

By Kate Legge Photography Julian Kingma

TAKE THE TEST OR HOPE FOR THE BEST?

They are cousins, but you wouldn't pick it. Simone Busija (opposite) is dark and quietly spoken; Kim Eishold is blonde and cheerfully loud. Simone lives with her husband and two sons in a renovated Victorian weatherboard to the west of Melbourne where the tidy interior suggests a super-organised working mother. Winter rain lashes the windows when I visit to talk about the rare genetic glitch that has struck this family.

Kim, by contrast, is a self-confessed gypsy. Single, without so much as a pet to tie her down, she's taken leave from her job to travel with her mother around Australia towing a caravan behind them. She's in a bikini the day we speak, bathing in the thermal springs of Mataranka, 400km southeast of Darwin, where she's landed a kitchen job out of the blue.

These women couldn't be less alike but they share a common sadness that marks their personal history. Their fathers, who were brothers, both died from stomach cancer in their mid-50s. Graeme Eishold, Simone's father,

and Peter Eishold, Kim's dad, did not know they carried the CDH1 gene mutation that predisposed them to a terminal illness. Their daughters, on the other hand, live in an era of genetic mapping; they don't have to live with the unknown or leave their health to fate. They each have a 50 per cent chance of having inherited the mutant gene that inflates their chances of a particular type of stomach cancer, but with the aid of a simple blood test – and drastic preventive surgery – they can beat the odds.

The precise science of this genetic defect on chromosome 16 determines the statistical likelihood of disease but the trickier uncertainties of personality and circumstance shape how each individual responds. Survivor or *pre-vivor*? Take the test, or leave well alone and hope for the best? Kim and Simone are dealing with the card that has been dealt them in very different ways.

A keen eye and a good memory for family history were once the only tools we had to untangle the crimson knot of bloodlines. Hours after the birth of new kin, relatives would gather around the crib playing I Spy to spot

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physical attributes that bind the members of a clan together. As a baby's personality emerged, other hereditary comparisons were drawn to link temperament and talents with aunts, uncles and grandparents. If fate was written in our genes, we had no way of finding out.

Today's parents are on alert for genetic abnormalities from conception. Tests for conditions such as Down syndrome or spina bifida are conducted in utero and resume after delivery with the heel-prick of newborns screening for cystic fibrosis and other congenital disorders. The Human Genome Project unleashed a revolution, with thousands of different tests now available in Australia.

As the menu of tests expands every year, so does the volume of patients. Surveys by the Royal College of Pathologists of Australasia found that almost 580,000 medical genetic tests were performed in 2011 – a leap of 280 per cent compared with five years earlier. Most common are the simpler, cheaper tests for iron overload, blood clot disorders and cystic fibrosis. Only 15 tests are covered by Medicare. Genetic pathologists are increasing pressure for a national response to boost staff and overhaul funding as our capacity to sequence genes races forward, encouraging public awareness of genetic predisposition to disease.

Angelina Jolie's double mastectomy this year alerted women to the radical preventive treatment available for carriers of the BRCA1 and BRCA2 mutant genes associated with breast and ovarian cancer. Other organs being targeted in therapy to reduce cancer risks include the colon, uterus and stomach. Genetic counselling services have burgeoned in lock-step with this advancing frontier of research but family members must first connect the dots.

The first hint of trouble in the Eishold family was shrugged off as misfortune. Kim and Simone's uncle, Geoff Eishold, was 30 when he began to feel not quite right. One of five siblings who shared a hardy work ethic and robust health, he put it down to stress. It was 1980 and DNA sequencing was in its infancy. "I started to lose a bit of weight," he recalls of what prompted him to visit the old-fashioned GP who had looked after his parents, Bill and Gwen Eishold, for years. The doctor did a couple of tests that were inconclusive. It's all in your mind, Geoff was told. "I didn't think of getting a second opinion." His weight continued to drop. One night he had a heavy drinking session with an old friend from interstate

and his younger brother Peter. Three days later he began vomiting blood. "Not a bit... a lot." His wife rushed him to hospital where a gastroenterologist diagnosed stomach cancer. Within days he'd undergone surgery to remove his stomach and spleen.

Almost 20 years later, Geoff's oldest brother, Graeme – Simone's dad – was being treated for bone cancer when he started to suffer reflux. An endoscopy confirmed he too had stomach cancer, but the disease was so advanced that nothing could save him. He died in 2000 aged 56, on the cusp of retirement. Then, when brother Peter – Kim's dad – was given weeks to live after being diagnosed with late-stage stomach cancer in 2005, the notion that this streak of bad luck was coincidence became too farfetched.

A fourth brother, Tony, was telling someone at a party what had happened to his siblings when "the penny dropped". After some ama-



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teur detective work he got wind of a killer gene peculiar to a handful of families around the world. The mutation was first publicised in a 1998 study by New Zealand scientist Professor Parry Guilford, who linked the genetic error to multiple cases of gastric cancer in three Maori families. But it took several years for genetic testing to become part of clinical programs that assess and manage the risk of various cancers. Around 100 kindred groups in the world have now tested positive for the CDH1 flaw.

Tony Eishold sounded the alarm within his clan but it was Geoff Eishold who took on the role of family guinea pig. He was the first to submit to DNA testing in 2008 through the Familial Cancer Centre at the Royal Melbourne Hospital. Confirmed as a carrier, he became chief bearer of bad news. But at least there was cause for optimism. Tests and treatment could save younger generations from the cancer that had picked off their uncles. He contacted his two surviving siblings, Tony and sister Jan, as well as their elderly father Bill, and every member of the extended family.

Deciding whether or not to get tested for CDH1 is a tough first threshold to cross. Once you know you carry a gene that gives you an 83 per cent chance of stomach cancer, the pressure to take preventive action is immense. Tony has never been tested. Jan tested positive, but delayed surgery until an endoscopy traced evidence of stomach cancer. Their father Bill, who sits at the top of the family tree, was found to carry the gene that has been passed around. He is 96, sound of body, with no hint of the disease that has devastated his offspring.

Kim Eishold, 35, is a fatalist. She knows about the family gene but she's decided not to be tested for it. "I'm one of those people who put my head in the sand. I only want to think about the good stuff. I don't want to know. If you find out you've got the gene you'll be caught up in so much agonising," she says. "I don't know if I want to change my whole life."

Her philosophy revolves around the theory that "everyone's got a number and when you're time's up, it's up". What will be, will be. "I don't have kids so I don't have to be around for children when they grow up. If something happens to me it doesn't matter. I'm different to everyone else, I'm the black sheep," she laughs.

Her brother Paul Eishold, 32, has three young children and he opted to have the gene test. "I'd rather know," he says of the discovery

that he carries the mutant gene. But he's not ready to undergo surgery. "It's a pretty big, life-changing thing. While I'm younger I want to enjoy my lifestyle for a while yet." Separated from his wife, who wanted him to have the surgery, he now faces most urging from his uncle Geoff, who is living proof that being young is no insurance policy. "I know it has to be done," Paul says of the surgery, but he prevaricates, explaining that recent back surgery for a herniated disc took priority.

His uncle nags, anxious at Paul's delay. "He keeps putting it off. He's always got something else to do," Geoff sighs. "You can't put these things off. It's not an old person's disease. I wish he'd have it done. And I wish Kim would have the test. You can only keep telling them. They've seen their father die," he says, exasperated.

Paul acknowledges his father Peter's early death casts a shadow. "I don't want my kids not to have their dad." He remembers the symptoms that plagued his father. "He let it go for a while. I'd go straight away," he says. The surgeon



Genetic lottery: from left, Tony, Peter, Geoff Eishold (sons), Bill Eishold (dad), Paul McHardie (grandson), Jan Lawrence (daughter), Graeme Eishold (son). Opposite page, Kim Eishold

has told him the sooner he acts the better. Relying on regular endoscopies is a poor substitute since these probes for cancer can only take a snapshot of a section of this large internal organ. His aunt Jan, Geoff's sister, opted initially for this less interventionist monitoring after finding she also carries the gene. But once cancer was detected she had a total gastrectomy. Since the operation Jan, 67, has suffered unusually severe repercussions. "That's one of the reasons I'm reluctant to do it now, because of the impact it's had on her," Paul says.

His and Kim's response to their father's genetic legacy may be due in part to a personality trait they inherited from him. Peter Eishold was a children's clown who didn't fuss overly when he began experiencing heartburn and reflux. Misdiagnosis from several doctors didn't help. "He'd been sick for a long time," Kim recalls of his sudden decline. "Dad and I were pretty close. I got him. He got me," she says, linking "my ignoring self" to that side of him.



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This equanimity contrasts starkly with the proactive stance adopted by their cousin Simone Busija, 41, whose father Graeme was the first to die from gastric cancer. She went for the test shortly after Uncle Geoff briefed the family on CDH1 and its potentially fatal impact. During the eight-week interlude waiting for results she thought carefully about what she would do if she had the mutant gene. "I was similar to my father so I was pretty sure I was going to have it," she recalls.

Her husband accompanied her to the Familial Cancer Centre in March 2011 to learn she was indeed positive. "I was very emotional on the day. I could tell immediately by the body language of the counsellor. I phoned Mum and she burst into tears," she recalls. Later, a close friend told her: "They've saved your life."

Although an endoscopy couldn't detect cancer, her consulting surgeon strongly recommended surgery to remove the stomach because of her age and the 83 per cent risk of gastric cancer associated with the mutant gene. Like Angelina Jolie, she wanted to do all she could to be around for her two sons. Living without a stomach was a small sacrifice for the promise of mothering her brood.

Before undergoing six hours of keyhole surgery in November 2011 she deliberately put on 7kg to cushion her body against likely weight loss during recovery. Having no stomach means she doesn't feel hungry or full. Eating must be carefully controlled because the body doesn't digest or absorb proteins in the normal way. Her oesophagus is joined to her small intestine, shrinking dramatically the area for food to travel through. "The hardest thing is getting the body and the brain into synch," she laughs.

Fielding curious questions when people discover she has no stomach also requires diplomacy. "Sometimes they ask whether I have to eat at all," she says. Breakfast is a piece of toast or a small bowl of cereal; mid-morning she often has a muesli bar, followed by some soup or half a sandwich for lunch; then a small serve of whatever she cooks for the family dinner. Tasting rich foods such as a creamy pasta sauce is a game of trial and error, particularly if she adds a glass of wine into the equation. "People often forget I'm not the same person," she says of her diminished appetite. "I definitely can't eat a meal and a dessert." Her mantra is "eat slowly and small amounts" since large amounts of food accompanied by too much liquid leads to what's known as "dumping" or episodes of

light-headedness, nausea and fatigue. When this happens, rest is the only cure.

Since her operation she has returned to her sales job part-time and become a vocal advocate for a support network called No Stomach for Cancer. She sometimes mourns the loss of her appetite and the freedom to binge, but the surgeon's discovery of a minute cancer in her stomach validates daily her preventive strike. The CDH1 gene mutation can cause breast and colon cancer, too, but the risks are much lower. Simone is committed to annual check-ups for both. "How many body parts do you want to lose?" she jokes.

If Simone is a picture of health, her aunty Jan is stick-thin, and takes anti-depressants to cope with myriad post-operative complications that make her feel unwell five days out of seven. "I was such a party person," Jan sighs. Her operation went smoothly but she suffers constant reflux and "plumbing problems" requiring

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numerous procedures to stretch her oesophagus. "It has been so life-changing," she says despairingly, even as she concedes her outlook is preferable to the death sentence both her brothers faced. "After seeing the others suffer, and they really did, I couldn't have gone like the boys." She lives in Bairnsdale, East Gippsland, close to their father, Bill, who passed on the mutant gene. "He's as good as gold, sharp as a tack."

Jan worries that her experience has discouraged Peter's children, Paul and Kim, from facing the risks inherent in the family's genetic footprint. Her son Warren had his stomach removed shortly after Simone's surgery. He's had problems but nothing to rival his mother.

Bound together by an inherited trait, members of the family deal with it as they see fit. Jan and Geoff had no choice. Both got cancer. "It must be a huge thing to have your stomach out or your breast off when you're not sick. I'm not sure I would have done it if I hadn't been ill. I just don't know," Geoff muses sympathetically of the stand taken by the youngest of the five siblings, Tony, 52, who refuses the test. "He won't have it done," shrugs Jan.

"I don't believe I've got the gene," Tony tells me when I ask why he's resisting. "I'm the same ilk as Kim. If I've got it, I've got it. I've seen everything I've wanted to see, done everything I've wanted to do. I just don't want to go through what Jan has endured. She's in terrible shape. Simone is younger and more resilient."

Tony has a daughter and a son who may carry the genetic error. "We should all get it checked," he agrees. "Jan's been in my kids' ear about it," he says, wavering while we talk. One minute he swears he's as "fit as a Mallee bull", the next he remembers how Simone's father Graeme died when he was 56. "I don't rule it out," he says. "But I'd rather die on my feet than live on my knees... It's a laid-back attitude but it's got me through so far." The clock ticks as he ponders his quandary.

There is no accounting for why one relative opts to know while another turns a deaf ear. According to Professor Graeme Suthers, who chairs the genetics advisory committee for the Royal College of Pathologists of Australasia, only about 40 per cent of at-risk relatives turn up for genetic testing and the rest may put it off for years. "It can be eight or 10 years later that someone rings up and says, 'I think I might come and talk to you about this now.' Sadly, sometimes we also get calls from their doctors,

who are treating them for cancer.” He suspects “busyness” and “fear” are the main roadblocks.

“These are the tough choices people have to face,” says genetic specialist Ingrid Winship, who counselled many of the family members involved in the pioneering New Zealand study that discovered CDH1. Now executive director of research at Melbourne Health and Professor of Clinical Genetics at the University of Melbourne, she says every sibling weighs the risks and benefits on a personalised set of scales that reflect “life experience, exposure to the genetic disorder, age, whether you’re partnered or not, whether you have children... these all figure in the decision-making”. She describes stomach removal as “significant surgery”.

Simone agrees. Lucky enough to enjoy a fairly normal existence, she says it has influenced her philosophy in profound ways. Not only has she made peace with her father’s death, because it wasn’t in vain; she has also gained a new perspective on the quotidian bugbears that used to stress her. She wants to be a role model for her sons in case they have inherited this flaw.

Last year they accompanied her on a fund-

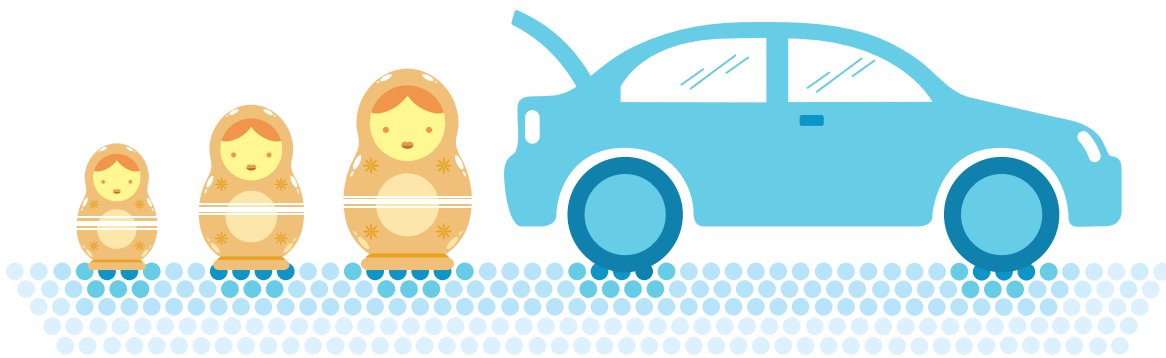


Preventive strike:
Jan, with her father
Bill. Opposite, Paul
Eishold, who had
the gene test but not
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raising walk to promote research and awareness and will do so again in November. “I want to be a pioneer for them. I want them to see that Mum has gone on to live a normal life, and who knows the medical technology that will be available when they grow up?” Her sons, aged nine and three, have a 50 per cent chance of inheriting the genetic error from her. She plans to get them tested in their teens. “They are not at risk [of cancer] until they are older, so it is too early to worry about it,” she says.

Winship acknowledges that biotechnology breakthroughs to switch off mutant genes are not fanciful science fiction, even though targeted genetic therapy is a way off yet.

In the meantime, the Eisholds manage the trick their ancestry has played on their lives as best they can. Uncle Geoff wonders whether he would have fathered offspring if he’d known then the secrets of a gene pool that science has since revealed. His two daughters have escaped the curse. “They’re so lucky,” he says. That’s the roll of the genetic dice. The blessing for all is that at least they now get a heads-up for a square fight. ●



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