standing dialysis patients, being treated for more than 20 years. For a child, exposure to an unwell older relative was disturbing. “The machine was like another person in the house,” she says. “It was confronting to learn that her blood would leave her body, get cleaned and get put back in again.”

Anna’s father provided a very different role model. “My father was diagnosed with PKD in his mid-20s,” she says. “He discovered he had the disease but was determined not to become ‘a sick man’. Dad’s been a very positive influence. Like him, I don’t define myself as a sick person or think that PKD should hold me back in any way.”

Anna’s father ignored the fact that he had PKD - he had no symptoms – until his kidneys failed with little warning 20 years after the diagnosis. After about 6 months on dialysis he had a kidney transplant from his brother. “Now in his late 60s he is well, goes to the gym every day, and lives like someone decades younger,” she says.

When Anna was 16 she and her older brother had an ultrasound to check whether they, too, had PKD. “At that point we didn’t know very much, but we had two extreme examples in the family - my grandmother having dialysis and my dad who said there was nothing wrong with him.”

“In hindsight, 16 is not a great age to find out something like this but, to be honest, I didn’t think much of it. My mother was very distressed so I knew it was something bad, but in my head I thought, ‘I’m going to be just like my dad’. I was invincible, like all teenagers, and I didn’t really change very much.”

Anna’s younger brother was also diagnosed with PKD, and noticed the first symptoms such as kidney pain and blood in the urine in his early teens. Most recently the family has discovered that Anna’s oldest son has PKD too, after suffering a series of kidney infections at the age of 15.

“I was with him when he had the ultrasound,” she says.
HE KNOWS ABOUT PKD.
HE HAS SEEN ME IN AND OUT OF HOSPITAL AND THINKS IT’S NO BIG DEAL, BECAUSE HE ALSO SEES ME WHEN I’M VERY HEALTHY.

"I saw the cysts on his kidneys, and I knew what it meant. He said, ‘It’s fine mum!’, but I did have a heavy feeling of guilt. It’s one thing to have a theoretical discussion when you’re pregnant about the risks for your children, but very different to find out your 15-year-old has the disease.

"He knows about PKD. He has seen me in and out of hospital and thinks it’s no big deal, because he also sees me when I’m very healthy.”

Having children
Anna had no symptoms or complications associated with PKD until she became pregnant and had her first child at the age of 25. "I was immediately told it was a high-risk pregnancy. This was the first time I thought that life might not be smooth sailing,” she says. Complications arising from her kidney disease included high blood pressure and pre-eclampsia, requiring multiple hospital stays.

Two years later her second pregnancy was relatively straightforward - although closely supervised by her medical team – and her second son was born.

Twelve years later Anna had her third child, a girl who is now almost three years old. It was another complicated pregnancy, but managed expertly by a team including her long-standing nephrologist, an obstetrician, and a physician who specialises in the medical aspects of complex pregnancies.

“When we found out we were pregnant with our first son, one of the first questions was whether we wanted genetic counselling,” Anna says. “I asked about the purpose and was told we could decide not to have that child. I was 25, I didn’t have any complications, I could see that my dad was healthy, so I asked myself, why would I not have this child?”

Anna remains very optimistic about the future for herself and her children and the potential advances in PKD treatment. “The world my grandmother lived in when she had PKD was different to the world of my father, and my world, and my children’s world,” she says.

Dealing with complications
“I bounced back to good health straight after my first two pregnancies,” Anna says. “The other effects of PKD didn’t happen until I turned 30 when I had three kidney infections and a haemorrhagic cyst in one year. Suddenly I thought, ‘OK, I have to think differently about this, it’s not going to be as simple as it has been for Dad. Since then I’ve averaged one major episode of infection or haemorrhage, with a trip to hospital, every year, and it can take up to a month to recover.”

Apart from a short break, Anna has been under the care of the same senior nephrologist since her early 20s. They have an excellent professional relationship in which Anna is acknowledged as an expert in her own disease, and able to recognise when she is becoming unwell.

Copies of test results are routinely sent to her as well as her doctor. “It’s your information,” her specialist has told her. ‘Knowing her numbers’, for example about her current kidney function, helps Anna communicate clearly when she needs to consult other health professionals.

She has been lucky to find a GP who understands PKD, including the fact that complications can extend beyond the kidneys. Some years ago she developed a severe and persistent headache and her GP urged her to have some tests. They identified a dissection of her vertebral artery – a small tear on the inside of the blood vessel - which can occur in people with PKD. It responded well to treatment.

“Having a good relationship with your GP is critical,” Anna says.

Her experiences have not always been positive. “One doctor was convinced I did not have a kidney infection, when I knew that I did. You do need to be an expert in your own disease, and find doctors who you can trust,” she says.

Her workplace has been very supportive during the bouts of illness. “I was very honest when I accepted my current role and have always explained why I need time off work. It can be hard for an employer to understand that someone can have a disease but not be sick. I always have a contingency plan in case I get unwell, and I have an amazing team around me who can pick up things at work.

“The long hours and travel are starting to make life harder. I’m not at optimum health and I push it by working hard, but I love what I do and don’t want to be constrained.”

Looking to the future
Anna’s nephrologist is optimistic about the status of her kidney function, telling her that the need for dialysis or transplantation is probably 10 years away. “But I know I’m coming up to the age when both my grandmother and my dad developed kidney failure,” she says. “All I can do is stay optimistic, do everything I can in terms of healthy lifestyle, take my medication, and continue to bounce back when I do have complications.”

She appreciates the work of organisations such as the PKD Foundation of Australia and Kidney Health Australia. Anna sees two major roles: connecting the community of people and families with the ‘invisible’ condition of PKD, and supporting research. “There’s also a lot more that can be done to support organ donation,” she says.

“There is so much scope for involvement and advocacy, but maybe that’s for when I don’t have a three year old and a big job!”

Being the third generation of her family with PKD, Anna has had the opportunity to come to terms with the disease and its possible implications. And her advice for other people with PKD? “Take life one step at a time.”
“The other effects of PKD didn’t happen until I turned 30 when I had three kidney infections and a haemorrhagic cyst in one year.”
Mimi, the heroine of Puccini’s opera La Bohème, sighs her last breath in the closing bars of tonight’s performance. The outdoor stage is suspended over the water of Sydney Harbour with the floodlit bridge as the backdrop. Opera Australia’s site and venue operations manager Mitchell (Mitch) also sighs, with relief, ticking off another success as this month-long production winds up.

Mitch, aged 36, has autosomal dominant polycystic disease (ADPKD) but it’s had no effect on his career. Initially a production designer working on kitchens and exhibitions, he then moved to the staging of major rock concerts and music festivals. He now has a full-time role managing the venues for Opera Australia’s annual cycle of outdoor performances in Sydney and Queensland’s Gold Coast. “It’s a challenge, but a good one,” he says.
MITCHELL
36 years old Sydney, NSW

PKD in the family
There is no obvious history of ADPKD, an inherited disease, in Mitch’s family so his condition probably resulted from a spontaneous mutation. His parents, brother and sister are free of the disease. When Mitch was 16 he had pneumonia and some routine X-rays suggested his kidneys were unusually large. A follow-up ultrasound confirmed they contained cysts typical of the disease.

“I didn’t think much about it at all,” Mitch says. “A 16-year old teenager doesn’t think much about the future, and I was told my doctors would just keep an eye on it.”

His GP referred Mitch to a renal physician who monitored his progress once a year during his 20s. “There

MY KIDNEYS LOOK LIKE A SACK FULL OF MARBLES. OCCASIONALLY DURING A CHECK THERE WILL BE A TRAINEE DOCTOR WHO WANTS TO HAVE A LOOK AT THE SCANS, AND I SEE THE SURPRISE ON THEIR FACES.”
were never any major concerns. When I was about 24 I started one tablet a day to treat my high blood pressure, but it’s well controlled.

“My kidneys look like a sack full of marbles. Occasionally during a check there will be a trainee doctor who wants to have a look at the scans, and I see the surprise on their faces. My kidneys are enlarged but I’m quite tall - six feet five - so that’s not causing any problem. I sometimes have back pain, but that could be because of my height rather than from my kidneys. I don’t have any other pain or other symptoms, so PKD has had very little effect on my health.”

**Having a family**
The biggest impact of PKD has been on the decisions Mitch and his wife have made about having children. Genetic tests identified the mutated gene in his DNA and a genetic counsellor provided expert information about the options.

“They did some DNA research into our family, including my parents and siblings, and worked out where the mutation was sitting. We decided that, rather than take the 50/50 chance of passing it on, we would go through IVF so they could scan our embryos and choose one without the ‘broken’ gene,” he says.

“We did talk about it a lot. IVF, apart from anything else, is a tough process for women, it’s very ‘medical’, and it’s expensive. But we decided that, for us, it made sense. We’re very lucky to live in a country as wealthy as Australia where we can make these types of choices. And if you have the choice, why would you not?”

Their daughter, now two and a half, is thriving, and they are thinking of having another child.

**Looking to the future**
Mitch still feels a little like the nonchalant 16 year old who discovered he had a kidney disease. “PKD doesn’t change my life, and I’m healthy. I had an occasional freak-out when I was younger, but I know that, for now, it’s not changing my standard of living,” he says.

His kidney function is currently about 40% of the normal level. “It’s not in my personality to follow it obsessively. If it gets too low I know someone will tell me, but if I track it myself, it’s just something else I have to worry about.”

He relies on his medical team to keep him up to date about his condition and any potential developments in treatment. “If you rely on something like Google, there’s probably a lot more misinformation than good information,” he says. “I have always trusted my GP and my kidney specialist, and I’ve never felt the need to look things up or double-check what they are telling me. You need to find doctors who strike the right balance between being overly concerned on one hand or, on the other hand, not concerned enough.”

Mitch says his apparently relaxed approach to PKD reflects the way he deals with his life, but he does take the disease seriously. “There are no decisions to make at the moment and it’s hard to be proactive. If the situation changes then we will deal with the new information at the time,” he says.
Pedalling 150 km a week on his bike, fast, is Fraser’s idea of relaxation. For this 51 year-old web designer who works from his Melbourne home, a diagnosis of autosomal polycystic kidney disease (ADPKD) four years ago has done nothing to slow him down.

“I was having an ultrasound to check for another problem when the radiographer turned the screen around and said, ‘This doesn’t look normal!’ I had to agree with her,” he says. “I could see my kidney and there seemed to be dots and lumps all over it, and then realised that both my kidneys were affected. I wasn’t too worried but I knew I’d need to get them looked at a bit more closely.”
Fraser had always been fit, healthy and active, and a keen cyclist for many years. He owned and operated a bike shop and expanded the business to include online parts sales in the early years of e-commerce, until the daily grind of a retail business prompted him to move in a different direction.

“Until I know there’s a problem, I try not to worry about things or dwell on the future too much,” he says.

“I saw my GP straight after my ultrasound. He’s older and very experienced, but it was the first case of PKD he had ever seen. He used me as a guinea pig a few times after that with trainee doctors, checking if they could pick up the condition.”

While Fraser was “a little” concerned about the diagnosis his wife was more assertive about the need for some specialist advice. He was very fortunate in having a trusted GP, as well a brother who is a GP and an uncle who is a kidney specialist.

“My uncle told me to find the best nephrologist possible. He said it’s a long-term condition, so look for a specialist of about the same age so you can grow old together.”

Clinical trial
Soon after his diagnosis Fraser was referred by his GP to the hospital renal clinic and has remained under its care. Initially he was told there were no options for active treatment, other than doing his best to stay healthy and fit. The aims of his care were to establish some baseline measures, including his kidney function and blood pressure, and follow his course over time.

About 18 months later there was an opportunity to participate in a clinical trial of a medication in PKD.

“I asked him about the pros and cons of the study, but I knew there was little hope of other treatment,” Fraser says. “There was the potential that the drug would help keep my kidneys working for longer, even though we don’t know the answer yet.

“The clinic already had some history about the path of my kidney function, so I was an ideal candidate. The decision really was a no-brainer for me. The medicine had been approved for the treatment of PKD in some countries, so it wasn’t really experimental, and I felt I had very little to lose.”
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“The alternative of doing nothing was not palatable for me. If there is anything that will help me get better – or at least not get worse – then I’ll consider it.”

Participating in the trial means that Fraser has regular check-ups with the team and has access to expert care. “Monthly visits could be difficult if I was working 9 to 5 for an employer, so it was fortunate I had the flexibility of working from home,” he says.

Living with PKD
Fraser’s healthy lifestyle, established decades before he learnt he had PKD, has stood him in good stead as he ponders the next steps. “There seems to be a good chance I will die of old age before I need to think about dialysis or transplantation,” he says.

His kidney function is now about 30% of the healthy level for a person his age, and it seems to be declining quite slowly. Apart from the clinical trial for ADPKD, he is not taking any other medication and he does not have high blood pressure.

In fact, he has thought about the next steps and is concerned about the possibility of needing dialysis. “It’s very limiting being tied to a machine for three days a week. I lead a pretty active life, so that would have a big impact,” he says.

His aunt and two brothers have both offered to donate a kidney if he ever needs a transplant.

Overall, he feels PKD has had very little impact on his life apart from the effects of taking a medicine for ADPKD. “There is a silver lining: the disease is an added incentive to keep fit,” he says.

Fraser appears to be the first member of his family with autosomal dominant PKD so it may be the result of a spontaneous mutation rather than being inherited from one of his parents. His siblings have been tested and are free of the disease.

“My two boys are 12 and 14, so we’re coming up to a time when they might have some scans,” he says. “We have chatted briefly but haven’t talked to them in detail about the possibility that they have PKD. We’ll deal with it at the time – worrying won’t help.”

“THERE IS A SILVER LINING: THE DISEASE IS AN ADDED INCENTIVE TO KEEP FIT.”
The moment you first learn you have autosomal dominant polycystic kidney disease (ADPKD) is one of the most challenging times for people with the condition, according to Alicia, aged 28.

“I went home with my ultrasound pictures and held them up to the window,” she says. “I compared them with some pictures I found online of polycystic kidneys, and figured out I probably had ADPKD. Then the report came and it said, yes, you have this disease. That day was really sad because I was home alone with my computer, trying to find information.

“You start out thinking, ‘Oh, I’m going to have to change all these things in my life,’ but actually it’s not that bad,” she says.

“At the moment it doesn’t affect me too much besides from a little bit of pain here and there and high blood pressure. But other than that, I don’t really pay attention to it. I don’t notice it.”

Alicia works part-time as an administration assistant in Sydney. She has just completed a Masters of Research in International Studies/Spanish Studies, examining relations between the government of Colombia and the country’s rebel FARC group.

Alicia was diagnosed with moderately high blood pressure when she was 22, which is unusual in a young, healthy person. She saw a new GP when she had a series of ear infections and a urinary tract infection. During a routine physical examination the GP was able to feel her enlarged kidneys through her abdominal wall: healthy kidneys are too small to be felt in this way.
He told me to get an ultrasound. While I was having it the radiographer asked if I had ever been diagnosed with ADPKD, but couldn’t say any more because that was the doctor’s job.

Among Alicia’s many reactions to receiving the diagnosis was a feeling that she had some kind of ‘time limit’ on her life. “I thought, I need to do all these things before it’s too late.”

After the ultrasound Alicia also had a CT scan to provide a clearer picture of her kidneys, and later an MRI of her brain to check for cerebral aneurysms that can result from the high blood pressure that occurs in ADPKD. She carries the CT image of her kidneys on her phone so she can easily show it to health care professionals if needed.

Finding information and support
After the diagnosis Alicia felt her GP offered little information or direction on the next steps. She found a new GP who responded more actively to her concerns and referred her to a nephrologist (kidney specialist). “My GP said I needed someone to monitor me and make sure that if anything is going wrong, then we can find out as early as possible,” Alicia says.

In the majority of PKD cases there is a family history of the disease. In Alicia’s case no-one – grandparents, parents or siblings – had PKD. “I’d never heard of it before,” she says. “For me it was a complete surprise, so it was a bit scary. I was the only one and I didn’t know what it meant.”

Google was one of the first stops in her search for information. One website included a presentation from an American woman with ADPKD who had received a kidney transplant. “She was really interesting and really positive. You think, ‘Oh well, she’s doing okay, she’s got a career, she’s making the most of her life’. But I felt she was too far away, I needed someone here, in Australia.”

Alicia was eventually put in touch with the PKD Foundation of Australia, and found that other people living with the disease are keen to share their stories and offer each other support.

The PKD community
Alicia has been working with the PKD Foundation of Australia to support other people with the condition, educate patients, families and the public, and encourage research. “The Foundation was set up by families, many of whom were already friends, so they already had their own community in a way. I’m keen to expand the range of people involved,
and I’ve written a few blogs, filmed a patient video with them and have been to some events,” she says. Alicia is a little unusual in not having other family members with PKD. “When a whole family is affected they automatically have their own support network. It’s a bit harder when you’re the only one,” she says. “But the Foundation has been good. I have benefited a lot because I was able to meet other people with the same disease who tell me, ‘It’s okay, it’s life, it’s nothing unusually special’.”

**Working with health professionals**

“I was confused by what I was told at first, like ‘Oh, just don’t eat junk food’. Things have improved since then,” she says.

She initially saw a nephrologist every six months, but as her disease is stable that has been wound back to once a year. Alicia is now a participant in a clinical study looking at whether a high water intake can slow the progression of ADPKD. “Now I feel I’m giving blood and urine samples all the time, so if anything is wrong they will tell my nephrologist.”

Having her health constantly monitored has its downside as well as benefits. Doctors tend to be cautious and can be a little negative about her plans, for example to travel the world. And every visit to a doctor or other health professional is a reminder that she is living with ADPKD.

Her nephrologist also told her about clinical trials of a new medication. “It’s good to keep up to date with new information that’s coming out,” Alicia says.

**Family and friends**

Alicia was living at home with her family when she was diagnosed, and praises them for their reaction. “My parents were especially worried at first because they didn’t know what it meant, just the same as me. They did the normal things that families do, giving you support and a lot of affection, making sure that you’re looking after yourself. One of my sisters was overseas. She got very worried but she handled it well. My two sisters have always been so supportive.

“I did tell my friends. It didn’t faze them, which was good because it can be scary for people when you tell them. When I tell new people now, for whatever reason, I need to explain everything to them. But my friends didn’t worry so much and it didn’t change how they felt about me, which is nice.”

**Life with ADPKD**

Alicia looks more carefully at her diet than in the past. She takes medication for high blood pressure, paracetamol occasionally for pain, and fish oil and vitamin D. She was told to avoid contact sport because of a risk of damaging her enlarged and vulnerable kidneys, but that’s not a burden because it was never her favourite past-time.

She feels she doesn’t have as much energy as in the past and tires more easily. There are no obvious signs of ADPKD, so it can be difficult for other people to understand the significance of the disease and its possible long-term effects. “That can be difficult when you don’t feel so good and people just assume you’re lazy or have some other problem,” Alicia says.

She has occasional abdominal and back pain. “If I eat too much, I get really sensitive around my abdominal area. One time after dinner I was washing up and my brother poked my sides and I yelled at him because it was so painful. My kidneys are really big so everything is a little ‘squished’ in there. But at the moment it’s not really affecting my life too much.”

When she was first diagnosed Alicia had multiple medical appointments and tests, and was also facing the uncertainty of what the condition would mean for her. “At that point it was a little bit traumatic because every day included something to do with the disease, and it was affecting me, but now it’s fine. Everything has settled a little bit and things are under control. I had to learn how to think about myself in a different way, accept myself as I am. That’s something you have to do when you’re a teenager and then I had to do it again as a young adult. Whenever I want to do something new, I do think, ‘Oh, can I do this because I have ADPKD?’”

Her health care team included a dietician who encouraged her to limit salt, fat and protein in her diet. “At first I was really, really strict but now I just try to eat healthily,” she says. “This is difficult sometimes because I like bread and meat. I probably should be a bit more diligent on what I’m eating, but I always say to myself, ‘I’ll start again tomorrow’.”

**The future**

Alicia’s kidney function is relatively stable for now. “I don’t worry so much about when I might develop kidney failure, but there’s this great unknown.”

Alicia’s sister has been assigned to the renal ward in the hospital where she works, seeing first hand some of the longer-term outcomes of ADPKD. While some patients with advanced disease are having dialysis, many have had active lives at home and at work, and have raised families. “My sister tells me not to worry, because that’s a long way off,” she says. Having children is a possibility sometime in the future. Alicia will work through the implications with her doctors, including any extra risks that pregnancy and childbirth might pose for her ADPKD and the best way to manage them.

For someone just diagnosed with ADPKD, Alicia has some simple advice. “It’s not as scary as it seems.”

*“When a whole family is affected they automatically have their own support network. It’s a bit harder when you’re the only one.”*
For Charmaine, aged 29, having polycystic kidney disease is simply a “fact of life”. It hasn’t stopped her from leading a normal, active life, having a child, and establishing a career as a scientist and researcher.

Charmaine’s condition is autosomal dominant PKD (ADPKD). There’s a 50/50 chance that a child of a person with ADPKD will inherit the condition. Her grandfather had ADPKD, as do her father and her sister. And when Charmaine’s son Oliver, now a toddler, was just a few weeks old, small cysts were discovered in his kidneys.

It’s a little poignant that her career has been in embryology - the science of how a single fertilised egg develops into a fully-formed human - given that her family is affected by this inherited condition. She has worked in IVF services in Canberra, and has been forced to make hard decisions about whether she should have IVF herself so that the potential risks of ADPKD in her own children can be addressed.

Her memories of being diagnosed at the age of eight are a little hazy. “I remember getting an ultrasound and my mother being quite upset,” Charmaine says. “But as a child, it didn’t really sink in and I didn’t take much notice of it. I saw my father living with ADPKD and thought, ‘Oh, he’s fine, so I’ll be fine’.”

In fact, her father later developed an aortic aneurysm associated with high blood pressure caused by ADPKD. This weakness and bulging in the wall of his largest artery was life-threatening. “They managed to do incredible surgery and basically save his life, and since then he’s been on blood pressure medication,” Charmaine says. “Now he’s older, his kidney function is starting to decline more rapidly than before. And my grandfather – my dad’s father, who had ADPKD - had a kidney transplant in his 50s after a kidney injury. He was on a lot of medication after the transplant, which affected his health and mental wellbeing. He is no longer with us.”

Charmaine started getting urinary tract infections (UTIs) from an early age. “Probably around eight was my first, and I’ve struggled
with them since then, especially through my teenage years,” she says. Repeated UTIs as a child prompted a GP to arrange an ultrasound which revealed the distinctive pattern of multiple cysts within both kidneys. Together with the strong history of the disease in her family, her GP and a nephrologist (kidney specialist) were confident the condition was ADPKD.

“I’ve also had kidney infections and cyst infections, and pain in my kidneys occasionally. During my pregnancy, my blood pressure skyrocketed towards the end,” Charmaine says. “My blood pressure had always been good until the end of my pregnancy.”

A fungal infection of a cyst was one of the most unpleasant complications. “It was so painful, one of the most painful things I’ve experienced,” she says. “I was sick for a long time. Although the treatment of the infection is relatively simple, it took some time to make the diagnosis.”

The progress of ADPKD varies widely between people with the condition, so nothing is inevitable and nothing is certain. Right now, Charmaine’s blood pressure and kidney function are close to normal. Charmaine’s sister has a different pattern of symptoms. “She’s two years younger and she has had high blood pressure. She was on medication really early to control it and now it’s stabilised,” Charmaine says. “She doesn’t have as many cysts as me. Mine...you couldn’t even count, there are so many.”

Her sister was a young adult when the symptoms of PKD became apparent. “She struggled with the information she was given and this was overwhelming for her, whereas I had learnt more about the condition over many years.

“It’s scary for me too, the thought that my kidney function will decline one day, and I might need a transplant, or that it might shorten my life. I don’t think my family told me at a young age what it meant to have ADPKD, so I never really understood the full implications as a child.”

Living with ADPKD
Apart from her bouts of infection and occasional pain, ADPKD has not had a major impact on Charmaine’s day-to-day life and she is not taking any medications. “We always exercised and ate relatively healthily as children,” she says. “I grew up playing soccer and I kept that up until I started university and ran out of time. Now it’s really hard to exercise with a toddler – except for chasing after him – but I used to swim a lot when I was working. “When I was younger I didn’t want to follow the advice to drink a lot of water, because it made me need to go to the bathroom a lot and that was really inconvenient. I’m much better at it now. My nephrologist is very adamant about me having a low salt diet.”

She sees a nephrologist once a year to monitor her blood pressure and kidney function, which includes blood tests and a 24-hour urine collection. “That’s not much fun,” she says. Charmaine recognises that monitoring is essential, but it is also a reminder that she has a health issue to deal with. “I liked having a period of my life where I didn’t think about ADPKD at all,” she says. She also sees her GP as needed if infections or other symptoms cause problems.

“I’m still young, and I still look healthy. People just assume I am healthy rather than having issues to deal with.”

“I’m still young, and I still look healthy. People just assume I am healthy rather than having issues to deal with.”

CHARMAINE
29 years old Collaroy, NSW

“I’m still young, and I still look healthy. People just assume I am healthy rather than having issues to deal with.”
is supportive. I now have a lot of friends who are studying medicine, and one of my previous managers was a genetic counsellor. They realise the implications, and it was great to have their understanding. My manager was always very caring and wanted to help, and if I needed to have time off work due to PKD she was really good about it.”

Charmaine thrived as a research student and was active in the Bosch Young Investigator Group. The initiative is supported by the Bosch Institute, a major centre for medical research at the University of Sydney. “We’re a group of young scientists who try to promote development opportunities for other young scientists,” she says. “It’s such great experience, practising your communication skills and collaborating with other researchers. I really love research – I’m very passionate about it.”

She is now working with the PKD Foundation of Australia, providing advice both as a person living with PKD and as someone familiar with the intricacies of scientific research. Her personal experiences provide unique insights that can help guide the Foundation’s work and support other people with PKD.

**Having a family**

Charmaine and her husband were friends for years before getting together. “He used to say he’d marry me one day. When I told him about my condition he just said straight away, ‘I’ll give you a kidney!’”

It’s possible to conceive a baby through IVF and then test the embryo for ADPKD before having it implanted and proceeding with the pregnancy. “When we were planning on having a family, we did discuss if we wanted to go through IVF, but we just didn’t feel like it was right for us at the time,” Charmaine says. “As an embryologist I saw people going through IVF all the time, but for a number of reasons I chose not to.”

After knowing that her first child most likely has ADPKD, Charmaine is faced with challenging choices if she wants another child. “It’s complicated to think about IVF and then removing embryos that have ADPKD, and I know there are other risks associated with IVF as well.

“If someone had removed me as an embryo then I wouldn’t exist today, and then my son wouldn’t exist, and I wouldn’t change his existence for the world!”

**HAVING ADPKD AFFECTS ME MUCH MORE NOW THAN IT EVER USED TO BECAUSE I’M MUCH MORE AWARE OF WHAT IT MEANS IN THE LONG TERM.**

She is also concerned about the toll that each pregnancy might have on her kidneys, and whether she will be healthy enough for another pregnancy if her kidney function starts to decline.

“I thought, I’ve got ADPKD, I feel fine, I think it’s something you can live with. But now I have my son, and he seems to have a more severe presentation. Having ADPKD affects me much more now than it ever used to because I’m much more aware of what it means in the long term.”

The hardest time for Charmaine was when her son was diagnosed with the condition at six weeks of age. “He was at the hospital at three weeks with a urinary tract infection (UTI) and when we had his ultrasound he had cysts, and also what they think is nephrocalcinosis – deposition of calcium in the kidneys.

“My ADPKD is stable. The thing that bothers me the most is not knowing what exactly is happening with my son’s kidneys. There’s not as much known about paediatric ADPKD as there is about adult ADPKD, and our doctor isn’t sure whether this nephrocalcinosis - or whatever it is - will go away. That worries me.”

**Looking forwards**

“My kidney function is still fine, but if it does decline, how will that make me feel?” she says. “I hope new treatments, and even a cure, become available, and that we can do something to prevent the shortening of life that can happen with ADPKD patients.”

Kidney dialysis and then transplantation are a possibility at some point in the course of the disease. “I haven’t really thought about that. At the moment it’s wait and see, and I feel I’ve got quite a while before it happens. There’s a lot of research going on and the situation is changing all the time.”

And what advice would Charmaine give to other people with ADPKD? “Don’t be afraid to talk about it if you feel that will help. My experience has told me people are very supportive. And look after yourself...your diet, at the moment, that is something we can control.”
Jodie’s father, a dairy farmer on Victoria’s south-west coast, devised a home-made shield to protect his enlarged and vulnerable kidneys from an untimely kick from a cow while milking. Jodie has inherited not only his autosomal dominant polycystic kidney disease (ADPKD), but also his down-to-earth attitude.

Each child has a one in two risk of inheriting this autosomal dominant form of the disease from a parent with the condition. Jodie’s generation has been unlucky as she and each of her three siblings have ADPKD. It was just a one in sixteen chance that all four children would be affected.

The family history of ADPKD extends at least as far as her father’s mother. “My grandmother passed away at the age of 60 from a heart attack, just as she was about to start dialysis for kidney failure,” Jodie says. “My father was on home dialysis, but still milking cows, when he developed kidney failure at the age of 52. He had a transplant six months later, and lived until he was 65.”

Jodie has two boys in their early 20s, but they haven’t yet been checked for PKD. “They know about the strong family history, but I have said to them, ‘Enjoy yourself boys, it doesn’t impact on us until later in life. Our family mantra is, live your life to the full and then pull your head in if you need to, but be mindful of your health.’”
Living with PKD
Now aged 47, Jodie has been a nurse since she left school. “I first found out that I had ADPKD when I was 19 and training to be a nurse,” she says. Jodie volunteered as a ‘patient’ for radiographers who were practising how to do ultrasounds. “They said, ‘Ahhhh…there’s something not quite right,’ because they could see cysts on my kidneys. And I said, ‘I have a strong family history of PKD, don’t worry about it’.”

Her parents were also philosophical about the diagnosis, and the family followed the example set by her father. “Dad didn’t get sick until his late 40s or early 50s and PKD wasn’t a big presence in the family - we never put great emphasis on it,” Jodie says. “He led a normal and healthy life. Even after he had his transplant he still milked the cows, did his own thing, and kept working until he decided to retire from the farm.”

A diagnosis of ADPKD had no immediate impact on Jodie’s life, or her medical care. Her GP was aware of it but she did not need any specific treatment. Her second pregnancy was complicated by hypertension and gestational diabetes, and she has taken medication to control her blood pressure since then.

Over time, Jodie has become increasingly aware of her enlarged kidneys. She stopped playing netball in her mid-30s because of concern about causing them damage during heavy contact. Working on her hobby farm where her family breeds sheep, she instinctively protects her abdomen when working with stock to make sure she is not kicked. Any pressure on her kidneys, for example having an ultrasound, is very uncomfortable and she avoids it if possible.

Jodie’s renal function is still relatively good, at about 60% of the normal level, and it has been stable for some years. She has recently had two urinary tract infections, which is unusual for her. They could be a consequence of PKD, so her medical team has armed her with a pathology form and an antibiotic prescription to ensure she can diagnose and treat infections as quickly as possible if they recur. She has no other symptoms of ADPKD.

Family and friends
Having children herself was not a difficult decision for Jodie, as she saw her father live a full and rewarding life without major health problems until he was older. “I don’t really want this disease passed on, but with every generation we are getting more information and better treatment,” she says.

“I only have a few close friends who are aware I have PKD,” she says. “I have a disease, but I’m not sick.”

Similarly, there is little discussion among the family even when she and her brothers get together. “We don’t have ‘wallowing’ parties, we don’t talk about kidneys. We just get on with it.”
WHEN I WAS OFFERED A PLACE IN A TRIAL, AND I THOUGHT, ‘WHAT DO I HAVE TO LOSE?’ IT MIGHT NOT BE SOMETHING THAT ASSISTS ME, BUT IT COULD HELP THE NEXT GENERATION, INCLUDING MY BOYS IF THEY HAVE ADPKD.
"I think the world is a wonderful place and life is what you make it."
The top’s down on the Ford Mustang convertible driving America’s iconic Route 66 from Los Angeles to Chicago, and long purple hair is flying behind the Australian squeezed into the back seat. The proud owner of the hair is Julie, who can usually be found living quietly in a small town a little north of Adelaide.

Julie has autosomal dominant polycystic kidney disease (ADPKD), so the cramped back seat wasn’t the most comfortable way to travel. “I think the world is a wonderful place and life is what you make it,” she says. “But sometimes you discover the rules that you think have been set for your life haven’t really been set at all.”
**PKD in the family**

Julie’s grandmother, mother, aunt and son also inherited the condition. Her grandmother started peritoneal dialysis as her kidneys started to fail and about 10 years later had a kidney transplant. The transplant was a success and she lived over another 20 years, into her 80s. “Nana did really well,” Julie says.

Her mother’s course was a little more difficult. She, too, had a transplant, using an unmatched kidney donated by Julie’s father. The transplant performed well for about five years before it failed and her mother moved on to dialysis for a further four years, but dialysis had its complications. Her mother’s sister passed away from a brain aneurysm related to PKD at the young age of 38.

Julie was diagnosed with PKD when she had an ultrasound of her kidneys at the age of 12, but her two brothers are free of the condition. “I started thinking immediately, ‘That’s it, I’m going to die, I’m not going to make 30,’” she says. “So, I lived fairly hard, making the most of it. It’s not nice watching your Nana and your Mum going through what I saw, and thinking, ‘Well, that’s what I have to look forward to.’”

In fact, Julie is now just over 50 and has already beaten her target by 20 years. Until the last 10 years or so she has generally been well, but is now working with her medical team on managing the early signs of progressive kidney damage as well as some difficult symptoms. “I see the changes to my body, and it’s like looking in the mirror and seeing my mother,” she says. “But Nana and Mum were both sicker than me at a younger age.”

**Having a child**

Julie and her family were convinced that PKD occurred only in girls in their family, as there hadn’t been a case in a male for more than 60 years. When she thought of having children herself, she briefly considered IVF so she could choose to only have boys. “I thought it would keep us safe,” she says. In fact, PKD does not discriminate between males and females, and there is a 50/50 chance that a mother or father with the mutation will pass it on to a child.

“You have to go through the discussion with your husband, saying it’s hereditary, I might get sick. When you are young, it’s quite embarrassing to have to admit there’s something wrong with you,” she says.

Julie had two sons and one was unlucky enough to inherit PKD. “When I was diagnosed my Mum apologised to me so many times. ‘I’m so sorry,’ she said. Now that my son has been diagnosed, I know why she was like that. It is guilt, absolutely.” Julie’s challenges in life have been compounded by a brain injury her son suffered when he was just two years old, leaving him with special needs to the present day.

**A changing body**

Julie says that at the age of 40 she was still “hot”, happy about her flat stomach, good health and ability to live life to the full. Since then the symptoms of PKD, which can vary widely from one person to the next, have started to slowly accumulate.
“Underneath this dress I look like I’m seven months pregnant,” she says. One kidney is 26 cm long, and together they weigh about 6 kg.

“I’ve had to learn to dress for it, and it affects my self-esteem. My Nanna’s stomach never got big and I always wanted to be little like her, but I came out like Mum who also had very enlarged kidneys. Why did I have to get the big stomach? Ageing is bad enough, but having this as well is a pain.”

Her purple hair, matching nails and multi-coloured tattoos help her to think about other aspects of her body, and divert people’s attention from her less-than-flat stomach.

“As a woman, I am judged,” she says. “I’ve been asked if I’m pregnant, and I’m judged for seeming to be overweight. I can’t wear a sign saying, ‘This is because I have PKD.’

She also has high blood pressure, a common result of PKD, which now requires medication. A stroke at the age of 27, which initially left her with vision problems in one eye, was probably related to the condition as well. Her stomach is compressed by her kidneys, so she can eat only very small meals and sometimes vomits afterwards.

Julie had been working for many years in disability services, initially with special needs children in schools but more recently in adult rehabilitation. She left work last year on the advice of her GP because of difficulties with her kidney function, which now requires medication.

Discomfort from her kidneys, sometimes manifesting as outright pain, is a growing concern. “They ache, and they rub on my ribs,” she says. “We looked at the possibility of draining the cysts but they are multiple, so it wasn’t possible.”

Sitting immobile in an airline seat to America, and being cramped in the back seat of the Mustang convertible, was extremely uncomfortable. She desperately wants to go overseas again, but the prospect of any long trip is daunting. “And I’ve never been able to get life insurance or travel insurance,” Julie says.

**Health care**

When Julie was a teenager her medical care was overseen by her mother’s nephrologist, but as a young adult she found her own kidney specialist who has guided her ever since. Initially she saw her doctor for monitoring every few years, then annually, then six-monthly, and now every three months.

Her medications include treatment for high blood pressure and high cholesterol level, occasional pain relief, and she is involved in a clinical trial of a potential new treatment. Julie agreed to join the trial about seven years ago. “I was a great candidate because I had normal kidney function but was well into the disease,” she says.

Working closely with her medical team has been vital for Julie in managing her PKD. She is full of praise for the care provided by the hospital, especially her nephrologist and specialist nurses. “The staff I deal with have never made me feel like a number or just another patient,” she says. “My nephrologist considers my point of view and discusses the benefits and possible complications of treatment with me.”

PKD is not a common condition and people living with it tend to become experts in their own disease. Julie has found it a little difficult to identify a GP with an understanding of her medical issues, but thinks she has now succeeded. Her GP and nephrologist collaborate in providing comprehensive care, both for her PKD and the usual medical issues of mid-life.

**Looking forwards**

Although her kidney function has been completely normal for many years, recent tests have shown some deterioration and discussions about dialysis and transplantation have a heightened focus. “I wasn’t surprised. I’ve spent most of my life expecting it,” Julie says.

One possible benefit of transplantation may be the removal of one or both of her enlarged, problematic cystic kidneys, reducing her constant discomfort and restoring the body shape of her early 40s. “I like the idea of a transplant and see it as a natural progression of PKD. My mother’s experience wasn’t what I would call a huge success, but my Nana’s was brilliant,” she says. “It’s all about our own choices. It’s our body so ultimately we have to take responsibility for that.

“I can’t stress enough the importance of being able to talk frankly with the treating doctor and having the type of good and honest relationship that I’m lucky enough to have. Doctors are different these days compared to when my Mum and Nana were being treated – they are people you can have a discussion with.”

Julie’s message for someone just diagnosed with PKD is to find a medical team that you are comfortable with. “You need to be comfortable, because it’s such an intimate disease,” she says. “If you have trouble finding that, then look at the networks that have been formed for people with PKD where you can ask other people questions.

“And remember that it’s not a life sentence. I have purple hair, and I’m an optimist.”
Laura has a role model for responding to the challenges of living with a chronic illness. Her mother had autosomal dominant polycystic kidney disease (ADPKD) and dealt with the illness with grace and dignity.

Laura, 29, also has the condition and is following her mother’s example, leading a productive and active professional and personal life while working hard to maintain her health.

“My mum was the strongest person I have ever seen,” Laura says. “She never said, I can’t do this because of my disease or because I’m unwell.”

A social worker who is passionate about her contribution to disability care in Melbourne, Laura was aware from the age of five or six that she, too, could have this inherited condition. “It has always been part of my life,” she says.

Laura had a medical check-up before starting a volunteer role when she was 21 and chose to check whether she had PKD too. Until that time there was little point in pursuing the question, as she was healthy, had no symptoms, and knew that problems with kidney function did not usually emerge until a little later in life.
Laura's aunt, as well as her mother, inherited the condition. “For us, having a mother with PKD was normal,” she says. “We never knew anything else. Mum was quite unwell for a lot of my life, and as the oldest child I had to pick up things for the family as I watched her on her journey.”

Her mother’s path became complicated when she developed breast cancer and was treated with aggressive chemotherapy which may have hastened the progress of her kidney disease. She started peritoneal dialysis when she was about 40 and then progressed to haemodialysis after about seven years. A kidney transplant was not successful, and she passed away. Tragically, her aunt with PKD had died six weeks earlier in the same hospital.

Laura says she was pragmatic when she was diagnosed with the disease herself, as she was not surprised and wanted to focus on how to live her life. “The hardest thing was telling Mum, so I kept it a secret for quite a while. But my father found out and then I had to tell her.”

She has stayed healthy, active and positive in the eight years since her diagnosis. She takes two medications for high blood pressure, a common result of the condition, but has no symptoms and has never been admitted to hospital even though her kidney function is currently about 20% of the healthy level. “I’ve been really lucky,” Laura says.

Laura was monitored by kidney specialists, initially every 12 to 18 months. About two years ago she agreed to participate in a clinical trial of a medication in autosomal dominant PKD. “There was very little the clinic could offer until the trial came up,” she says.

“Kidney health has always been a big thing for me, growing up with it in my life. After watching my mother’s progress, if there was a way I could delay that for myself, or support any advancement in the field, I knew it would be a good thing.”

Close monitoring and regular assessment as a participant in the trial has been a positive experience for Laura. She has also benefited from having a GP who understands her PKD, is committed to her care, and works with her kidney specialists to keep her well.

Knowledge is power!

“ ’I’m inquisitive, so I ask questions,” Laura says. “I search for information if I’m not offered it, and that way I can feel in control of my life.”

Laura has found information sessions organised by the Australian PKD Foundation (pkdaustralia.org) helpful for both her and her partner, and she also accesses the resources offered by the American PKD Foundation (www.pkdcure.org) including regular webinars.

She has been an active fundraiser for Kidney Health Australia, attracting up to $3000 in sponsorship in walks and other events. “My friends know about PKD and my family, and they want to help,” she says.

Laura would like to see more recognition of the fact that kidney disease can affect young people, not just older people. “Young people need support too,” she says. “It’s also important to recognise that everyone is an individual, and affected in their own way by disease.”

And her advice for someone who has recently been diagnosed with PKD? “It sounds like a cliché, but the more you are aware, the more you can prepare!”

Living with PKD

Laura is about to travel to the United States to celebrate her 30th birthday, including time exploring New Orleans. The trip symbolises the fact that PKD is not imposing any limits on her life. “I play netball once a week, I go to the gym twice a week, and I like swimming,” she says. “My mum was a home economics teacher and an amazing cook. Food is a big thing for
Laura, and I still enjoy it.”

Although she’s careful about “burdening” other people with her condition, Laura told her partner about PKD very early in their relationship. Now almost 30, she is facing difficult decisions about the possibility of having children. “It’s complicated,” she says. The issues include her ability to carry a pregnancy, the risks of her child inheriting PKD, and the very difficult dilemmas of pursuing IVF and testing whether or not an embryo is carrying the gene. “If I had a child with PKD, I would hope that science will have advanced enough to support people with the condition,” she says.

**Dialysis and transplantation**

A graph printed by her GP shows a steady reduction in Laura’s kidney function, measured by glomerular filtration rate (GFR). It was about 60% of the normal value when she was first diagnosed eight years ago but is now hovering around 20%. “I think that’s quite a decline,” she says.

Her medical team tells Laura that she will probably need dialysis when her kidney function drops to below about 15%, but the path is unpredictable. “I’m not scared of dialysis because it’s such an ingrained part of my life, having seen my mum go through it,” she says. “But not knowing when this might happen is a problem. I would prefer to know when I’ll need it, so I can plan my life.”

And transplantation? “I won’t accept a living donor from a family member,” she says. “It’s too much pressure on them, and I saw my mum have an unsuccessful transplant. I would hate that to happen if someone had given me a kidney.”

Laura acknowledges that PKD is a significant part of her family’s history, and her own life. “But I don’t feel it defines me,” she says. “There are so many other important things that take precedence, like a strong relationship, a fantastic family, and being supportive of them. I’m now the matriarch in my family – it’s a role you have to accept.”

“MY MUM WAS THE STRONGEST PERSON I HAVE EVER SEEN. SHE NEVER SAID, I CAN’T DO THIS BECAUSE OF MY DISEASE.”

“THERE ARE SO MANY OTHER IMPORTANT THINGS THAT TAKE PRECEDENCE, LIKE A STRONG RELATIONSHIP, A FANTASTIC FAMILY, AND BEING SUPPORTIVE OF THEM. I’M NOW THE MATRIARCH IN MY FAMILY – IT’S A ROLE YOU HAVE TO ACCEPT.”
When Jodie recently had a small skin cancer removed from her face, covered by a surgical dressing, people were concerned, sympathetic and offered to help. It was a different story when, 12 years ago, she told her friends she had autosomal dominant polycystic kidney disease (ADPKD). Some said she was simply making it up because she looked healthy, the condition was ‘invisible’, and kidney disease was ‘for old people’.

“You have to put up with a lot of judgment and misunderstanding, because no-one can see your kidneys,” she says.

Jodie is now 33 and telling her story in a tranquil park in a small town just outside Melbourne. She explains that, as a 21-year-old, she had a new diagnosis of ADPKD but very little information to guide her. She was rejected for a time by her friends who didn’t accept that she was unwell. Years later, her marriage suffered when difficult decisions about having children had to be addressed.

Her family, too, took time to come to terms with the diagnosis. Because ADPKD is inherited, there’s usually a parent, grandparent, aunt or uncle in the family who has been diagnosed. In an unusual twist, Jodie’s father was found to have the condition about seven years after she had been diagnosed, when he had some investigations for the high blood pressure which had been present all his adult life.

“He always said I’d inherited his high blood pressure, but he was shocked to find out he also had ADPKD and I’d inherited that as well,” Jodie says. (High blood pressure is a common result of ADPKD). “When he put two and two together he had a devastated look on his face, realising that he had passed it on, and it was a very uncomfortable topic for a long time.”

In recent years she and her father have strengthened their relationship based on the challenges they have in common, and are both dealing with the fact that their kidney function is slowly declining.

“Dad’s kidneys and mine are working at about the same level. That’s a bit of a worry for me considering that I’m so much younger,” Jodie says.

Although ADPKD is rare, Jodie discovered a former co-worker had the condition. “It had been diagnosed in her family for generations, so she knew everything about it,” she says. “It was very different for me, because I was the first person diagnosed in my family and I started out knowing nothing.”
Pathway to diagnosis and care

Some practical sessions at university first alerted Jodie to her medical problems. She volunteered to be the ‘guinea pig’ as students explored ECGs and blood pressure.

At the age of 18 her blood pressure was 173/120 mmHg compared to a normal level of 120/80 mmHg or less. She started treatment for her high blood pressure immediately after being sent to the campus doctors, but discovering ADPKD took another three years.

Jodie had persistent back pain after a fall at work and her GP ordered an ultrasound a few weeks later to check for any damage. “The radiographer doing the scan noticed something and said, ‘Can I have your consent to ultrasound your kidneys, even though it’s not part of the request?’ And then she said, ‘You need to wait right here until I’ve processed the scans and then take them straight to your GP’.”

Jodie’s GP instantly recognised the distinctive image of her polycystic kidneys: they were about 40% larger than normal and contained multiple clear cysts, up to 5 cm in diameter.

Although being told she had ADPKD, Jodie received very little information about the implications or what to do next. Referred to nephrology outpatients services, she felt she “bounced around” the health service, spent hours in clinic waiting rooms, saw different doctors at every visit, and still learnt very little about her situation.

Some of the information was very negative. Jodie was told, ‘You’re quite young to have advanced disease. We think you’ve got until your 30s until you’ll be on dialysis, so anything you’ve got to do, do it right now’.

“They focussed on my blood pressure and ordered some tests, but no-one had explained what it meant. I thought, they don’t seem to care so much or be worried, so why should I take this seriously?”

“MY FAMILY AND FRIENDS WERE WORRIED ABOUT SOMETHING BAD HAPPENING IN THE TRIAL BUT I SAID, WHAT IF SOMETHING GOOD HAPPENS?”
Access to expert care
As a young woman building her life, Jodie decided that ADPKD, doctors and hospital clinics were not for her. She ignored her condition for six or seven years until a GP was alarmed by the results of some routine blood tests showing that her kidney function had dropped to about half the healthy level, and her blood pressure was high.

“I asked, is that bad? And he said, ‘Well, you’re 30 years old, so it’s not great’. I had to stop pretending I didn’t have it and decided to take control,” Jodie says. “I arranged to be seen by the nephrology clinic at the hospital. I knew about its good reputation and I also knew about clinical trials.”

On her first assessment in the clinic she was asked to consider entering a clinical trial evaluating a new treatment for PKD.

“My family and friends were worried about something bad happening in the trial but I said, what if something good happens? I wasn’t doing it for me. I know where I’m headed, but what about people in the future? Regardless whether the results are positive or negative, we need the data.”

Participating in a clinical trial means that Jodie has been reviewed every month by the clinic, now for a total of about three years, and she also talks regularly to the trial coordinator by phone to discuss any difficulties. “Even without being in the trial I know I still would have had great care,” she says.

A healthy life
Jodie’s life and medical care is complicated by several other medical conditions including lifelong severe eczema (which is now well-treated and under control), food intolerances and hypermobility of her joints. She has had difficulty with her heart rhythm, as her heart rate is often over 100 beats a minute but at times is too slow.

Her reduced kidney function limits the range of medications that she can take, so her doctors have been working together to provide the best possible care.

Jodie walks around her home town every day, wears a FitBit to monitor her heart rate, and will shortly get a new device which will also record her blood pressure.

Her enlarged kidneys sometimes cause a little abdominal pain, but it is not a major problem. She’s had to limit her enthusiasm for kickboxing and paint ball, following her doctors’ concern that they could be dangerous for her vulnerable kidneys.

Having grown up in a “family of foodies” Jodie loves cooking but her diet is limited by several food intolerances, except for a planned monthly indulgence. She follows advice from a clinic dietitian to limit her salt and protein intake.

Jodie takes two medications to control her blood pressure and heart rate, and a statin to lower her cholesterol, together reducing the risk of cardiovascular disease that accompanies ADPKD. Fish oil and glucosamine help control joint pain caused by hypermobility syndrome. She also takes three tablets of the trial medication in the morning and one in the afternoon.

A daily tablet box which she fills each month helps ensure she takes her daily quota of medicines. Alarms on her phone provide reminders, but Jodie has found they are easy to dismiss without actually taking the tablets.

She finds her daily pill box valuable, because it provides a very obvious visual reminder about whether or not she has taken her prescribed doses.

Impact on work
Jodie worked full-time until about six months ago, having developed a successful management career in a major retail organisation. A very long commute, tiredness and her accumulating health problems meant she had to take a break, and she’s still considering her options.

Although earning a living is an increasing challenge, Jodie is pessimistic about the prospect of support from Centrelink. “When a 33 year old is able to walk into their office, it’s hard to convince them of the real disability of an invisible disease,” she says. “If I had kidney cancer rather than ADPKD, people would be much more likely to understand. Everyone’s heard about cancer, and knows how serious it can be.”

Looking forward
As her kidney function continues to slowly decline, Jodie knows that dialysis and transplantation may become a reality for her. “We don’t actually talk about it at the clinic, but for me I think it’s inevitable,” she says. “People in the future might have the option of an implantable artificial kidney, but we’re not there yet.”

In contrast to her earlier experiences, Jodie now has a strong and supportive network of friends and her family members have come to terms with the condition. She is full of praise for her healthcare team.

“I can’t fault them. I know I can have an honest discussion, and they are so supportive.”

For someone newly diagnosed with ADPKD, Jodie would advise them to talk to people, get to understand the condition, and have open and honest discussions with family, friends and healthcare professionals.

“And you don’t know how strong you are until you’re tested by life,” she says.

YOU HAVE TO PUT UP WITH A LOT OF JUDGMENT AND MISUNDERSTANDING, BECAUSE NO-ONE CAN SEE YOUR KIDNEYS.”

AND YOU DON’T KNOW HOW STRONG YOU ARE UNTIL YOU’RE TESTED BY LIFE.”
Melissa, now 47, was told she shouldn’t have children because she had autosomal dominant polycystic kidney disease (ADPKD). “Bad luck,” she told her doctor, an internationally-renowned kidney specialist, “I’m having three.” And she did.

Melissa is well and leads a busy life focussed on those three children, now aged 17, 14 and 10. She returned to full-time work after her husband had a major health scare seven years ago, she has just transitioned her parents into an aged care facility, and her office has recently moved to a new site, so there’s little time left to worry about her ADPKD.

With a background in marketing, Melissa works as an events organiser with a church organisation. She plans about 70 events a year, ranging from day-long training session for 20 people to major weekend meetings attended by more than 1,000.

“We have a lovely group of people and a very positive, supportive team culture in the office,” she says. “It’s a nice place to be.”
“I’ve been open with them, so it won’t be a surprise if I eventually become unwell. At the moment, though, I’m a picture of health and my children know that their grandmother with ADPKD is still with us.”
**PKD in the family**

There is a 50/50 chance that a child of someone with ADPKD will also have this inherited condition. Melissa’s grandfather, who she never knew, had the disease and died at the age of 52 because of its complications.

Her mother, now in her mid-70s, also has ADPKD. She received a transplant from her husband when she was aged 52. Melissa was a young adult at the time and recalls the stress of knowing that both her parents were on operating tables at the same time, one donating a kidney, the other receiving it.

The transplant went well, lasting about 20 years until Melissa’s mother needed a second procedure, this time receiving a kidney donated by her mother’s sister.

“Mum’s been going well and has managed to live independently until now,” Melissa says. “She’s not strong health-wise – she’s weak and frail – but her kidney is fine.”

Melissa’s sister, four years older, has ADPKD too. “She is struggling. She had a kidney transplant from her husband but it was rejected and a second transplant two years ago. The second one is now showing signs of rejection and she’s having treatment trying to control it.

**Melissa’s diagnosis**

Melissa found out she had ADPKD at the age of about 14. Her family had agreed to participate in a study of ADPKD genetics so she had some blood tests, and then had an ultrasound which revealed the typical cysts.

As a child she didn’t know that her grandfather had the condition. Although the family knew about her mother having PKD, it was not an obvious feature of the family’s life.

“PKD was not a ‘presence’ in my family,” Melissa says. “When I found out I had it, I was never really concerned. Even when I started taking blood pressure medication and regularly seeing a nephrologist, I didn’t give it any weight because at that point in time I hadn’t seen any detrimental effects. Mum didn’t seem to be sick, so I didn’t think it would make any difference to me.”

**Having children**

As Melissa learnt more about her family’s history she realised that her mother’s journey had been smoother than her grandfather’s. She hoped that she, and her children if they developed the disease, would benefit from further advances in treatment over the years.

“The disease didn’t come into any decision-making process about having children. I knew PKD was inherited, but I didn’t think it would be a major issue, because I still hadn’t seen any consequences for the people around me,” she says.

Melissa briefly explored genetic testing for her children to see if they carried the ADPKD gene, but discovered the out-of-pocket cost would amount to many thousands of dollars. “I’ve discussed it with my nephrologist, and we have decided to wait. When we think it’s right for each child, we will do an ultrasound. He doesn’t think it’s necessary to know at this stage as there is still little we can do, and there are consequences like insurance coverage once you know you have a pre-existing disease.

“All my children know about the family history,” she says. “I’ve been open with them, so it won’t be a surprise if I eventually become unwell. At the moment, though, I’m a picture of health and my children know that their grandmother with ADPKD is still with us.”

Melissa has taken medication for high blood pressure associated with ADPKD since she was about 18 and it is well-controlled. She has occasional urinary tract infections and back pain also caused by the condition, and has had several episodes of a kidney cyst bursting or bleeding. “It’s like having the flu for a couple of days when that happens, but I’ve been very lucky and haven’t had any major problems at all,” she says.

Melissa says her medical team are “amazing” and have done everything possible to care for her. The family’s GP, who still looks after her parents, has a good understanding of ADPKD and is an important member of the team.

**The future**

Melissa has watched as her mother and sister have had kidney transplants, and knows she may need one in the future. Her husband has heart disease and is not a candidate for donation. The team also made it clear they would not use anyone who is the parent of dependent children, which has ruled out some friends who had initially considered donation. “So, I don’t really have a lot of options,” she says.

Her kidney function is not yet at the level where dialysis or transplantation are essential. When the time comes, if she is unable to find a live donor she will start dialysis and be placed on a transplant waiting list, perhaps for two years before she can have the procedure.

“I try not to worry, and I regard dialysis and transplantation as inevitable,” she says. “I have faith in God. I know that just because I have faith, doesn’t mean bad things can’t happen, but I feel a sense of comfort. I worry more about the children than about myself, but as every year passes they are older and better able to cope.”

Melissa has seen the range of possible outcomes in her family, from the early death of her grandfather to successful transplants for her mother, and greater challenges with transplants in her sister.

“I relate my story more to my mum. Looking down the barrel of the future, I look at her and think, that’s what I’m going to be like. But I’m a generation younger, my medical care has been better, and I’m in better health than she was at my age, so I think I will be healthier at the time of a transplant.

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