https://medium.com/@KatieCincotta/the-genetic-lottery-ddaff0bd3fed#.kuo3bkk1cThe genetic lottery

By Katie Cincotta

When it comes to winning and losing in life – having good luck or bad luck – most of us think about the lottery: about what winning a few cool millions could do to change our life. Tropical holidays, a pool, a tummy tuck, a never-ending supply of French champagne?

Not many of us think about the genetic lottery we are born with – whether or not that exclusive DNA code will keep us alive and kicking long enough to enjoy a retirement in the sun, and see our grandchildren grow up.

It was peacemaker Mahatma Gandhi that said 'Your Health is Your Real Wealth'. All the science and common sense tells us that eating well, exercising and finding ways to relax and manage stress certainly makes a difference, but our genetic lottery remains the key in determining our health and longevity.

At the moment of conception, we are all thrown a dice. We inherit the genetic code from both our mother and father: 23 chromosomes from the mother's egg and 23 chromosomes from the father's sperm, which together will determine what we will look like, how smart we are, and what might be in store for our body.

For my mother Josephine Cincotta, it came as a huge shock to be told at the age of 64 that she was the carrier of a rare genetic disorder – X-ALD. X-linked Adrenoleukodystrophy – that this strange sounding disease, one of over 40 known Leukodystrophies, could cause serious neurological symptoms, including numbness in her hands and feet, stiffness, loss of balance, bladder incontinence – that she was facing a degenerating nervous system that in the most severe cases could impair mobility enough to warrant a wheelchair.
After three and half years of testing, of countless appointments, of s
cans with specialists, she finally had an answer for why she had been getting tightness in her legs, her arms and back, numbness and tingling in her fingers and toes, and a persistent rash on her legs.

I remember sitting alongside my mother when she received the diagnosis from her neurologist Dr Mark Faragher. It was in January 2015 after extensive genetic testing that they found the ABCD1 gene that would explain her muscle spasms, her stiffening gait, her fatigue (problems that had initially been incorrectly diagnosed as chronic inflammatory demyelinating polyneuropathy (CIDP), which required her to have lengthy blood plasma transfusions, which we now know had no effect.

We asked many questions that day. What will happen? Is there a cure for Leukodystrophy? Is anyone else in the family at risk? The neurologist said he simply didn't have the answers. In more than 20 years of practice, he'd only seen two cases of Adrenoleukodystrophy. We would have to seek out expert opinion from a genetic neurologist, an appointment that would not eventuate for three months because of the demands on the specialist neurologist at the Alfred Hospital, Professor Elsdon Storey.

In the months that passed we Googled frantically, worried, googled some more, and remained baffled by what this neurological disease would mean for our mother's future.
The session with Professor Storey was incredibly valuable and revealed that my grandmother was a carrier of the mutated gene, even though she never had any symptoms in her 86 years. 'Nonna Pina' was a character and a troublemaker and we joked that even from her vault in the Fawkner Cemetery she was still causing trouble.

X-ALD brought confusion and unease, not just to my mother but our entire family. We had no known affected male relatives in our family history, so came to this disease without warning.
As children of a female carrier of Leukodystrophy, we had to decide whether or not to get tested. Whether or not we wanted to know if we too were victims of this gene mutation, if we too would come up in the random roll of the ALD dice – which was a 50 per cent chance for us as children of a mother who is a carrier.
Leukodystrophy currently has no cure or proven therapy, so my sister and I and our brother who is 40 with no symptoms, have decided not to be tested. We figure living with the knowledge of carrier status isn't worth the mental anguish, especially as only 15-20 per cent of women with ever develop symptoms.

My mother is one of those unlucky few that has developed symptoms from X-ALD. They began most noticeably when she turned 60, and possibly decades before with ongoing bladder infections and incontinence which she thought was an after-effect of childbirth.
Sadly, ALD is not the only health problem she has come up against: rheumatoid arthritis, a weak kidney and a damaged mitral valve from contracting rheumatic fever as a child have also taken their toll on her body.

Each day she takes a concoction of some 20 pills – a list she keeps in her handbag, as it's too lengthy to reel off by heart to every doctor.

Her mornings are slow. Hot showers help. Her walk is often unsteady. She has fallen several times, when her leg doesn't get the signal from her brain to move. Recently, she stayed too long on the couch and had so much trouble getting up that she tore a ligament in her shoulder and went to the emergency department at 3am when she could no longer bear the pain.
She is doing what she can to find relief and keep her body moving: a weekly hydrotherapy session at the pool with her girlfriends, physiotherapy, using the B laser device, and taking a raft of drugs to help ease the pain.

Still, my mother felt disheartened, afraid and alone. It was Neurogenetics Professor Michael Faye who suggested making contact with the ALD support group Leukodystrophy Australia to help mum cope with the mental strain of this disease. That connection has been life changing.
As an ALD sufferer herself, Sister Julie Thomas founded The Australian Leukodystrophy Support Group (now known as Leukodystrophy Australia), and without this incredible network many of those afflicted would be facing this disease alone.

As my mother says: "I felt isolated with a disease nobody knows anything about. Since I've spoken to my support group ladies, it has lightened my pain. The simple act of talking, listening and sharing our stories makes us feel less alone, less forgotten, better able to face the daily struggles."

What none of these health issues can take from my mother is her fighting spirit, her stubborn fortitude is something we all admire. Where she finds peace and purpose is in the garden. She seems to move more easily there, her mind content with the task at hand. Even pulling weeds gives her joy.

My father recently found a group in London called Raremark who bring people with rare diseases together to share information, and consider clinical drug trials.

Finding out about the latest research into Leukodystrophy and connecting with people who know something about this rare and debilitating disease gives my mother comfort and hope.
She admits there's a sense of frustration with an illness that is so obscure. For cancer, mental health and more mainstream neurological diseases like MS and Motor Neuron Disease, there is awareness and funding. For diseases on the fringe like ALD, which is only relatively new to the medical community, it's harder to get answers and research dollars.

It's also difficult to explain to people exactly what ALD is, what caused it, what it does to the body, without discussing the complexity of genetics, the brain, the adrenal glands, and very long chain fatty acids.

We have found the easiest way to describe X-ALD is that it is a genetic mutation that causes a build-up of very long chain fatty acids in the brain, which damages the protective layer around the nerves commonly known as Leukodystrophy.

My mother's form of the neurodegenerative disease is slow progressing, and not nearly as debilitating as what young boys with ALD face, so in a sense we feel grateful. She hopes that with the push for newborn testing of Adrenoleukodystrophy we might be able to save those young boys from the deadly progression of the most severe form of the disease - childhood cerebral ALD – which can only rarely be slowed with Lorenzo's oil and stem cell transplants.
In the grand scheme of life, perhaps the only positive way to deal with ALD, and the myriad of diseases which can strike the body, is to increase awareness, to offer our stories, our data, to contribute to Leukodystrophy research and support groups. There is power in numbers – not just for scientists trying to find a cure, but for the peace of mind of sufferers who have been dealt this hand and are struggling to accept the loss of control and freedom that goes with a broken nervous system.

Despite the misfortune of landing one wonky gene in the genetic lottery – my mother is happy to have a loving family, a tight-knit circle of friends, and a determined mindset that she will continue to live an active and fulfilling life despite the constraints on her body.
She is about to join a group of friends for a trip to Fiji. They've booked rooms on the ground floor so she doesn't have to battle the stairs.

She hopes sharing her story will encourage other people to share theirs. It's a simple but powerful way to deal with an illness that has no cure. When you tell others about the long and confusing road to being diagnosed, how difficult it is/ was to process that you have inherited a rare genetic disease that was only classified in the 1990s, what it is like to live with pain and mobility issues – it's a way of moving forward with confidence, a way of feeling less alienated, a way of accepting that you are a part of this strange Leukodystrophy family called Adrenoleukodystrophy, despite never having a choice in the matter. Maybe together, we can bear that burden, better than we can alone.