



Issue 9 Winter 2018



Big plans for Center Parcs 2019!

Join our Teddy Bears' Tea Party to mark International Dravet Day 2019

After many months of dedicated fundraising, careful planning and extensive preparations, we are pleased to announce plans for our Annual Family Weekend Away in the heart of Sherwood Forest.

This year, we are thrilled to once again be able to offer a Center Parcs grant of £250 to every registered family who wants to join us for a weekend of fun-filled activities, all due to the continued support of our inspirational fundraising community. Thank you everybody!

The weekend is confirmed for **Friday 21st – Monday 24th June 2019**, and this year we have new sibling activities planned such as 'Crazy Science' alongside the ever-popular 'Laser Combat'.

We have also scheduled the weekend to coincide with **International Dravet Day**, which falls on **Sunday 23rd June**. To mark the day, we have planned an extra-special event with our Teddy Bears' Tea Party including tea, coffee, sandwiches and cakes! The party will run from 12:30pm – 4:30pm offering fun for all the family, with lots more surprises in store and details to be announced soon.

If you are unable to join us at Center Parcs, we will also be encouraging everybody to hold their very own Teddy Bears' Tea Party at home or in your local community, to help us to mark International Dravet Day 2019 and raise awareness of our fantastic cause. So watch this space!

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TRANSCRANIAL MAGNETIC STIMULATION

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DEVELOPMENTS IN DRAVET SYNDROME 2018-2019

Hello!

Meet Linzi, our fab Fundraising Co-ordinator



My name is Linzi and I am Fundraising Co-ordinator here at Dravet Syndrome UK.

I live in Derbyshire and am married with two children; in my spare time I enjoy reading, knitting, walking my dog (Stanley) and spending time with family and friends.

I am also proud auntie to Amy Finch, who is 23 years old and living with Dravet Syndrome. This year I celebrate my 5th year working with the charity. I take care of all applications that we receive, handle all merchandise orders and make extra sure that all of our lovely children and their siblings receive a birthday card... plus getting involved wherever I can!

My background includes working with children with special needs, working for Mencap and being a nursery nurse at a special needs school. I also spent a decade as a support worker and activity coordinator in a residential home for young adults, with a range of challenging needs from learning disabilities, mental health issues, physical disabilities, behavioural needs, epilepsy and sensory impairment.

I am looking forward to making 2019 the most successful year yet for our wonderful charity and if you have a fundraising idea or need help with an event get in touch!

Linzi Cowley, Fundraising Co-ordinator

Contact Us



We love hearing from you, whichever way you choose to contact us!

Please share your stories, photos and fundraising successes using any of the methods below.

Phone:

You can call us on 07874 866937

Post:

Some people still like to write to us. Our address is:
Dravet Syndrome UK
PO Box 756, Chesterfield, S43 9EB

Email:

Most people like to contact us by email because it's quicker. Send to: **info@dravet.org.uk**

Social Media:

Why not send us a message via Facebook or Twitter?

Facebook: **Dravet Syndrome UK**

Twitter: **@dravetUK**

You can find us on:

LinkedIn: **linkedin.com/company/10990437**

YouTube: **DravetSyndromeUK**

Instagram: **dravetsyndromeuk**

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How to book

When booking through the contact centre, please advise that you are attending the Dravet Weekend at Sherwood in 2019. The advisors will automatically offer free central preference as part of the booking process.

Alternatively if booking online you should book your lodge as normal and select a central lodge. You will then need to send a separate email to **callcentre.specialservices@centerparcs.co.uk** explaining that you have a booking as part of the Dravet Weekend at Sherwood 2019 and advise of your booking reference number. The refund adjustment will be made manually.

Once you have booked your accommodation with Center Parcs you will need to complete our grant form to claim your £250 grant but should you have any queries or questions in the meantime, please contact our wonderful Relationship Manager and Trustee Teresa on **teresa.f@dravet.org.uk** who will be very happy to help.

We hope as many of you as possible will be able to join us on our woodland adventure in 2019, see you there!

SAVE THE DATE
Center Parcs, Sherwood Forest
Friday 21st –
Monday 24th June 2019



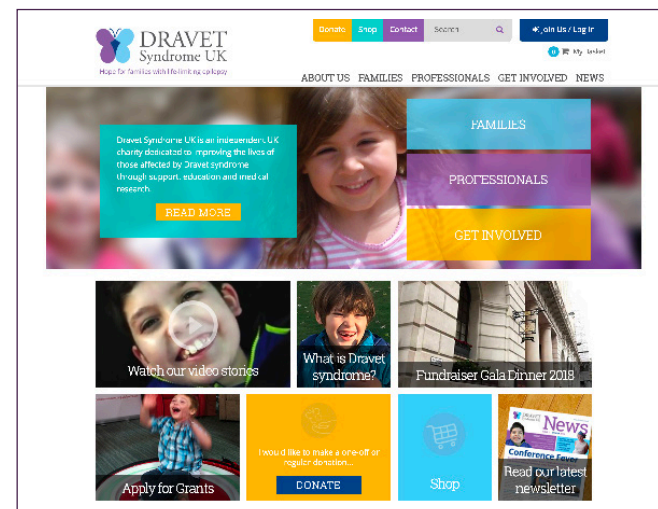
The wonderful world of web! New website planned for 2019

In early 2017 we launched a new and improved website to help make life easier for those of you applying for grants and registering your interest for events.

As we all know, the world of technology doesn't stand still for long. Two years on we have begun scoping and planning work for a new and refreshed website with the aim of further improving the ability for registered users to manage their account and applications, along with refreshing and updating their personal and contact data.

We also hope to achieve greater flexibility for managing the site content, meaning we are able to add and update content more regularly to ensure we continue to bring you the latest news and updates from the world of Dravet Syndrome.

Users will be required to reset their account password once the new site launches but don't worry, we will keep you fully updated as work progresses.



If there is anything you would find useful or would like to see on the new website please email **gary.j@dravet.org.uk** with your suggestions!

DSUK film shortlisted for Charity Film Awards 2019

Fundraising film impresses and makes the final cut



Our recent awareness film that shines a light on living with Dravet Syndrome through the story of Samantha Slocombe and her young family, has made the shortlist for Charity Film Awards 2019.

There was stiff competition to make the shortlist for the awards with over 40,000 members of the public voting for their favourite charity film. For us to have even made the shortlist is a great testament to the enthusiasm of our community of amazing supporters, and the effectiveness of our video.

The shortlisted videos will remain on the Charity Film Awards website and you can watch our video here:

<https://www.charityfilmawards.com/id/118464>

Final judging by the panel will be in February with finalists due to be announced in March, followed by a red carpet awards ceremony for winners in April. Whatever happens from here, to have brought so much attention to our video through the entry and help to raise awareness of Dravet Syndrome has amazing value.

We are incredibly proud of the film we produced for our organisation and the families we support. A huge thank you again to Samantha Slocombe and her lovely young family for agreeing to share their experiences with the world, along with Red Goat Productions for their specialist knowledge and guidance in producing films for charities.

Thank you to everyone who cast their vote for us and we shall be sure to keep you updated of further news and announcements.





Transcranial magnetic stimulation

An insightful window into how the brain functions

On the 15th November 2018, our Charity Chair, Galia Wilson and Charity Manager, Sam Reed were invited by Professor SM Sisodiya and Dr Katri Silvennoinen to see first hand their research study looking at brain excitability in genetic neurological disorders at the Clinical Neuroscience Centre in London.

Dr Katri Silvennoinen kindly explained how this technique works and its possible impact on Dravet Syndrome.

"TMS, Transcranial magnetic stimulation, has been used to study neurological disorders and brain function in healthy people since 1985. TMS, is a special means of studying the excitability of the living brain. The magnetic pulses transiently activate brain cells over a very small area on the surface of the brain. This activation travels to the hand muscle through nerves, causing a wave in the electric muscle trace (EMG), which is measured with special stickers. The size of this activation tells us how excitable the surface of the brain is.

The technique involves giving brief magnetic pulses over the area of the brain that controls the movement of the hand. Participants experience a clicking sound with each pulse.

Other possible sensations include movement of the hand. Sometimes participants experience transient tingling of the scalp or twitching of the facial muscles, which is caused by the nerves in the scalp being activated by the magnetic pulse.

By measuring responses to pairs of pulses, we are able to test the function of specific brain circuits. These circuits are very interesting in Dravet Syndrome, as they appear to be particularly affected by the abnormality in sodium channels associated with the condition. Indeed, a previous study undertaken at Epilepsy Society showed that a group of people with Dravet Syndrome had a different response to this test compared to people with other types of epilepsy, or no epilepsy.

As we are now expanding this study, however, we have noticed that there is variability within people with Dravet syndrome with respect to these changes. We are now studying whether this variability corresponds to other variability observed between different individuals with Dravet Syndrome, e.g. differences in response to certain antiepileptic medications, or the severity of epilepsy or

cognitive difficulties. If we are able to find such correlations, TMS could become a useful tool for informing us about each individual's condition. It could also help in choosing the most effective treatments for each individual."

We would like to thank Dr Katri Silvennoinen and her colleague for their time and their thorough demonstration of TMS technique.



Super Siblings 2018 Announced

Recognising inspirational bravery and selflessness

With so many stories from proud parents, our Super Siblings Awards are designed to recognise the brave, selfless and inspirational brothers and sisters of those who are living with Dravet Syndrome.

On Friday 7th December we closed entries for our Super Siblings Awards 2018 and we are proud to present the names of our 13 amazing Super Siblings who have received our award in 2018!

In no particular order we would like to congratulate the fabulous:

Joshua Robertson
Simon Stone
Nellie Phypers
Megan Tebay
Isla Ramsay King
Daniel Sims
Phoebe Parnham
Sam Davison
Hajar Ali
Danny Tambling-Goggin
Rishi Pradeesh
Teagan Dickinson
Ciara Clare



By now each of you should have received your Super Siblings certificate along with your gift voucher as a small token of thanks from us all...

Congratulations, you are all amazing!



Christmas Wreath fundraising by The Flowerpot Ladies



Over 80 stunning Christmas decorations sold

The Flowerpot Ladies (Angela and Sarah) is located in Fulham, SW London, and offers perfectly planted pots and window boxes delivered to your door.

Angela's son James is 17 years old and living with Dravet Syndrome, and to help support our cause at Christmas, The Flowerpot Ladies came up with a fantastic festive fundraising idea to help support our cause... festive wreaths!



Angela told us a bit about this festive fundraising plan:

"We decorated around 80 wreaths, with all the profit from sales going to Dravet Syndrome UK. We organise everything and then we host a couple of evenings along with our helpers (who we couldn't do without!) to sell the wreaths. We have also managed to offer a recycling service, so once we have collected the wreath we can reuse the decorations as much as possible, so even more profit can go to the charity."

As you can see, the wreaths are truly festive and are simply stunning – this year the green-fingered Flowerpot Ladies raised just over £2,000 for our cause... amazing!

We would like to say thank you to Angela, Sarah and their dedicated helpers for such a wonderful Christmas gift.



A Christmas Tree for Dravet

1st Gosforth Guides enter Seascale's Christmas tree festival

We were delighted to receive a 'Merry Christmas' message from Julie Dickinson and the Gosforth Guides, along with a delightful Christmas donation!

The Gosforth Guides decided to enter their own tree at St Cuthbert's Church as part of the 4th Annual Seascale Christmas Tree Festival in Cumbria.

After the girls held a vote to nominate a charity to support, it was decided they would support Dravet Syndrome UK. Our cause is close to the hearts of the group as Guide Leader Julie's daughter, and Guide Kayla's older sister, Jessica, is living with Dravet Syndrome.

This year the festival had a nursery rhyme theme for the trees and this group of intrepid young ladies decorated a stunning 'Mary had a Little Lamb' themed Christmas tree in support of our fantastic cause!

Not only this, but the girls also helped raise much-needed awareness of Dravet Syndrome and Jessica was keen to get involved too, helping the Gosforth Guides decorate their amazing tree to raise a festive £36.55 for our cause.

A massive thank you for your support and Christmas gift, you have shown the true meaning of Christmas spirit!



An epic year of fundraising in memory of Georgie Barnes

Memorial fundraising raises almost £10,000 for good causes

Many of you will remember the brave, courageous and inspirational fundraising mission of Geoff Barnes and friend Tim Ellams.

The duo decided to raise funds in memory of Geoff's daughter Georgie Barnes, who had Dravet Syndrome but tragically passed away just after her 3rd birthday.

Together they set out to raise a target of £15,000 by taking on many events over the course of 2018 including running, walking, trekking up mountains, cake sales and much more along the way. All funds received were to be equally split between three organisations that were pivotal during Georgie's life, and we are delighted to have now received £3,200, as Geoff got in touch to say:

"With special thanks to Tim Ellams for helping me on this journey in 2018; we pushed the boundaries of our abilities and fitness levels, racking up the miles in memory of the most beautiful and brave girl. I hope this makes you all smile, I'm proud to have been able to do it all and extremely grateful for all you've done for my family!"

We would like to say a massive thank you to Geoff and Tim for their dedicated fundraising and amazing achievement - Georgie would be very proud and will live forever in our hearts.



Attending 'Family Day' for the first time

We speak to Stephanie Smith for her story ahead of our London conference

We hold our Dravet Syndrome UK Family & Professional Conference every two years, with the hope of connecting families and professionals alike and to promote a sense of unity, purpose and knowledge within our community.

However, we appreciate attending for the first time can be daunting for some. We speak to mum Stephanie Smith from Cumbria, whose 7 year old son Jake has Dravet Syndrome, about the experiences of her family ahead of our 2019 return to London for this years conference. This is their story:

When you receive a diagnosis like Dravet Syndrome it can feel like your world has imploded. No one

can prepare you or predict what the future may hold for you and your family, which is part of what makes Dravet Syndrome so hard.

Jake was 18 months old when we received his diagnosis, and for the first few months we went through every emotion possible whilst we came to terms with it. Living in the Lakes limits our access to specialists and in most cases many of the doctors, community nurses and other specialists had never even heard of the condition or knew how to support Jake and us. It made the condition even harder to deal with as we felt alone, isolated and without support. Jake's condition began to become more complex and as parents we felt helpless, as did his team, to help him.

That's when we were introduced to Dravet Syndrome UK. The impact the charity and all of the incredible team

have had on our lives is difficult to put into words. To be in touch with other families who could empathise and understand how we felt, and have a database of advice and literature, helped us begin to understand Jake's condition.

In 2015 we attended our first Dravet Syndrome conference in London through Dravet Syndrome UK. We had no idea what to expect or how we would feel meeting other families face-to-face. I wouldn't be being honest if I didn't admit that a part of us felt scared that we may have to face up to hearing aspects of the condition we didn't want to hear, or that we may see older children or adults with the syndrome that had more severe symptoms. But we also knew that to be able to support Jake and his twin sister to the best of our ability, we had to arm ourselves with as much knowledge as we could!

The conference allowed us to receive vital information directly from leading specialists from around the world relating to every aspect of Dravet Syndrome. From seizure information, to behavioural issues, to mobility issues, to sleep and development... every area was covered!

Hearing from different specialists about current and emerging treatments gave us hope and knowledge to feed back to our local team and for the first time since Jake's diagnosis, we finally felt less helpless. It also allowed us to not only engage with specialists from all fields but to meet with other families and share our stories.

Hearing from different specialists about current and emerging treatments gave us hope and knowledge

Hearing that we were not alone and learning other people's journeys not only helped us emotionally, but also by sharing experiences it allowed all the families the chance to get advice from people who were actually going through the same thing. When one parent shares similar experiences and can offer some useful hints or things that work for them, it really is priceless and we left not feeling negative like we first thought, but more hopeful for the future.

Knowledge is power and whilst none of us want to hear the bad, it is far better to be prepared and know that you are doing everything in your power to keep your child safe. Dravet is unpredictable, unstable, and takes no prisoners but having a forum and platform that puts you in contact with the leading specialists, paving the way to fighting and beating this syndrome, allows you the chance to take back some control and fight it head on.

Since attending the first conference so much has changed for Jake and all of us for the better. The conference put us in touch with Dr Elaine Hughes who took Jake on as a patient. As part of a team based in London, she has used her expertise to better manage Jake's condition, seeing Jake twice a year and supporting his local team in how to better manage Dravet Syndrome.

Jake became critically ill in December 2015 with high ammonia levels resulting in Jake aspirating and developing pneumonia. Jake was on life support for a long time, but afterwards the team in London worked closely with our local hospital to find out how it had happened and how to make sure it never happened again. Dr Hughes referred Jake to Evelina's Metabolic team and placed him in her Dravet clinics, creating a bespoke plan to manage Jake's ammonia levels.

In 2017 we attended the conference in Liverpool along with Jake's local paediatrician and community nurse. On Professional Day Jake's local team gained so much from hearing from specialists who were experienced in the condition and came back with a wealth of ideas to better support Jake. For us not only did we get to meet with even more families, we also heard from more specialists and heard of more research and emerging treatments, which continue to give us hope.

After the Liverpool conference we received support from Fiona Scolding QC, a legal specialist at the conference who advised us to make sure Jake's needs were being met through his EHCP. We would never have known without hearing from her at the conference what rights Jake had, nor felt we had the knowledge and expertise to battle the education system. The conference allowed us to share our story about Jake and afterwards we received comments from families that told us through

hearing about Jake's symptoms with high ammonia that it allowed them to have their child checked and then treated.

After meeting so many people at the conference we made the decision to attend the DSUK Annual Family Weekend Away. This year will mark our 4th Centre Parcs trip and it's an event that Jake, Ella and both of us look forward to.

Life is a journey with none of us knowing what's around the corner, but we are all learning new things every day. The conference allows a safe, supportive environment for families to come together, question and learn, to better arm our Dravet warriors to be able to battle and beat this awful syndrome. If you take just one thing from the conference, it really is priceless... but for us not only have we taken so much, we've gained new friends and knowledge to help support us as we venture into the unknown.

Our family cannot thank the team enough for arranging this and will continue to attend and support them for as long as we can.

*We hope to see you there,
Love Wayne, Steph, Jake and Ella x*



Join us in London for our **Family & Professional Conference 2019**

Two days of expert speakers packed with information and updates

In 2017 we held our biennial Family & Professional Conference in Liverpool at Hilton One and were joined by guest speakers from around the world.

This year for 2019, we are looking forward to heading back to London for the first time since 2015 to bring you yet another packed agenda of leading voices in Dravet Syndrome offering the latest news, updates, advice and medical research.

The event also offers our attendees the chance to meet other Dravet families and professionals, to share experiences and stories and to learn about the latest exciting developments in medicine and research.

As in 2015, we will be returning to The Tower Hotel overlooking the world famous Tower Bridge with Professional Day to be held on Friday 15th November followed by Family Day on Saturday 16th November 2019.

With around 10 months to go until the big event, we are busy finalising our agenda that already includes Professor Helen Cross OBE, Professor Rima Nababout, Professor Sanjay Sisodiya, Dr Elaine Hughes, Professor Sameer Zuberi, Epilepsy Nurse Neil Williamson and Fiona Scolding QC. We expect to announce more speakers from around the world soon.

Ticket bookings are now open, along with further details and information, via our event microsite at www.dsuklondon2019.org

We have secured a discount code for families/delegates who wish to book rooms at the venue, however there are only a limited number of discounted rooms available and these are offered on a first come, first serve' basis.

The rooms on hold are allocated for families and delegates, The contact number is **0800 330 8005**, Option 1 and if you inquire you should provide the code **'DRAV071118'**.

We welcome all interested in learning more about Dravet Syndrome including medical professional, students, friends and families alike – so please do spread the word and we hope to see you in London later in the year!

SAVE THE DATE

The Tower Hotel, St Katharine's Way,
St Katharine's & Wapping, London E1W 1LD
Professional Day Friday 15th Nov 2019
Family Day Saturday 16th Nov 2019



XTX Markets partnership goes from strength to strength

DSUK named Charity of the Year for the second year running



As part of our corporate outreach strategy, early last year we were thrilled to be named Charity of the Year for XTX Markets following their support and attendance at our 10 Year Anniversary Fundraising dinner, held at The Ned in the heart of the City of London.

XTX Markets provide consistent liquidity to help market participants throughout the world obtain the best prices in the various assets classes they cover, regardless of changing market conditions.

Our partnership continues to develop, and on Friday 7th December 2018 we were pleased to be invited to host an awareness event for interested staff at their stunning offices in Kings Cross.

The presentation was entitled 'Cannabidiol as a Medicine - A Dravet Syndrome UK Case Study' and was given by our Charity Chair Galia Wilson, along with special guest speaker Professor Helen Cross OBE.

We welcomed around 30 XTX Markets employees to shine a light on this catastrophic condition and explain the significant contribution this key strategic partnership is making toward our cause.

The talk ran from 4:30pm until 6pm following which XTX Markets had kindly prepared a stunning selection of drinks and nibbles, offering staff the chance to meet some of the people behind the cause they are supporting and to ask questions about Dravet Syndrome.

The event was a huge success and we would like to pass on our sincere thanks to special guest Professor Helen Cross OBE, who took time out from her impossibly busy schedule to join us and support our charity.

We are also delighted to have been named XTX Markets Charity of the Year again for 2019 - XTX Markets Global Head of Marketing & Comms Tim Moxon, said:

"Dravet Syndrome UK's invaluable work in creating a greater awareness of this rare genetic epilepsy was a key factor in our decision to select them as one of our corporate charities."

Having researched various charities with a personal connection to our firm, when we engaged with DSUK we saw the dedication of the staff, the work they do, the community they are creating and most importantly - the direct effect of tireless fundraising on those suffering from Dravet Syndrome, and their families."

XTX Markets has been proud to support the charity and its wonderful work ever since and in guaranteeing a set % of our annual profits - we know this money and support will go to where it is needed most."

We would like to thank all at XTX Markets for their interest and support in our event and the work of our charity. Your contribution is helping to make a direct and significant difference to the lives of those affected by this catastrophic condition.





Developments in Dravet Syndrome 2018-2019

In review with Dr Andreas Brunklaus, Consultant Paediatric Neurologist

With so much going on relating to Dravet Syndrome, we invited Dr Andreas Brunklaus, (Consultant Paediatric Neurologist, Paediatric Neurosciences Research Group at the Royal Hospital for Children Glasgow) to give us an update on exciting research and the many developments happening in the world of Dravet Syndrome.

"Last year we saw a number of significant developments emerging for the treatment of children with Dravet Syndrome including the legalisation of certain cannabis-based medicinal products, evidence for the effectiveness of fenfluramine treatment and promising developments in the field of gene therapy.

On 1st November 2018 the UK government rescheduled certain cannabis-based products for medicinal use.

The marijuana plant called genus cannabis contains over a hundred different compounds called cannabinoids. [1] Two of the most well-known and researched cannabinoids are tetrahydrocannabinol (THC) and cannabidiol (CBD). Cannabis plant derived products contain a mixture of these cannabinoids. CBD has the most evidence in controlling seizures but does not have the psychoactive effects (known as a "high") seen with the use of THC.

Although cannabis-based products have been used as therapy for thousands of years they are by no means harmless. We know that accidental ingestion and overdosing of cannabis in young children can be harmful leading to lethargy and coma.

Every new medicinal product has to undergo a rigorous assessment to establish whether the product is effective and just as importantly whether it is safe to use.

In standard practice this is done via a so-called double-blind randomised controlled trial (RCT): a group of patients is given either the trial drug or a "dummy" drug (placebo) without the patient or their doctor knowing which drug they have received. After completion of the study the effectiveness of the trial drug is compared to that of the placebo. If the trial drug is significantly better than the placebo it might be accepted as a new treatment given it is safe to use and the study has not shown significant or dangerous side effects. Throughout the trial any illnesses or problems are reported as adverse events so that the safety profile of the drug can be determined.

There has recently been a randomized controlled trial in Dravet Syndrome that found that after a 3-month treatment period on cannabidiol (CBD, Epidiolex GW Pharma) the number of convulsive seizures had at least halved in 43% of patients compared to 27% on placebo. Five per cent of children became seizure free. The main side effects were sleepiness, diarrhoea and reduced appetite.

Therefore, the only rigorously trialled cannabis-based product with good evidence to support its use to treat epilepsy in Dravet Syndrome is CBD as Epidiolex. At present Epidiolex is being reviewed by the European Medicines Agency as to whether a licence for use should be granted. It will thereafter be reviewed by the National Institute for Health and Care Excellence (NICE).

It is worth pointing out that during the double-blind randomised controlled trial with stiripentol in DS the number of convulsive seizures had at least halved in 71% of patients compared to 5% on placebo. Forty-three per cent of children became seizure free.

Fenfluramine, a previous weight-loss drug, recently underwent a double-blind randomised controlled trial after having been successfully used to treat children with DS in Belgium for many years.

After a 3-month treatment period on fenfluramine the number of convulsive seizures had at least halved in 70% of patients compared to 8% on placebo. In 25% the seizure activity was reduced to one or no seizures. The main side effect was decreased appetite. There were no cases of cardiac valve defects and no evidence of pulmonary hypertension.

References [1] Ali et al. Efficacy of cannabinoids in paediatric epilepsy. Dev Med Child Neurol. 2019 Jan;61(1):13-18 [2] Isom et al. Targeted augmentation of nuclear gene output (TANGO) of SCN1A prevents seizures and SUDEP in a mouse model of Dravet Syndrome. Abst. 1.051, AES 2018.

Fenfluramine is currently also undergoing licensing procedures so it can be prescribed in the UK to treat children with DS.

Although response rates for stiripentol and fenfluramine in DS appear much higher compared to CBD there has been no face-to-face study between the different products and therefore they cannot be easily compared.

Gene therapy in Dravet Syndrome has recently become a major research focus. Dravet Syndrome is caused by a mutation in the SCN1A gene which leads to reduced expression of the SCN1A sodium channel and loss of its function in the brain. The consequences of this are difficult to control seizures, significant learning disability, problems with movement and behavioural difficulties. A technology called antisense oligonucleotide (ASO) has been used to increase the expression of the SCN1A gene to raise the SCN1A protein and sodium channel to normal levels. [2] The aim of this treatment would not only be to control seizures but also to improve the other disabilities associated with Dravet Syndrome.

As a first step this approach was tested in an animal model of DS and the researchers were able to show that ASO therapy restored normal protein levels, reduced seizures and increased survival in DS mice. [2] These results are very encouraging and this method could lead to the first disease-modifying treatment for patients with DS. The aim is now to advance this program to the clinical stage and undertake human trials to assess its feasibility for use in children.

Finally, 10 years after the first UK-wide study on "Long-term outcome and quality of life in Dravet Syndrome" we are now inviting families affected by DS to tell us what is important to them and how it impacts on their lives. By studying children and adults over a longer period of time we hope to gain valuable information on whether individuals benefit from specific treatments and we hope to better understand any additional needs children and adults with DS have. We wish to thank all children and adults with DS and their families again for their ongoing participation in this research project."

Our thanks to Dr Andreas Brunklaus for sharing this update; Dr Brunklaus is a scheduled speaker at our conference in London this November – you can find out more and get your tickets by visiting

www.dsuklondon2019.org





Continued support for exciting milestone research projects

Research announcements and updates

In 2017 we announced continued funding of £18,560 for the updated research application received from Professor Sameer M Zuberi at the Royal Hospital for Children in Glasgow. The planned study incorporated testing up to 5000 individuals with epilepsy and identified around 700 pathogenic variants in SCN1A, the majority of which have Dravet Syndrome phenotypes.

Our funding in 2017 for the Research & Database Manager, Leigh Hamilton, for 2.5 days per week has been key to the epidemiological studies in Dravet Syndrome offering a key reference for incidence of Dravet Syndrome and to support early genetic testing in all infants with epilepsy or complex febrile seizures.

One of the key aims of our charity is to identify and fund research into Dravet Syndrome and due to the incredible work of our fundraisers, we are thrilled to announce our continued support for the research project lead by Professor Sameer Zuberi with a further grant of £22,328 in 2018!

Our on-going funding for Research & Database Manager, Leigh Hamilton will allow further development of the database.

Professor Sameer Zuberi recently updated us on the research:

"Historically our database has been used to provide information for many projects linked to Dravet Syndrome; we have completed work which provides the most accurate data on how common the condition is and we feel that this will encourage children with complex febrile seizures and epilepsy to have early genetic testing."

"We continue to collect detailed information on this group and also their families. We will also use our database to begin a ten-year follow up of a large cohort of people with Dravet Syndrome. We hope that gathering this information will give a better picture of what life with Dravet Syndrome is like in the long-term for affected individuals and their families."

The project continues to progress the build of a 'Childhood Epileptic Encephalopathy Database'. The genetics lab has now tested more than 6000 individuals with epilepsy and the SCN1A database, which now has more than 3000 patients who have been referred for SCN1A single gene testing. Leigh will also be able to continue her work on a number of other databases such as the Genetic epilepsy clinic database, GACE Study database and the Developmental assessments database

We are also very pleased to offer an insight into the 2017 project with Professor Simon Waddington **'Gene therapy approach for onset childhood epilepsy, Dravet Syndrome'**

Professor Waddington and his research team are developing a treatment for children with Dravet Syndrome that aims to deliver a corrected copy of the gene into tissues and organs. They aim to first test the treatment in the lab, then in animals that carry the Dravet mutation. They hope to be able to fix the genetic mistake, allowing nerve cells to function normally and avoiding the seizures and other debilitating symptoms of Dravet Syndrome. If successful, this gene therapy could provide long-term treatment or even a cure for children with the devastating condition.

One way of delivering a corrected gene to a child's cells is to use a modified virus, known as a vector. Normally, harmful viruses take hold within the body by inserting their own DNA into cells. In gene therapy, researchers remove the harmful viral DNA, replace it with the required corrected gene, then release those cells into the body. The modified virus seeks out cells and inserts the corrected DNA into them, fixing the genetic mistake causing the child's disease.

Because the gene linked to Dravet Syndrome is very large, it is very difficult to create a gene therapy vector that contains the entire gene. This has meant that, until now, researchers have struggled to develop a gene therapy for Dravet Syndrome. Professor Waddington's team hope to overcome this problem by developing a technique to deliver the gene in two halves. They will develop two new gene therapy vectors, each designed to carry half of the corrected gene.

Update on the project's first year funded by Dravet Syndrome UK

The team have successfully produced these new gene therapy vectors, each capable of carrying half of the corrected Dravet gene. Through initial laboratory testing, they showed that, working in tandem, these new vectors appear to successfully fix the genetic mistake and help to restore normal function to nerve cells.



Royal Hospital for Children in Glasgow



This exciting progress, seen in both cell populations and some mouse test subjects, suggests that the approach could hold promise as a viable treatment for children with Dravet.

A crucial element of the study is developing an animal model that accurately reflects both the underlying genetic and physiological characteristics of Dravet Syndrome. This will allow the team to test their treatment under conditions that most closely mirror the human condition. During the first year of the project, they have successfully created a population of mice that carry the genetic mistake associated with Dravet Syndrome and display symptoms comparable to the human form of the condition.

Next steps

The team will now focus on rigorously testing the effectiveness of their gene therapy in their Dravet mouse model. They hope to gather evidence to show that the vectors successfully deliver the gene to cells and correct their function, leading to a marked reduction in the symptoms of Dravet Syndrome in the mouse test subjects.

As ever we would like to thank all of our incredible fundraisers for their dedicated and inspirational efforts, without which such research projects would not be possible. Through increased understanding and improved treatments, your commitment is making a direct and significant difference to the lives of those living with Dravet Syndrome.



Join us for our **marathon 26-mile walk in London**

Take in world-famous sights and complete a marathon for our cause

In 2017 we took on Ben Nevis together raising a staggering £52,000. Then in 2018, we took on Snowdon raising a dizzy £20,000.

This year for 2019, why not join us to take on another epic bucket list challenge as we trace a 26-mile route walking through London (you don't have to run, unless you really want to!)

On Saturday 28th September, Team Dravet Syndrome UK will congregate in central London to take on a marathon walk around our capital city. This marathon walk takes in world-famous sights and some less well-known corners too.

This is a tough one-day event; at approx. 26 miles it forms an enormous challenge for walkers (there is a half-distance option available) but we will be taking each step together knowing that we are raising money to help support the lives of those living with Dravet Syndrome.



And rest assured, we plan to have plenty of fun along the way too! So if you want to take on the walk in fancy dress then go for it! Plus we plan to stay together after the walk for food and drinks to celebrate our achievement.

The team will set off early on our route that takes us through peaceful parks and alongside waterways, and past iconic landmarks including St Paul's Cathedral, Big Ben, Buckingham Palace and the London Eye as we trek through the historic heart of the city.

You can choose your registration option and register to secure your spot with us on the challenge by following the link below:

<https://www.discoveradventure.com/challenges/discover-adventure-marathon-walk>

Once you have booked your spot, please email our fantastic Fundraising Co-ordinator linzi.c@dravet.org.uk who will send out your fundraising pack and add you to the Team Dravet Syndrome UK list to keep you up-to-date with plans ahead of the day.

What are you waiting for? Let's do it for Dravet!

SAVE THE DATE

Thames Marathon Walk Challenge 2019
Saturday 28th September 2019, London

