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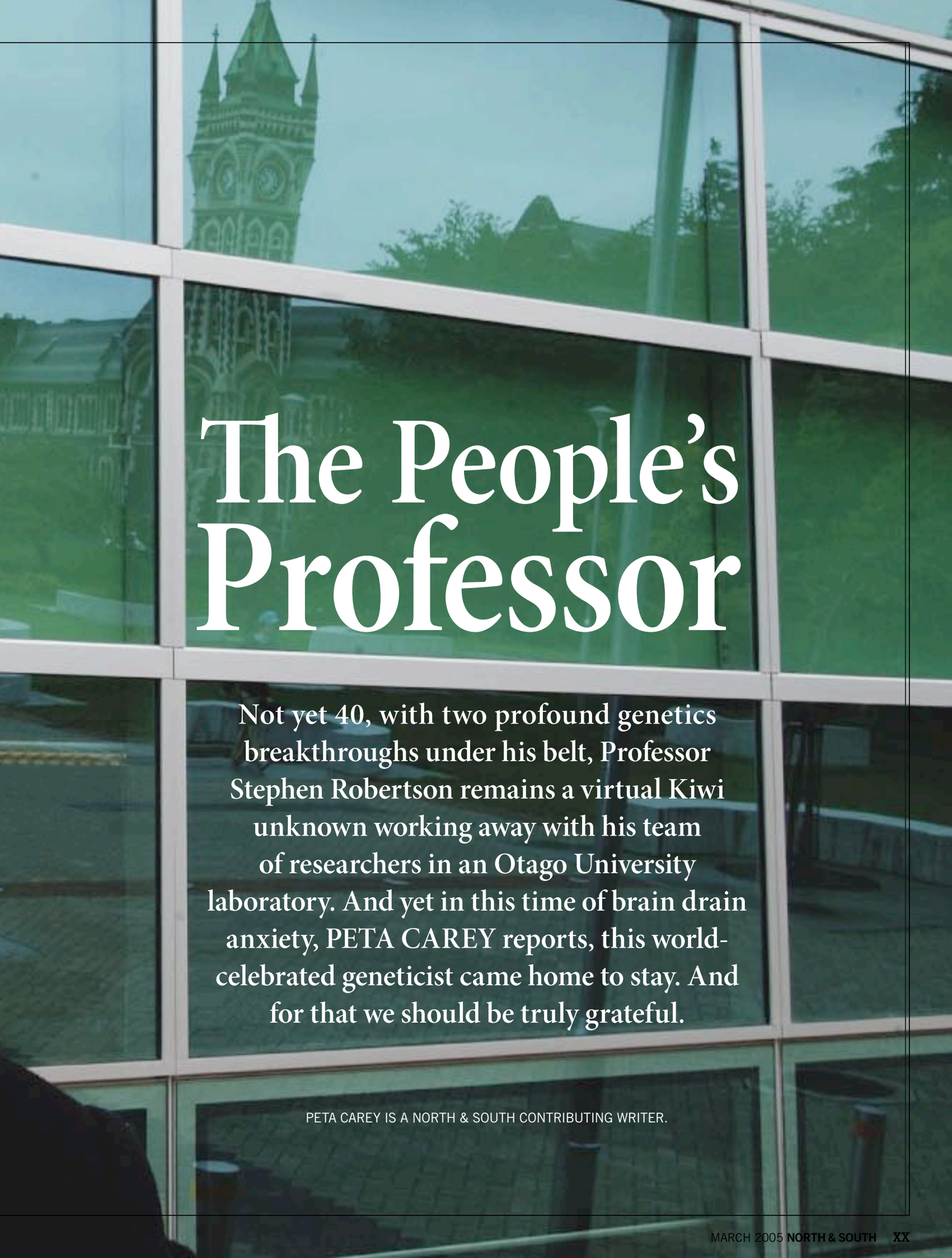

The People's Professor

Not yet 40, with two profound genetics breakthroughs under his belt, Professor Stephen Robertson remains a virtual Kiwi unknown working away with his team of researchers in an Otago University laboratory. And yet in this time of brain drain anxiety, PETA CAREY reports, this world-celebrated geneticist came home to stay. And for that we should be truly grateful.

PETA CAREY IS A NORTH & SOUTH CONTRIBUTING WRITER.

In July 2004 Stephen Robertson brought the science back to the turangawaewae of those he was caring for. From left at Tangiteroria marae: June Miru, Robertson and June's daughter Noki-Jane and Karen.

The 500 Queenstown rich listers were well into their cocktails at the black tie Child Health Research Foundation fundraiser. Having sat rapt through speeches from Peter Hillary and Tashi Tenzing (son and grandson respectively of the Everest greats), the audience's attention wavered as the Professor of Molecular Genetics took the stage.

Stephen Robertson was well aware that a scientist didn't have the same immediate appeal as those who'd made it to the top of the world. The crowd seemed more drawn to the slightly inebriated antics around the tables than on the tall, reserved man speaking quietly into the microphone. Then, as Robertson softly uttered the words: "When the seventh baby died" there was a gathering silence throughout the hall.

For the rest of his speech the audience was pin-quiet and close to tears. As Robertson concluded applause swept the hall like gunfire, and the 500 guests in black tuxedoes and silk frocks dug deep.

Ironically, in the world of genetics research, the man standing before this Queenstown gathering was more than the equivalent of an Everest summiter. With a major discovery in human genetics in 2002, Stephen Robertson shot from research fellow to professor overnight and was suddenly being courted by some of the world's most eminent research institutes.

Stephen Robertson's story — a career path from medicine, specialising in paediatrics, to one of Australasia's leading medical geneticists — is a poignant one. The man who never sought the limelight, never pursued the accolades of the world's scientific elite, found success simply because he encountered a family whose suffering touched him, and he wanted to help.

The family was the Miru/Tito whanau. When Robertson first met them in 1996, five of their baby boys had died at birth. And no one could tell them why.

June Miru is matriarch of the close-knit West Auckland family. Married twice (hence her children's surnames Tito and Miru), she is mother to two sons (one of whom was adopted), four daughters, and several grandchildren. There is always laughter, love and exuberant chaos in their neighbouring households **(suburb?).**

Despite their seeming health and well-being, this family carries an overwhelming load of grief. Since 1988 June's adult daughters Philly, Shona, Karen and Noki-Jane have lost seven baby boys between them, the first three within three months in 1988. All died hours, sometimes days, after birth from severe abnormalities. June had lost her own first born, a son, in 1963.

Robertson was a trainee paediatrician at Auckland's Starship children's hospital when he first heard of the Miru/Tito tragedy and the fact the family could be given no explanation why their boys had died. It was evident the condition was the result of genetic mutation, somewhere "down the line" and that the women were silent carriers. Only when the gene was passed to a male would that baby then die.

Robertson pays homage to the work of his "inspirational" colleagues — Dr David Becroft and Associate Professor Tania Gunn — who introduced him to the family. Tania Gunn, who has since died of cancer, played an early and pivotal role, gathering all the necessary information on the family and moving towards the correct diagnosis. "Tania knew me as an up and coming paediatrician interested in

genetics and thought this was something I should bend my mind to. It's a huge regret she never saw the final fruits of all her initial hard work."

One meeting with June's family led to another, one small step in diagnosis led to the remote but exciting possibility of being able to find the gene responsible. From Auckland's Starship to Melbourne Royal Children's Hospital, then on to Oxford University, Robertson would spend the next seven years of his life pursuing an answer for June's family. No one could have foreseen, back then, that not only would Robertson change their lives for the better, but they, in turn, would irrevocably change his.

The upstairs playroom of the Robertsons' Mosgiel home is strewn with Harry Potter characters — Lego pieces making up castles, cats and wizards. His young sons Nicholas (8) and Mark (6) are giving their father a lesson in tactical manoeuvres. Four-year-old Isabelle dances in, watches for a while, then returns to her mother downstairs.

Paediatrician and geneticist, Stephen Robertson insists his main job in life is husband and father. He's also lecturer and all round good Kiwi bloke. He takes his family tramping in the Otago hills, plays piano and propagates natives. He likes rugby and enjoys a beer. How can anyone so normal also be so brilliant?

"It's not that he's any brainier than anyone else. He just works really hard, He's very much dedicated to being a good father, a good husband, a good scientist and a good doctor. Those are the things that are important to him," says Robyn Blake, Steve's GP wife and greatest advocate. "Despite working hard he tries to keep a balance in his life because he realises all work and no play is not good for you. It's not good for anyone around you either."

"I'm about helping people who are suffering from disease to be more healthy. Be free of some of the pain, reduce the suffering. What genetic research isn't about is making us a better species, it's not about making designer babies."

Robertson never wanted to be a doctor. He simply loved science. Growing up in Hawke's Bay he won the first of many subsequent awards topping the national Secondary Schools Science Fair. "I was enamoured by biology, I thought it fascinating. In the emerging area of biochemistry there seemed to be a chemical explanation for the most complex phenomena that we see in front of us, and that's life. I went to medical school with the ambition of learning medical science to pursue science as a career. But very soon the human aspects of being a physician became apparent — being engaged with human struggles, the intense emotions, joys and dilemmas of clinical practice. The human side of medicine wasn't something I was willing to give away."

And it was children who inspired him most, hence paediatrics. "It's because kids get better and when they get better they show joy, and they get back to living life with such verve, such enthusiasm that you can't not want to try and help deliver that for a young person. Every paediatrician I've met identifies with that."

But somehow being a physician was never enough. Robertson wanted to pursue the answers to the ailments he was confronted with, and in 1996 — when he first met the Miru/Tito family — began his seven-year search into molecular genetics.

Today he's a "physician-scientist". In the course of a week he moves from outpatient paediatric clinics throughout the South Island to his third floor genetics laboratory in the Otago Medical School. "One of the great joys of what I do is being able to deliver clinical help to families at the same time as pursue questions about them scientifically."

As a geneticist those lines of scientific enquiry are never random forays into areas of curiosity. It's important to Robertson that his science *matters*, that it is simply an extension of his work as a physician.

"I'm about helping people who are suffering from disease to be more healthy. Be free of some of the pain, reduce the suffering. What genetic research isn't about is making us a better species, it's not about making designer babies, or manipulating our constitution so we are somehow not the organism we think we are. It's simply about attacking disease."

For the Miru/Tito family that disease, back in 1996, didn't even have a name. Nor did any of the women, if pregnant, have access to a definitive test. Each pregnancy was like Russian roulette, having to carry a baby almost to full term and then give birth — to see whether or not the child, if a boy, was affected. And, if so, then to watch him die. There was also no way of knowing which of the Miru/Tito granddaughters carried the gene. These young women approached child-bearing years unsure whether they'd face the same trauma as their mothers.

Yet in this family, where whakapapa is so essential, despite the possibility of further grief, healthy children continued to be born including three precious, unaffected boys.

This was the overriding motivation for Robertson. "The ability that June and her family have to go on living life, to put this thing in some kind of place, to carry on having kids, to know that kids are important and that even a tragedy like this will not derail the life-force of this family, I think is inspiring. To deliver something for a family like that — because it's so essential and so important to them — was a large part of actually continuing doggedly to pursue an answer."

Ironically it wasn't any of the headline grabbing childhood diseases that took Robertson's attention. What he was pursuing was the cause behind a condition that was incredibly rare (two known families in New Zealand and another few hundred throughout the world), and largely unknown. It was finally named Otopalatodigital Syndrome (OPDS).

What Robertson did know was that the condition was the result of a gene change that had caused significant malformations (he prefers that word over "deformity") not only to the skeleton but also the heart, kidneys, intestines and brains of these baby boys, so they could never survive long after birth.

The chances of finding the gene whose mutation had led to this condition was "akin to looking for a needle in the haystack". The fact the women were silent carriers meant the gene was carried on the X chromosome, narrowing the search from 23 chromosomes to only one. But on that chromosome alone there are more than 150 million letters strung along as a single strand of DNA. Robertson needed to find the equivalent of a single spelling mistake among 150 million genetic letters.

Just what had led to the genetic change in June and her family was a cruel roll of the dice. Genetic mutations occur to many of us with

Paediatrician and geneticist, Stephen Robertson insists his main job in life is husband and father to GP wife Robyn Blake and Isabella, Nicholas and Mark.

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little or no noticeable effect. When they occur in the midst of a crucial gene (a particular functional sequence of DNA), the results can be devastating.

The Miru/Tito clan — full of life, laughter, a "spade's a spade" and say it straight to your face attitude — could be a terrifying prospect for any quietly spoken, Pakeha doctor.

But so too might a scientist — probing the genetic constitution of deceased babies — be potentially off-putting to anyone Maori.

"Genetics is viewed with some suspicion among some in Maoridom," says Robertson. "But in my dealings with this family, they didn't decide to invest trust with genetics as such, but more with the face, more with me."

All five women are emphatic in their praise of the way Robertson dealt with the issues. And enormously grateful. Before meeting him they'd gone years without even a confirmed diagnosis. As June Miru says: "Somebody took the time out to care about what was happening to this Maori family."

In return, Robertson is indebted to their level of commitment to the research. "I had to be very sensitive to, not just the fact that we had some of their DNA, some of their genetic material with me, but also that I was forever dealing with pictures of those little boys, pictures of them."

Not once, Robertson says, was any request denied.

One issue, though, was foreign to Robertson's scientific view of the problem. Since the first baby deaths in 1988 there had been talk of a *makutu* having been placed on the family.

"The idea of *makutu* or a curse actually seems to be something which transcends all cultures when it comes to explaining genetic illness." He cites examples from throughout the world, and throughout history, where many malformations were explained by a curse. Stealing an apple while pregnant, for example, could result in the malformed hand of the unborn child.

"The concept of *makutu* can arise when an individual or family can't understand this thing that's affecting them, when they can't put a face or a cause to it. Too often a curse therefore becomes something which is malign, overshadowing and disabling."

Research, science, samples, blood tests. It was a big ask of a family who'd already been through hell. Robertson was well aware of raising false hopes. "It [weighed] on my mind that I must finish this. I didn't want to start something, and then go back and say, 'Well actually I said we could probably do this, but in fact it's not going to be possible. Before I drew blood on that family I contacted colleagues overseas and asked them if they would be able to help with molecular analysis. That undertaking was a very conscious one."

Little did he know that the research would go far beyond simply finding a name and a diagnostic gene test for the women in early stages of pregnancy, or for subsequent generations. It would turn a corner in the understanding of human development and lead to insights into many diseases well beyond the condition afflicting June Miru's family.

A maze of stone walls and gilt gateways marks the centuries of research at Oxford University. It was here they cured the first patient with penicillin, Edmund Halley predicted the return of his comet and Tolkien penned his *Lord Of The Rings*. Above the portals are the carved titles of the various institutions: Biology, Chemistry, Geology. Nowhere in the rough hewn, stone walls do you find "Genetics". The word didn't come into usage until the 20th century.

Wandering beyond the cathedral spires, up a hill to the John Radcliffe Hospital you come to Weatherall Institute of Molecular Medicine right next door. In comparison to the luxury of space afforded to scientists in New Zealand laboratories, here white coated academics work shoulder to shoulder. But the funding funnelled into this one four-storeyed building far exceeds New Zealand's annual health research budget.

Robertson stepped through the door in 1999, having been awarded a three-year Nuffield Medical Fellowship. "The institute is very famous in world medical molecular research. One of the first purpose-built institutes, its raison d'être was to have physicians who are trained in medicine then come into science, to use their knowledge from treating patients in the clinic to find new and better treatments in the laboratory. It's a powerhouse for research in the UK. There's an intense concentration of talent and resolve to answer some pretty critical questions for world health."

The decision to take up the invitation to go to Oxford was one both Stephen and Robyn agreed to, but with two young children, Nicholas and Mark, it still meant sacrifices.

"Robyn has her own career, but I think she knew she was married not just to me but also married to my dream, so she went for it. That says a lot about the sort of person she is."

Once in Oxford Robyn gained accreditation to work as a UK GP and gave birth to daughter Isabelle. (You get the feeling that "extraordinary and multi-skilled" are attributes easily applicable to both husband and wife.)

Robyn Blake happily embraces her husband's research: "Otopalatodigital syndrome's been a big part of our life, and no-one's ever heard of it, and the Miru/Titos are like an extension of the family really." She tells the story of friends coming to dinner, and the phone ringing. "I said to our friends, 'Oh, that'll be Stephen talking to his family', and they said, 'Oh, you mean his brother?' and it was well, 'No, his *other* family'."

Robertson had three years of funding and laboratory resources at Oxford, tilted towards a DPhil, a PhD in genetics. The only letters he was concentrating on, though, was the DNA code of an elusive gene and the clock was ticking.

Professor Andrew Wilkie, Nuffield Professor of Pathology and "one of the world's best researching physician scientists in clinical genetics", presides over the confines of the Weatherall Institute third floor laboratory. Chocolate wrappers sprinkle his desk among sheaves of scientific papers. Between two computers and reams of bookshelves there's barely room to swing a pipette.

Wilkie is now a close friend, and an ongoing collaborator but at the outset Robertson was just hugely privileged to have met him. "He has a small laboratory but he's achieved a massive amount. To be part of that machine was thrilling, it really was."

What becomes apparent is how much Stephen Robertson enjoys "this stuff of life". Conversing with Wilkie — talking for the most part in four-syllable scientific terms, a language incomprehensible to anyone eavesdropping — there is also laughter, animated and rigorous discussion, and an obvious enthusiasm and love for what they do. It could be two mates talking rugby, fishing, or the anticipation of a weekend away in the wilds.

But there is also no mistaking the responsibility they carry. During the three years at Oxford Robertson was painfully aware of "the name on the sample", keeping in regular telephone contact with the family. During that time Noki-Jane Miru, June's youngest daughter, lost her second son.

> "What we've stumbled upon is not just an area of biology where we've found one gene pivotal to human development; we've found a family of genes. I don't know why that's important, I don't know what they do during development. That's why we're researchers. There'll be answers around the corner and there will be further questions beyond that."

By this stage June's family was not alone. Robertson had located 50 families with the same or related conditions in South America, Eastern Europe, Europe, United States and Australia. But the gene seemed to elude him at every turn.

Robyn remembers her husband's rollercoaster existence. Ever the attentive father he'd come home to put the children to bed before returning to the laboratory in the evenings, getting home finally in the early hours of the morning.

"He went through a bit of a downer at the end of the first year. At the end of two years he went through a bigger downer because he still didn't have it, and that went on for probably two or three months.

"He'd had several false alarms where he thought he'd found it. We hadn't quite celebrated with the champagne every time, but we'd got our hopes up that this was going to be it. And we got more desperate as we were coming to the end of three years. But he'd get through those down times by spending time with the kids. It always picked him up."

From the 150 million letters of the X chromosome, Robertson and Wilkie had narrowed the search to one specific area, a "mere" two million letters located at its very tip. But still it thwarted them.

In early 2002, as Robertson was fervently trying to find the gene responsible for OPDS, scientists around the world were in the final stages of the Human Genome Project — a map of the genetic constitution of the human species, three *billion* DNA letters long. Of those billions of DNA letters, some sequences were known to constitute specific genes — like a computer programme, each gene responsible for a particular function. By 2002 the genes responsible for haemophilia, muscular dystrophy and colour-blindness had all been located and characterised on the X chromosome.

Of particular interest to Robertson was a gene located at the tip of the X that had been shown to be associated with a neurological, epileptic type condition. "These folk are otherwise quite healthy. You could walk past them in the street and never suspect such a critical gene was not functioning properly."

At first Robertson never bothered to look at these genes. Genetic research thus far had shown no suggestion that one gene, when mutated, could result in such markedly different conditions. Robertson had been going down a blind alley — for two years overlooking the culprit.

It was the 11th hour. Reluctantly he'd begun writing up his thesis — a "null thesis that described the genes which were *not* implicated in this group of disorders". As he describes it: "It was the beginning of an English winter, a very glum feeling." On a hunch Robertson decided to go back for one last look at a gene he'd considered out of the running, the gene causing epilepsy. Immediately the results looked interesting.

He'd been to the pub with friends to watch a football game, heading home after midnight in February. "I was on my bicycle and it was achingly cold. This experiment was running slowly overnight and it could have waited until the morning but I decided just to duck in, a little bit giddy and there the result was. There wasn't a eureka

For Robertson returning to Otago was an easy choice. He came back because of Cure Kids, the fundraising arm of the Child Health Research Foundation.

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moment, it was just the satisfaction I'd nailed the proof." He'd found the gene.

For the Miru/Tito family they finally had a name for the gene responsible: *FLNA*. A name and a face.

It was months before Robertson could return to New Zealand to present his findings to the family. All he had was a name, and the ability to offer a diagnostic test to female family members and to any foetus they might be carrying. But to the family — and in particular the next generation of granddaughters — the knowledge was a godsend. They finally had the power to make their own reproductive choices.

Robertson credits June and her family with delivering the crucial genetic information, a gift to families throughout the world. He feels strongly that the power of simply "knowing what it is" is not to be underestimated. "Medical advances must not always be viewed in terms of diagnostics and therapeutics; the value of knowledge for individuals can be just as empowering.

"I think the disability of having something in the family you don't know the name of is something which is much worse than actually being able to give it a face and then square off against it."

Robertson, when pressed, allows himself a small smile and an admission of enormous satisfaction at the final discovery. He refers to the scientists who had been charting and sequencing the human genome at the same time. "There was an analogy written in scientific journal *Nature*, in which they were compared to Magellan, the "Magellans of our genetic constitution". I don't rate myself as a Magellan but somehow I'd had the privilege during this work to actually stumble across my own little rock off Terra del Fuego that I can point to and say I was the first one to find that."

But he'd found more than a rock, more than the gene responsible for a rare and disabling condition. He'd actually uncovered something previously unsuspected — that the gene, mutated in different ways, was responsible for a group of vastly different conditions. That a gene could code for a neurological condition, as well as one resulting in organ and skeletal malformation, raised eyebrows. "The human genetics fraternity sort of turned its head, and that was nice to see. We were excited about that."

He'd also found a gene critical to human development, a gene that coded for particular proteins whose behaviour — in terms of how they set about building the elements of the human body — held surprising new information.

What had previously been considered a gene responsible for the "scaffolding" of our cells — a component of the cell's architecture — was suddenly shown to be the foreman, or project manager, a "master regulator" of signalling between cells. This discovery opened up a whole new direction of scientific enquiry.

"We had to readjust our focus and think 'where to next?' and that's what research is like. It's heading down that corridor and there's a corner ahead and we don't know what's beyond the next corner. It's continually testing your ability to give away your fond ideas, to abandon your preconceptions."

In 2003, not yet 40 and with not a grey hair in sight, Robertson was appointed Professor of Molecular Genetics at Otago University. From a myriad choice of universities abroad he'd chosen Otago.

Professor David Skegg, university vice-chancellor, and world- renowned scientist, was integral in luring Robertson back to Otago, following his discovery in Oxford. Skeggs had also been on the Nuffield fellowship appointment panel back in 1999.

"Provided we can give people like Stephen the resources to work to their full potential, the future for New Zealand will be bright," says a happy Skegg, alluding to recent pessimism about our brain drain.

For Robertson returning to Otago was an easy choice. He came back, he says, because of Cure Kids, the fundraising arm of the Child Health Research Foundation. The newly-created Chair of Child Health at Otago University, which he accepted, is funded by Cure

Kids, an organisation dedicated to funding at least part of his research, hopefully ensuring his ability to follow his research dreams in New Zealand.

Cure Kids CEO Kaye Parker raves about Robertson. Despite a hugely demanding workload, he always makes time to turn out and spread the Cure Kids word. Parker guides Robertson gently from one fundraising event to another, from celebrity golf matches, to the essential black tie executive dinners, like the Queenstown fundraiser. She's accustomed to some scientists who are "extremely good at what they do", but when it comes to public speaking might be a little "reticent". That's not so with Robertson.

"He has an extraordinary ability to speak to corporates, and for them to understand what he does. They're touched by his sincerity."

Parker says he not only helps families with congenital disabilities within his area of expertise, but now, because of contacts with international institutes, he's able to refer families with rare genetic disorders — outside his orbit — to colleagues throughout the world.

"We're extremely lucky to have him. He could have gone elsewhere in the world, but instead chose Otago." The irony, of that, she suggests, is that despite all efforts from Cure Kids (whose contribution makes up only a portion of the funding Robertson requires for his research), there's no security of tenure. "We should be looking after our best and brightest," she says. "It's bizarre we can have a geneticist with two world breakthroughs, a whole team of clinicians working with him and there's no certainty of funding for the following year."

Today Stephen Robertson's work extends far beyond New Zealand. Since that first crucial 2002 breakthrough he's forged international links with research institutes, among them premier US genetics research facility, the Cedars-Sinai Research Institute, allied to the University of California.

His colleague there is highly respected clinical geneticist Dr Deborah Krakow. On the phone from Los Angeles there's a cheer chorus from her family at the mention of Stephen Robertson's name. "My kids love him, we all just love him!"

This is not just your typical American gush factor. Krakow regards him as a "high-quality scientist with stunning dedication. "He really underplays just how hard he had to work to find FLNA. To stay committed for that length of time, to such a rare disorder, and not give up — that's dedication."

Working in a closely associated field of research Krakow learned of Robertson's findings, and approached him to combine their efforts — rare in the fiercely competitive world of scientific research. "Life's too short," says Krakow. "I knew that despite all the work I'd done, we'd get the results far quicker if we combined our work."

In just 12 months of collaboration with Krakow, working between New Zealand and California, Robertson made a second, major breakthrough. The first gene — FLNA — encodes for a protein called "Filamin A" and was found to be associated with a total of six disorders. In this, the second discovery in early 2004, they found its brother gene — FLNB — which encodes for a protein known as Filamin B. Mutations in this gene result in at least five disorders associated with joint and bone conditions.

Explains Robertson: "What we've stumbled upon is not just an area of biology where we've found one gene pivotal to human development; we've found a family of genes. I don't know why that's important, I don't know what they do during development. That's why we're researchers. There'll be answers around the corner and there will be further questions beyond that."

It's a tangled web, this genetics business, an immense three

dimensional maze of components — building blocks, signal stations, and engineering wizardry that could well be beyond the scope of even our imagination.

The irony of Robertson's story is that by pursuing the cause for a rare and little known disorder (to ease the pain of one grieving family) he might well have stumbled on a crucial jigsaw piece that in years to come could unlock the rest of the puzzle.

"It's my hope that by studying these rare conditions, we can make real progress on understanding more common, but genetically more complex, malformations that affect children."

In July 2004 Robertson was finally able to take up the invitation of the Miru/Tito family to visit their Tangiteroria marae, north-west of Dargaville. Five of the family's baby boys are buried in the urupa on the hill behind the marae.

Journeying with him was almost every member of the Miru/Tito whanau. Netball championships had been left behind, work commitments put aside. They were there to honour the scientist and doctor who'd changed their lives. "I love Stephen like a brother," says Karen Tito.

After welcoming formalities Robertson was invited to speak. There was nothing incongruous about this physician scientist on a rural marae. On the contrary, it seemed utterly right for a geneticist to talk about whakapapa, to honour those who'd gone before and to acknowledge the importance of family yet to come. The poignancy of the moment was not lost on Robertson. From the many laboratories and halls of learning — Otago, Auckland, Melbourne, Oxford and Los Angeles — he had brought the science back to the turangawaewae of those he was caring for.

Marae kaumatua Teihe Tito, addressing Robertson as *rangatira*, thanked him, saying: "You've taken away that thing of the *makutu*, the *mata maori*."

It was a still, clear shining winter's day as they made their way up to the urupa. The red and white of the ubiquitous Northland church gleamed as women spoke, often through tears, of their dreams and spiritual beliefs, of where their boys might be now, and who might be looking after them.

Science and spirituality side by side: Neither can explain it all, and yet both could be a comfort. "Getting our head around what makes a human being at least physically, genetically is going to bring a lot of benefit to medicine, I think there's little doubt about that," says Robertson.

"What we can't slip into, though, is [thinking] our genetics is necessarily going to help us fully understand what it is to be human. There are many, many more dimensions to humanness than just our DNA."

January 2005 and the Robertsons have their family vehicle packed for the holidays with five bikes aboard. For a week they wend their way over the Otago Rail Trail. Even little Isabelle manages to pedal a good distance. And Stephen's even managed to pick up a novel. Hardly light reading, he chooses Walter Scott's *Rob Roy* for relaxation.

He admits, however, that even in the Maniatoto, well away from the menial tasks of the laboratory, he never truly switches off. There are still questions nagging to be answered. "There's so much buried in those two genes we haven't uncovered yet. And there are other genes just around the corner," although he refuses to be drawn on the next big breakthrough.

Instead he talks about the families coming to his attention on almost a weekly basis — families with conditions not unlike the Miru/Titos, both Maori and Pakeha, in desperate need of answers.

It's strange then that there are so few people in New Zealand who know of Stephen Robertson and his scientific endeavours. Those few insist he's one of our true heroes deserving of the respect we usually heap upon adventurers, politicians, rugby greats or Olympians.

Roberstson has helped change Ria Rudolph's life for the better but his connection with her whanau has irrevocably changed his too.

Robertson shakes his head and says he's nowhere close to the highly respected upper circle of scientific "greats". And he refuses to have any expectations forced on him.

He's far more intent on drawing attention to the benefits of his science than to himself; on the future possibilities that genetics research can offer. "What we're not doing is trying to make designer human beings. We're not trying to make better babies, we're not trying to change our species in some way so that we're no longer definable as human. That I think is one of the great misconceptions of genetics today."

Instead, for this physician, it's simply about the health of our children. "The equivalent of nearly one child in every classroom in this country is born with some kind of genetic problem seriously affecting their health. And just because it's hardwired, just because it's there in our blueprint, doesn't mean we can't understand it, we can't conceive of it as disease and we can't find a way around it.

"Genetic disease now lies well within the horizon which we should be reaching towards and trying to understand and prevent suffering. Just because it's genetic, just because it's part of our constitution doesn't mean that its effects can't be understood, and hopefully controlled or ameliorated — the suffering somehow made less. And that, quite simply, is what we're about." ■

FOOTNOTE: *Lifting Of The Makutu*, a documentary written and produced by Peta Carey on Stephen Robertson, screens on *Documentary New Zealand*, TV One, February 28 at 8.30pm.