NEWSLETTER

Genetic Overgrowth PI3K Support
www.gopi3ks.com

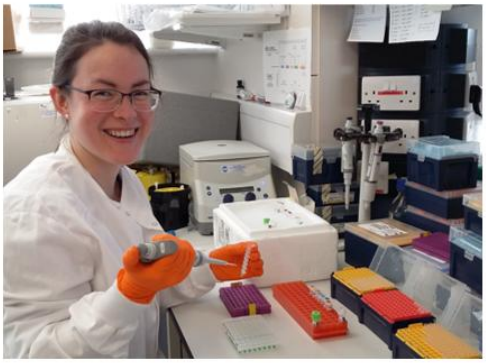
January 2016 Issue 2

Welcome to the second newsletter for GoPI3Ks – Genetic Overgrowth PI3K Support. Here we will keep you informed of the current research, interesting news articles and stories from overgrowth patients.

Recent developments from the research team at Addenbrokes, Cambridge:

SPEEDING UP DIAGNOSIS- NEWS FROM THE LAB

We have now recruited over 240 study participants, and so far we have been able to give a definite genetic diagnosis to 80. For many others we are eagerly awaiting the next round of gene sequencing, and from this we hope to make many more new diagnoses. One of the most important pieces of work we are doing in the lab at the moment is to try to increase the speed of this process, which is challenging because the gene changes that drive the overgrowth are only found in some cells and tissues in the body.



Rachel Knox
Research Assistant

Our approach is first to look for a panel of the eight commonest genetic causes, mostly in the gene called *PIK3CA*. We usually have these results within a month of a person being recruited. If we don't find the answer doing this, we then broaden the search to look at 60 different genes which control growth. This has taken 3-6 months, as we have to run samples in batches, but we are now in the process of changing to a new method of gene sequencing (called MiSeq) which will allow us to run many more samples at the same time, making things a lot faster. If after this we still don't find a genetic diagnosis, we review all available information and either recommend analysis of a second tissue sample (for example, from a skin biopsy) or else we go on to read all genes simultaneously in a final attempt to identify new, rare causes of overgrowth.

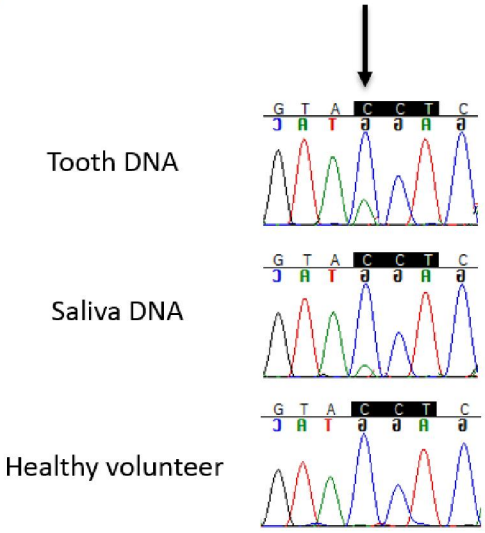
Genetic diagnosis from a baby tooth!

Many thanks to study participants who have sent us their baby teeth!

Tooth DNA

Saliva DNA

Healthy volunteer

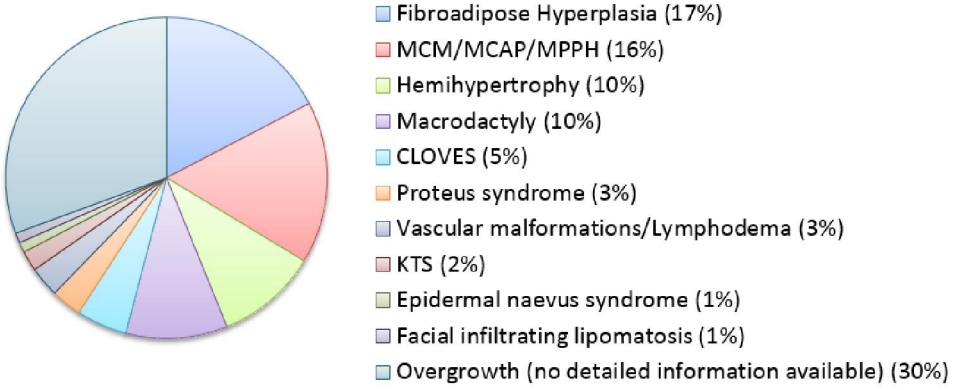


For MCAP/M-CM patients, we have been using saliva as 'affected' DNA to make a genetic diagnosis. However, baby teeth (which have already fallen out) are also a good source of DNA, and often carry the overgrowth-causing mutation, as Professor Clayton-Smith's group in Manchester recently discovered.

We tried this out for one participant who we had previously diagnosed from saliva. As you can see from the DNA sequencing opposite, the small green peak indicating a mosaic mutation is higher in tooth DNA than saliva DNA!

So step aside, tooth fairy! We may have a better use for baby teeth than you...

Clinical diagnoses of our participants upon recruitment



IMPROVING UNDERSTANDING OF LONG TERM OUTLOOK

Thanks to our second grant from the National Institute for Health Research, our new research co-ordinator, Leena de Silva, joined our team in May 2015. Some of you may have met her already during your visits to the Clinical Research Facility. We see 1-2 study participants each week in Cambridge, as well as doing genetic testing on samples sent to us from all over the world!

Perhaps the first critical questions about any medical problem for patients and their families is what the future holds. Answering that question is particularly complex in the rare overgrowth disorders that we study, where

not all parts of the body are affected. This means that different people can have very different severities of problem even with the very same gene change. A key aim of our studies is to draw information about the long term outlook so that we can offer the most accurate advice to families, and so we can target new treatments to those who need them most. There is no quick way to answer this need. Instead it relies on gathering information from as many patients as we possibly can, on continuing to follow their progress as time passes, and on swapping notes with researchers and doctors in other countries who see many similar patients.

It is incredibly important that we register any major medical events that happen to patients who are part of our study in between visits, so we are always grateful for being kept in the loop by our participants and their doctors.

TURNING RESEARCH INTO AN NHS SERVICE

All our studies to date have been funded by research grants. However in our view it is critical that we use the new information we have learnt about overgrowth syndromes and the challenges they pose for patients and their families to improve the dedicated services that can be offered as part of NHS care. In June this year, we met with teams of specialised doctors from Great Ormond Street, St George's, and the Royal Free Hospitals in London as well as specialists from Manchester to discuss a bid to persuade the Department of Health to set up an NHS highly specialised service for segmental overgrowth. The aim of this bid will be to strengthen the "joining up" and co-ordination of care, so there is always a known National Expert Centre and freely accessible advice to both families and their doctors. If successful, this will include 'one-stop-shop' clinics in Cambridge, Manchester and London to see a team of overgrowth specialists for reviews as regularly as needed. Financial times are challenging in the NHS, and this bid will be multi-step, slow process which can't lead to new funding any earlier than April 2017, but the first stage will be submitted shortly and we will keep you informed as to progress.

DR ROBERT SEMPLE
WELLCOME TRUST SENIOR RESEARCH FELLOW HONORARY CONSULTANT ENDOCRINOLOGIST

CLINICAL TRIALS UPDATE

DR VICTORIA PARKER
REGISTRAR IN ENDOCRINOLOGIST

We are about to start a clinical trial with a drug called sirolimus or rapamycin in 30 patients with some of the more severe growth problems. 10 patients in the UK with progressive overgrowth and confirmed mutations in the gene *PIK3CA* are going to take this as a tablet once a day for 6 months, along with others in France and the USA. They will have regular MRI and DXA scans to measure growth before and after taking the tablet and will be monitored for side effects.

In the lab, we have also tested other promising experimental drugs. We are discussing with pharmaceutical companies possible future studies using their drugs, and we foresee a series of clinical trials in the next few years with the potential to transform treatment of overgrowth syndromes.

Julie Harris
Research Sister

We remain hugely grateful to all our study participants, as without you none of this work would be possible! For more information, please visit our website, Facebook page or follow us on Twitter:

http://www.overgrowthstudy.medschl.cam.ac.uk
Facebook: https://www.facebook.com/segmental.overgrowthstudy
Twitter #overgrowthstudy

wellcome trust | NHS National Institute for Health Research | Cambridge University Hospitals NHS Foundation Trust | UNIVERSITY OF CAMBRIDGE

This is James, he has recently been diagnosed with a PIK3CA mutation. I asked if he would kindly share his story with us and its one that I can identify with and maybe you will too.

I'm James Vincent, 32 and I Live in a small village near Cardiff.
Mandy asked me to tell my story about my overgrowth. So this is how I have been diagnosed with Proteus syndrome. Growing up with it. The physical & mental challenges. Other suggestions on what I have and finally PROS-Segmental overgrowth diagnosis.

At the age of 5, my parents took me to Great Ormond St: Hospital in London. We needed to find out why various parts of my body were overgrown. From birth it showed completely throughout, but mainly affecting my left leg with quite a big difference in those early years. Also, my right leg was shorter to the point I needed my one shoe built up. I don't remember that, but by the time I was maybe around 7 years old, the length had caught up with my left leg but the size of my right leg had stayed how it was. Unaffected by the overgrowth. Well, at least size wise anyway.
During those first five years we didn't have anyone that we could go to to ask questions and we didn't know anyone else with overgrowth. So my parents felt going to London was something we needed to do.
I don't remember much of that week in Great Ormond street. Only that I hated it. I sat through many different tests and there were lots of other children also with different disorders. *[I'm using 'disorder' as I don't like the word 'disease' & 'condition' doesn't seem right]*

At the end of it all the best that the doctors could come up with was Proteus Syndrome. Leaving us with that, they just sent us on our way as there was no community to go to for support. So that's what we settled with and that's what my parents lived with. The uncertainty and not knowing of what could come.

> *Proteus is a progressive disorder that grows out of control at a much faster pace than the unaffected parts of the body. It is mostly bone affected, but can be tissue too. This often results in the need for surgery multiple times, to keep cutting back the extreme overgrown parts which in the end my lead to amputation as the best option to save the patient of severe distress in the long run. Or something even worse.*

At this point I should point out that this has impacted my mobility more than anything. With the distance I'm able to walk decreasing through the years, the last four years is where I've seen the biggest change. I've been silently preparing myself mentally for where I could be with this in 5 to 10 years' time.
Also the pain that I'm living with in my right leg on a daily basis is much more. It leaves me wondering what options I'll be left with in the future as painkillers are now almost un-affective.

Doctors insisted on monitoring me from a young age as they feared that this overgrowth could kill me before my teenage years, as proteus is known to be a killer. So there were many regular hospital visits each year for various things, which continued well into my teenage years. These were to check my internal organs like lungs, kidney and whatever else were not growing at a faster rate than what they should be. The dreaded hearing tests became regular also as sometimes I can't hear, but this is due to the tubes [the ones connecting Ear, nose, throat] as when breathing in and out, our tubes open and close but mine can't always do that due to them being larger. But again that's something many doctors didn't understand and many still don't.
We also had to monitor my varicose veins in both legs, size of my legs, what difficulties the physical side of things brought and how the overgrowth was progressing. The pain that they brought was generally forgotten about on these visits so was never really taken into much consideration by the doctors & consultants. So that's what most of the visits consisted of. Mostly to the University Hospital of Wales [UHW].
What we did know is that the bigger parts, the leg, fingers, toes, ear etc… were growing steadily, at the same rate as the non-affected parts.
Which was showing this is not typical Proteus because it was not 'progressive'. But again, no one could tell us anything else.
My facial features look fine and no one would notice anything different unless I point them out as they have smaller differences. Hands and fingers look different, but not by much. It's what isn't visible to people is where things change and the complexity of it all shows.
But as mum always said… I'm one of the lucky ones.
In one way I suppose I am. Though this didn't stop it from feeling incredibly hard at times and isolating.

High school presented itself as the most challenging. Emotionally more than anything. With not being able to take part in most P.E activities, some teachers not understanding why I couldn't run or join in a game of basketball, rounders or anything that required plenty of

energy. I eventually stopped attempting to join in to do what I could in a game as there was only frustration and insults from other class members due to the risk of the other teams winning.
Looking different, not being able to walk far, finding stairs hard some days, seemed to be not good enough for the other teenagers. Because if teenagers don't know or don't understand then they throw judgement. Whether it be comments, looks, torment or even just sheer hate shown from the odd few. All this made me develop many insecurities that I've fought over the years, mostly on my physical looks.
Confidence, trust, showing my legs, talking about my overgrowth, taking up some new opportunities, things like that. I've managed to get on top of them, but sometimes I still feel them.
I did have support, patience and good times from a few classmates, my little circle of friends, but that didn't take away the upset and most of all isolation. On top of my own personal life with things like hospital visits, daily difficulties, I had to deal with school.

Around the age of 19, mum and I were put in contact with the PSF, [Proteus Syndrome Foundation]. This is Tracey Whitewood-Neal's charity. Tracey set this up as her son Jordan, has 'typical' progressive proteus. Tracey has been very supportive over the years to everyone who joins the charity and attends the family gatherings.

I met Dr Leslie Biesecker, a specialist in Proteus, from America at the first family meeting myself and mum attended in Kent. I was about 19/20. All I remember from those two days is that he took one look at me and said, 'You don't have proteus'. Part of me expected that, but it still left me feeling disappointed and confused. He told me I may have HHML - Hemi Hype Multiple Liptocious. Hemi meaning half. I can't remember the breakdown of the rest. But it was getting closer, though this was still some sort of guess. We left it on doctors' notes as Proteus as we didn't want to cause confusion.
Since then I needed to find someone to give me a solid answer. That was my mission!
Two years later we attended the next family weekend. This was when Dr Biesecker threw 'HHP - Hemi Hyper Plaisir' at me. Basically the same thing. But more acronyms was just confusing.
The years went on, we attended the family weekends, but I still very much felt that I was on my own in this and the isolation remained.
Even though Tracey told us we have support from her anytime we wanted, we only spoke and saw each other and the other families at these meetings. With no social media to connect people back then, hardly many email addresses, just maybe a phone number. But what was I supposed to say if I did call someone?

As I was going through my 20s I battled with the physical appearance of my body the most and always feeling judged by it, to trust someone not to do that was not possible.
I needed to find my inner peace to become happy in my skin, but that I didn't know how to do. Though comments and looks were sometimes passed my way, usually by kids or a few college students for a short time [college was generally rather enjoyable]. Some even by family members. I would hide my legs, never wearing shorts and not wanting to go anywhere hot for holiday. Always wearing a t-shirt as I have some patches of pink markings above my waist line. I felt like I should continue to hide myself away. Not seeing any point in

talking to anyone about it as there wasn't and still isn't anyone who actually 'really does' understand. I just kept silent and didn't mention anything to anyone.
After many years of debating and getting upset with myself, also feeling incredibly frustrated that other people were doing things I wanted to do, like swimming and going on holiday. I forced myself to change my attitude and turn my confidence issues around. I decided that if I wasn't good enough for someone, then I didn't need to know that person. I needed to surround myself with positive supportive people.
My peace with myself eventually came, I was 27. I can't tell you how exactly, but I finally felt comfortable in my own skin. I learnt that this is me. I had to be happy in life and only I could make that choice. We ourselves choose if we are to be happy or sad. No one else.
Feeling happy that I found some sense of peace, I thought life could be easier for me. But there was something else showing more by my
mid-20s. I just chose to ignore it.

My varicose veins were putting more pressure on the surface of my legs.
The pain was sharper and more often. Walking was much more of a challenge. I knew what I should have done when I was 18, but as a teenager who wanted to be the kid that fitted in with the others without limitations, I was stubborn.
At the age of 26 I gave in and was referred to a department in the NHS that makes knee high length compression socks. Even though they are extremely tight, they are designed to ease pressure on the legs and keep one standing a while longer.
They have helped greatly and we have also seen a reduction in the amount of fluid in my legs. Without these my left leg would be much worse off and my right just wouldn't be capable of anything.
This is my sixth year with them now, having new ones every six months.
But I still hate them. They aren't designed for comfort but do need some sort of comfort fitting to sit right, if they don't sit or fit correctly then they can cut into me which is very unpleasant. When I first got them I took it for granted that they would solve the pain issues I had. But they weren't designed for that.
I'm still not comfortable within myself having them but I'm not going to let that get on top of me. I'm forcing myself just to get on with it and know that these are my aids for walking. They do the same job as crutches, walking frame and a false leg.
Upon receiving my first pair, it did set my confidence backwards but I knew I had to push on. Although on becoming happier within myself, I was self-conscious of these compression socks and kept it to myself for a while. I've eventually developed the courage to say what they are If anyone asks why. I've just got to show some confidence and make them understand.

As I was heading to my 30s I was still so desperate to know what am I really living with. Over the years, watching various documentaries with Jordan, Mandy and other people with different types of overgrowth. Part of me wished I could be the one someone was spending time and money on.
Then came February 2013. I had no idea things were going to change…
Mandy Sellars' Shrinking my 17 Stone legs aired on TV. I always find it hard to watch these documentaries. So when possible I tend to watch them on my own. I finally watched it one Saturday night the following month in March.

I reached out to Mandy the following day on Twitter. We exchanged a few messages. I happened to mention that Proteus/HHP was guess work and I didn't know what else to do. Mandy asked for my email address to give it to Victoria Parker at Addenbrooke's Hospital, part of The University Hospital of Cambridge.
I said yes. Thinking will anything actually come of this?
The next morning I had an email from Victoria, introducing herself and explaining a little about what the team at Addenbrooke's is all about and to arrange a phone call.
The next day on the Tuesday, we had that phone call. I explained the basics and talked in more detail about what is done there.
NHS England invests money into research for people with rare disorders and work with other charities around Britain with similar goals.
A week or two later I had a date for the 1st May to spend one night, two days there.
All this happened so fast before I realise that I had opened myself up to more tests. The thought of Great Ormond Street came back to me. But I quickly pushed that aside as I knew I had the reassurance that if there was any test I did not want to take part in I did not have to. But I'm the type of person that if I say I'll do it and I'm there, I'll have to because if I don't, I'll regret it all the way home.
During my first visit the team made me feel welcome and looked after me really well.
There were of course the blood tests but they quickly learnt not to show me the tools!
There was a glucose test. Down a pint of Lucozade then monitor my blood to see how I react to a quick intake. All fine there of course.
I had a tissue sample taken for 'growing' in the lab. This was to see if they could find the cause of my overgrowth and get some solid answers.
This was going to take quite a few months, so I knew I had to be patient.
There was a DXA scan. Think of that as being scanned and printed.
My skeleton, body fat, tissue, muscle. All laid out nicely on paper and colour coded.
That brought the first discovery. Not only did it show that I had overgrown tissue in my legs, mostly on the left leg.
But it was the fact that it showed me that my skeleton was not affected by the overgrowth. This was something which no one knew. My overgrowth has created much more tissue throughout my body.
After that I was left for a while in my room. I needed some quiet time as I found that I just cried for a while. I was here the other side of Britain with this new challenge and adventure. I was finally getting answers to the questions I've been so desperate to ask for so long. Though some part of me didn't want to do this, after this discovery that changed.
I dried my eyes, put on that smile and continued with business.
There were a few other tests during my time there like the MRI scan [really didn't realise how noisy those things are]. But the biggest and hardest one was saved till last.
As it's a University hospital, just like my main one home here in Wales, there are students. I was asked if I could help them learn from me and I always do like to say yes, though face and identity would not be revealed.
It was all explained to me and I was all set to go.
I remember being in that waiting room with Victoria and one of the nurses. I think they could see that my nerves were on the ceiling.
Myself and Victoria went into the photography room. Even though Victoria could probably see at this point I was extremely hesitant, I knew I had to do this. I was helping people study overgrowth and learn from me. After all, they were helping me. I took off my clothes with

just my boxers remaining on and the photographer took pictures of different parts of me. This would give the students a look at my type of overgrowth.
Feeling exposed and a little raw about how I was feeling I knew I had done good.
After returning home it took me a few days to come down from feeling poked and cut open!
During my second visit later that year, they needed another tissue sample. The first one didn't have much of an affected area.
Then the news eventually came...
I had a phone call later in 2013, towards the end of the year.
Victoria called me with some good news. Well, sort of. 'James you don't have proteus syndrome'. She said
This time I knew it was real. But I waited for Victoria to say 'but you do have this…'
That didn't come. More time was needed.
It was quite some months that I felt I was just hanging. But I knew Victoria's team were doing what they could. During that wait I had another visit, I was given all the details and kept up to date every step of the way. It didn't take away some worry of what they could find.
I found it was a horrible feeling. This was something I had been waiting for and trying to prepare myself. But now it's here, I didn't know how to feel. I stuck to Proteus when talking about it to nurses and doctors as I didn't want to say something I couldn't give details on and was worried that an answer might not actually come.
For someone who's always had a strong sense of faith in everything, I realised that I'd lost it here.
August 2014 finally came and Victoria called me to give me the details of what I really do have.
PIK3CA. It is the gene change in the 81st Amino Acid in the protein [E81K] in around 20% of my leg and back. *[Seems more than 20% to me but can't dispute science!]* But it's not in my blood.
Oh and it's not inherited from the parents either.
I'm the second to be found in the UK with this gene change. [Mandy's the first]
Those are the basics of the explanation.
The easiest way to put it is Segmental Overgrowth.
Meaning no pattern to it, just randomness.
So, finally. It took 31 years to find out what has caused my overgrowth. It is still just a bunch of acronyms, but there's some explanation to it.
Myself and Mandy have the same gene change but are also very different. My change happened late in my developing stage, where Mandy's gene changed right at the start of her development of life.
Although the feeling of isolation has always been with me, I have had some good talks with Mandy. She's given me a smile after every conversation. I know she does understand how I feel
and on some level, I too know how she may feel at times.
I can't describe how I feel through all this, with what the team at Addenbrooke's has done for me. All I can say is I'm grateful.
They sit and listen to me. They work hard. They made me feel important.
One more challenge lies ahead. Do I take part in the drugs trial to shrink my overgrown leg? I know it may change all overgrown parts too.
Will it work? What will happen if I stop taking the drugs?

Will the changes remain when I stop?
So many questions. If I do it'll be the biggest leap of faith ever. I pray every day that I find the courage to take part.
So much to learn in 2016…

As I end this I'd like to say all is not lost. I am feeling a lot happier and comfortable in myself. I've taken up swimming and I wear those shorts on holiday.
There is support out there. Myself and Mandy have recently joined a new group set up by NHS England for Rare Diseases - Translational Research Collaboration, National Institute for Health Research.
Building a community where we can support each other

Here are some articles that you may find interesting and/or helpful.

The perks and pitfalls of disabled travelling

By Srin Madipalli Guest blogger

Accomable is a new app to find accessible holiday rental properties. Set up by friends Srin Madipalli and Martyn Sibley, it was inspired by a joint wanderlust and some colourful experiences of disabled travel.

I love travelling. I have had adventures all around Europe, the US, Southern Africa, Singapore and Indonesia. I have gone scuba diving in Bali, camped in the South African savannah and trekked through a snowy

Yosemite National Park.

Image copyright Srin Madipalli

I have Spinal Muscular Atrophy (SMA) and use a wheelchair. I need 24/7 personal care to help with everything from getting out of bed and getting ready, to help with washing and feeding and most daily physical tasks as I can't move my legs and have very little use of my arms. I always travel with a personal assistant.

My disability makes travel much harder, but over the years I've found ways around it.

Five years ago I was working as a corporate lawyer in the City. I was having a horrid time on many levels, and as is often the case with horrid periods, you can feel like you have nothing to lose and you become open to trying new things outside your comfort zone.

I decided to take six months off to go travelling. It was the first time I had really attempted it, and while it was an amazing experience, it also opened my eyes to how difficult it is to travel as a disabled person.

"I choose not to place "DIS", in my ability."
Robert M. Hensel

Image copyright Srin Madipalli

Prior to that I had never really travelled. I was terrified of things going wrong. I feared my wheelchair breaking down in a land far away. I feared accommodation being ill-suited on arrival. I feared transport not being sufficiently adapted and leaving me stranded. I feared problems arising with my personal assistants and being abandoned when I needed them the most. The list felt endless and I usually convinced myself to stay in my comfort zone and never try anything.

While I had an amazing time on that trip and every adventure afterwards, pretty much all those fears I had about travel have actually come to fruition. But I found solutions.

As with most fears and difficulties, you realise when they actually happen that there's usually a way to fix it.

In Sydney, I arrived after a nine hour flight from Singapore to a smashed up wheelchair that apparently had been dropped while being unloaded. I was stranded in the middle of Sydney at night. Luckily, the airport staff were very supportive and found a workshop still open and a spare wheelchair to get me to a hotel. This mechanic also managed to patch up my wheelchair temporarily a couple of days later to allow me to get on with my trip.

In Los Angeles, Martyn and I turned up to our hotel in the early hours of the morning to realise that there was a step into the bathroom, even though the hotel listing said it was step-free. We ended up having to work with the hotel porter to create a makeshift ramp to somehow get us in there and at least provide a temporary solution until the morning.

Business partners and lifelong friends Srin and Martyn met as children at an SMA conference

Image copyright Srin Madipalli

In Bali, my wheelchair charger was broken and my wheelchair only had a few hours power remaining. I had to frantically phone around the manufacturer's offices in Germany looking for local suppliers who could help. After being passed on by about 10 different people, I ended up getting a kind recommendation of a small shop in Bali that sold replica Chinese made medical equipment on the cheap, which included a charger that seemed to work.

"When everyone else says you can't, determination says,"YES YOU CAN."
Robert M. Hensel

I've had more extreme experiences too. Baboons have attack my van in South Africa (not much one can do apart from hit the accelerator), and in Las Vegas, while I was asleep, my personal assistant went for a drink and ended up getting into a drunken brawl and got arrested, leaving me stranded in my room. I was stuck in bed till late morning unable to reach my phone, forced to wait until he was released by hotel security and able to get me out.

Image copyright Srin Madipalli

As with most fears and difficulties, you realise when they actually happen that there's usually a way to fix it, and I believe people are fundamentally decent and helpful when you are in need.

But the travel experiences Martyn and I have had led us to thinking that travel shouldn't have to be a risk or a major leap into the unknown just because you have a disability or are elderly. We believe it is possible to create a platform to find and inspect accommodation before travelling, and to find providers of support and equipment around the world, so if things break down it never has to be a terrible nightmare. Moreover, if we can get more disabled people to travel, we believe airlines will become better at treating their equipment with care as it would become something very usual.

Accomable now has properties in 18 countries and we are constantly searching for more. We are under no illusions that we have a long way to go, but we'll keep working hard to achieve our mission to make travel accessible to all. Because travelling is brilliant - angry baboons and all.

The people who race mobility scooters

By Caroline Lowbridge BBC News

Colin Furze raced his mobility scooter against a plane at an airfield in Lincolnshire

With a top speed of eight or 10 miles per hour, mobility scooters are not designed with motorsport in mind. However, souped-up models are increasingly being used for racing and record attempts. Why?

"I don't have a dis-ability,
I have a different-ability."
Robert M. Hensel

The high-speed rider

Colin Furze adapted the mobility scooter by using a 125cc motocross bike engine

"I've often said to people if you want to turn heads don't buy a Ferrari, pimp a mobility scooter up," says Colin Furze, who set a record of 71.59mph in a mobility scooter in 2010.

He has "pimped" three mobility scooters so far, by fitting them with motocross bikes engines.

"It's an experience," he says. "These things have been designed to turn around in a shopping aisle so the steering is quite responsive. You have to keep it dead straight."

Mr Furze used to be a plumber but now makes money by posting videos of his creations on YouTube.

One shows him racing an aircraft at an airfield in Lincolnshire - and accelerating faster than it.

He was winning the race until the aircraft took off.

Colin Furze found he could accelerate faster than the aircraft

"It was very strange having a plane next to you while you were riding on it," he says. "It was a silly race anyway but it was a good match."

His record was narrowly beaten in Denmark in 2012 and since then two Isle of Man mechanics have built a mobility scooter that can travel at 107.6mph.

He thinks using mobility scooters for record attempts is appealing because everyone is used to seeing them being driven at 4mph in the supermarket.

"When you see them in their normal life they are so slow and not built for speed, so to make something go fast that shouldn't normally go fast is the appeal," he says.

"People like things that move. Anything that moves, people will try and race them and get some kind of competition."

Old ones can also be picked up quite cheaply, he says, or are inherited from grandparents.

"There are all these old ones lying around that people have got," he says. "Why not race them?"

Colin Furze filmed himself making a 60mph scooter for YouTube

The endurance rider

Steve Tarrant has been a motorsport marshal for 25 years

Steve Tarrant has set endurance records in mobility scooters since being injured while marshalling at the Goodwood Festival of Speed, a heritage vehicle racing event, in 2000.

A car hit him at about 100mph and the driver and another marshal died, but Mr Tarrant narrowly survived, although his right leg was amputated.

He has made two attempts to break the record for the greatest distance covered in 24 hours.

The endurance challenge - which involves pulling a lever and sitting in a chair while continually going around in circles - requires skill and constant concentration.

Wet and muddy conditions hampered Steve Tarrant's first record attempt in 2012

"I probably adopted the motorsport mentality," says the 54-year-old, who lives in King's Stag in Dorset.

"There were tyre marks where I was effectively skidding around each corner because I was doing it flat out."

His scooter only travels at a maximum of 10mph and he had to allow time for "pit stops" so the battery could be changed.

But on his second record attempt he achieved 190.2 miles (306.1km), exceeding the 170.63 mile (274.60km) record set in California in 2013.

His efforts have not been recognised by Guinness World Records, however, because of a technicality relating to the track, but he raised thousands of pounds for charities.

"Just because a man lacks the use of his eyes doesn't mean he lacks vision."
Stevie Wonder

Steve Tarrant did his record attempts on a TGA mobility scooter

The banger racers

Mobility scooter banger racing is less about the racing and more about the ramming

Banger racing, mobility scooter-style, is known for its low-octane thrills and spills.

Scooters topple over and shopping baskets go flying as competitors repeatedly ram into each other.

"We weren't taking the Mick out of anyone, it was all for fun," says Matthew Watson, who promoted a recent event at Buxton Raceway, in Derbyshire.

"We did try to get people who used them for day-to-day use to come out and have a laugh with us."

Some of the mobility scooters were damaged in the race

He said it went so well he is doing it again on 14 June.

Video footage of the event reveals many of the racers took it seriously, despite their scooters being slower than a child's go-kart.

"We weren't allowed to adapt them," said Mr Watson. "I had one man, he rang me up and said 'I've got one with an engine in'.

"I said 'You can bring it up but you are not in the race'. So he went up and down the straights and entertained the crowd."

"Courage doesn't mean you don't get afraid.
Courage means you don't let fear stop you."
Bethany Hamilton

About 30 riders took part in the race

"However difficult life may seem, there is always something you can do and succeed in"

Stephen Hawking

The long-distance rider

Andy McIntosh's partner and two daughters supported him when he did a 100-mile challenge in November

Former soldier Andy McIntosh says he has "been in some very dark places" since becoming disabled in 2008, due to complications from corrective surgery following an Army injury.

He has a rare vascular condition that means he can only walk very short distances.

"When they turned around and told me this was going to be me for life, I needed something for me to focus on," the 42-year-old says.

He heard about a disabled former Royal Engineer called Simon Angel riding from John o' Groats to Land's End on a mobility scooter.

He decided to do the same, except he wanted to smash the world record of 24 days and nine hours.

"I'm ex-Army and I'm very competitive," he says. "I'm the type of person that if I set my mind to something I'm going to do it."

Andy McIntosh served in The Cheshire Regiment for 14 years

He plans to set off on 5 May and complete his journey in just 16 days - more than a week quicker than the current record of 24 days and nine hours.

The challenge will raise money for military charities including Poppy Scotland and Erskine.

"They just helped me and my family so much and it's our chance to give something back and to help others," he says.

"This is my way of being able to help other people that are going to face the same."

'People want a challenge'

Helen Dolphin, director of policy and campaigns for the charity Disability Motoring UK, does not have a problem with mobility scooters being raced by people who do not need them.

"I try not to take offence at this sort of thing. There are far more serious things to take offence at," said Ms Dolphin, who lost all four limbs to meningitis when she was 22.

"As long as it's not on the road and it's not harming anybody I tend to take the view live and let live.

"What people do on private property they do on private property. Let's hope they don't end up genuinely needing one for good."

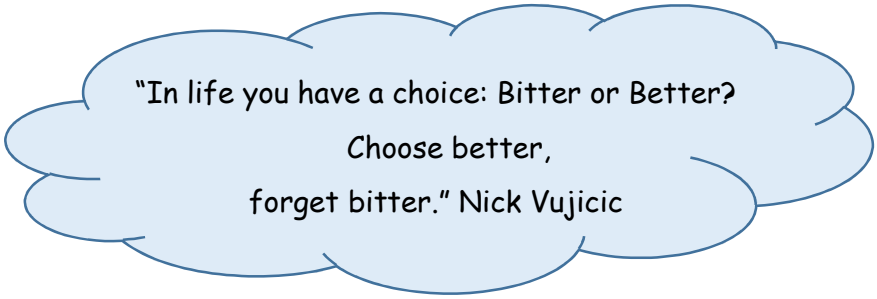
Disabled people have used mobility scooters for records and challenges for about 15 years, according to Tim Ross from supplier TGA Mobility.

He gets about six people each year approaching him for support with their attempts.

"I get a lot more inquiries than I used to," he said.

"The people who have done it with us have been people who wanted a challenge. They may have had some hard times and they want to prove themselves.

"Just because they are disabled they don't want to sit in their house watching Jeremy Kyle.



Viewpoint: Is it time to stop using the word "disability"?

By Rebecca Atkinson Guest blogger
30 September 2015

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After running a campaign to urge toy manufacturers to include disabled characters in their collections, Rebecca Atkinson started to wonder if the word "disability" might also need a positive makeover.

Cripple, deaf-mute and lame all fell out of favour a long time ago and are now considered insults. By the 1980s and 90s "handicapped"

was gradually replaced with "disabled" as a new way of thinking about disability emerged - called the social model. Attitudes change and as a consequence so does language.

Recently there has been a shift towards person-first language and now "people with disabilities" is often more popular in general usage over its predecessor "disabled people". I have noticed too that people in the disability community sometimes like to emphasise the "ability" part of the word with hyphens or capital letters: dis-ability or disAbility.

In April this year I started an online campaign urging the toy industry to include positive representation for the 150 million children worldwide with disabilities. I began making-over toys by marrying princesses with guide dogs or wheelchairs and giving hearing aids to fairies to create a fun and colourful disability aesthetic. I took photos of my creations and posted them on the web under the name ToyLikeMe.

Changing parlance will do nothing if there is not a shift in attitudes towards disability. Tom Shakespeare, University of East Anglia

The images conveyed not a shred of pity, no hint of inability, no inkling of dependence - the many things that people associate with the word disabled. It went viral and, what had started as a hobby, soon gobbled up my life. I quickly found myself writing post after post on the subject, late into the night.

But as I typed the d-word again and again, I started to see it in a new light. It felt like a great lump of a word, stout, ugly, cumbersome and dour. Whilst it conveyed the meaning, it did not carry the modern or celebratory sentiment I wanted. It seemed to focus on the negative when the toys I was creating were singing with fun and colour. For the first time I began to wonder if "disabled", had become outdated, and needed a replacement.

If we don't use the term disabled, though, what do we use to describe someone who has an impairment to set them apart from the majority?

Reading posts from my followers, many of whom are parents of disabled children, I noticed the words and phrases they use: special needs, differently-able, different, ability not disability, inclusive. It seemed many people were choosing not to use the d-word, or were trying to divorce the "dis" from the ability.

The definition of "dis" in one English dictionary is to "have a primitive, negative or reversing force". To discredit. To disengage. And in recent parlance "diss", with an extra s, has been popularised as an abbreviation of disrespect - "Don't diss me."

"Dis" is not a thing that many people want prefixed on their child or themselves. It is, after all, inherently negative.

One adult follower wrote to me in outrage after I posted an image of a toy with a facial birthmark under the banner "dolls with disabilities".

"I am not disabled," she wrote, as if the term was highly offensive. "I have a port wine stain and although it's nice that you create a doll with a birthmark, I think it's a poor choice of words..."

"A disability is "a physical or mental handicap, especially one that prevents a person from living a full,

normal life or from holding a gainful job," she continued. "I have a normal life. I am a teacher. I think you should seriously reconsider the use of the word."

I read her message and agree you should be able to choose your own identifying label but I wasn't sure how to take her rejection of disability. She sees it as separate from the idea of living a "normal life" or having a "gainful job". But, looking at the many disabled people I know and have worked with, they do have these things.

Perhaps a facial disfigurement or birthmark isn't a disability but, for many people, constant stares and negative remarks are disabling, and I feel there is still a need for children growing up with these differences to see themselves reflected in the toy box to help build self-esteem.

So I began to search for a catch-all term that could be used to describe disability but also include those who wanted dolls with glasses, eye patches, birthmarks and scars - differences that do not always fit under the d-word banner.

Find out more: Why campaigners are giving toys disabilities

Media caption Toy Like Me campaigner Rebecca Atkinson wants to see ''disability taken out of the hospital and into a more fun setting''

The comedian Adam Hills has a part of his leg missing and, in an interview with the Guardian in 2012, declared "mutant" a much better word than "disabled". "It sounds so much cooler," he said. I have sympathy with this, my sister and I have long referred to me as a mutant in private but I wasn't sure this level of irony would work for everyone.

The phrase "disability and difference" sprang to my mind as a replacement but felt a little wordy, overly PC and problematic. Humans, after all, are all different from one another so where does normal human difference end and disability begin?

Disability is a delicate flower around which the public tread with care. People with disabilities are used to being described as unable or hailed as an inspiration but may dislike both. So are disabled people "special"? Or do they span the spectrum of human life from talented to mediocre just like all humans? And does a benign descriptive word, free from negative connotation, actually exist?

The bio-etheticist Rosemary Garland Thomson came near to a solution when she spoke in April this year of disability as a form of "human variation" arguing that disability should be understood as a reality to be accommodated, not a problem to be eliminated.

Human variation? It kind of works.

We are all definitely variants of the human race, and the term can indeed encompass everything from a facial birthmark to someone with glasses, and someone with no limbs, but I suspect it's a term wide open to a charge of "political correctness gone mad".

As the academic and broadcaster Tom Shakespeare said when I met to ask his views: "Negative association will pin itself to any word. Changing parlance will do nothing if there is not a shift in attitudes towards disability".

This will undoubtedly take more than the evolution of words alone, so for now I'm sticking with the d-word.

FINALLY:

I am looking at getting this support group out there in the social media world, on Facebook and Twitter. Here I am hoping that we can get more regular and updated information from Dr Semple and Dr Parker about their work, the drug trials etc. Plus it will help to raise more awareness of not just the PIK3CA overgrowth condition, but overgrowth conditions in general.

Once I have these pages up and running I will send you the links and it would be great if you could like the pages, share with family and friends and contribute too, so that we can help other people out there who haven't been lucky enough to receive a diagnosis, start treatment or have the chance to talk with others who have the same or similar condition.

From all the team at Addenbrokes & myself
we hope you all had a very

and a wonderful New Year.
I hope that 2016 is filled with lots of love,
laughter and good health.

Thank you for reading our newsletter, if anyone would like to contribute to the next one, or have any ideas that could help the group progress, please email me (Mandy) at:
funnypaws2004@hotmail.com