DRAVET Syndrome UK

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NEWSLETTER
OCTOBER 2012

TEAM DRAVET CLIMB BEN NEVIS RAISING £23,500

On June 30th this year 36 individuals, comprising of parents, siblings, friends and extended family members of Dravet patients, came together to climb Ben Nevis, the biggest mountain in the UK. The fundraising target for this expedition was £10,000, which was the total figure required for Dravet Syndrome UK's first year's commitment to part-funding a Research Administrator to work alongside Dr Sameer Zuberi, one of the charity's Professional Advisors, at Yorkhill Hospital in Glasgow. The research administrator's role includes tasks such as:

* Maintaining and analysing the database of referrals to the SCN1a diagnostic service. The database includes clinical data and genetic findings, which is probably the most comprehensive in the world.
* Integrating other genes tested, and their associated phenotypes - PCDH19, SLC2A1, STXBP1, CDKL5 and others, into a comprehensive Childhood Epileptic Encephalopathy Database, which has been granted ethical approval as a research database.
* Supporting the genetics team in making research grant applications and ethics committee applications.
* Organising the practical aspects of collaborations with other centres, including those in London, Wales, Australia and the USA.

The climb itself was long and drizzly with no view whatsoever at the top. However every single member of Team Dravet completed the climb.

Special mention must go to Sue Groves, who unbeknown to her had a broken foot, yet still battled on and completed the climb. Other special mentions go to Rachel Spray and Dave Lay, who battled on even though I'm sure they wanted to give up many times; in fact Dave was quite ready to charter a personal helicopter to get him off that mountain!

Marie McLeish deserves a mention for being our top fundraiser and raising well over £3,000 and finally huge thanks to committee member Teresa Finch who had the initial idea and worked tirelessly ensuring everything was planned to precision.

The fundraising target was completely and utterly smashed with the final figure coming in at £23,500. Thanks to this amazing achievement we have been able to agree an extra £7,500 taking our total commitment to Yorkhill to £37,500 over 3 years. This money will directly benefit patients with Dravet Syndrome through creating a greater understanding of the condition and ensuring that accurate diagnosis is made for children in the future.

Huge thanks to everyone who took part in the climb, all those parents left at home caring for the children whilst their spouses were away and everyone who donated and supported.

SUPER SIBLINGS

By Martin Baker

Thank you again for all of your "Super Sibling" nominations since the last newsletter. It is great to hear more stories of the amazing brothers and sisters out there in our community. As previously, all of the nominations will be carried forward and considered for the next award. This quarter the winners are slightly different in that they were not nominated by their parents. Both of the winners this time round are older siblings who took part in Team Dravet's Ben Nevis expedition and were both nominated by Marie McLeish. Marie, who also took part in the climb and is Mum to Amanda with Dravet, was so impressed with the support these siblings showed their Dravet brothers that she was compelled to nominate them. Not only do these siblings support their Dravet brothers on a daily basis, they literally climbed a mountain to help raise money to further research and understanding into their condition.

Ok so let's meet the latest "Dravet Syndrome UK Super Siblings"

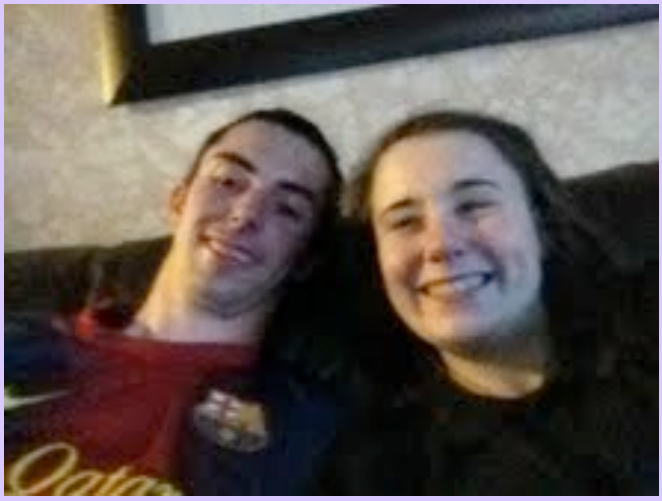
16 year old Hayden is brother to Iwan

Hayden is 16 this month and is in year 11 doing his GCSE exams next summer. He plays guitar, electric guitar and bass guitar. He plays bass and is lead singer is in his band Diesel Dirt Circus. He enjoys going to see other local bands and recently enjoyed a "Stiff Little Fingers" concert which he saw with his dad.

He suggested Dravet Syndrome at school for a charity fundraiser, stood up in front of his class to give reasons why they should choose this charity and he won. Elfed High School in Buckley did cake sales, guess the amount of sweets in the jar and bag packing.

He was not phased at all about the Ben Nevis climb and wasn't bothered that he was to miss a school trip to Alton Towers because of it. He has lots of great memories of the weekend.

Hayden & Iwan



Cara and Gary

19 year old Cara Collins is sister to Gary

Cara enjoys baking cakes and has recently begun to sell them on to people. She has a real talent for photography and has won a few competitions. Cara was joined on the Ben Nevis climb by her boyfriend Russ. Cara and Russ are keen hill walkers and were both hugely enthusiastic about the climbing Ben Nevis.

As always our Super Siblings nomination process is an ongoing one so if any of our readers would like to nominate a Dravet Super Sibling please do get in touch with me via Marie to get a nomination form and I look forward to announcing our next Super Siblings in the next newsletters.

Many thanks,

Martin

EUROPEAN CONGRESS ON EPILEPTOLOGY

Monday 1st October saw the start of the 4 day European Congress on Epileptology, held at the ExCel Centre in London Docklands. With over 4,000 delegates expected from all over the world Dravet Syndrome UK were thrilled to be able to exhibit, giving us the opportunity to meet with numerous professionals.

It was incredibly gratifying that all the professionals we spoke to had an understanding of Dravet Syndrome and many were already working with either adults or children diagnosed with the condition.

This was also the perfect platform for us to launch our 'Professional Guide to Diagnosis'. The guide was produced in an effort for us to raise awareness of Dravet Syndrome, the symptoms of the condition, how the condition progresses, the benefits of accurate diagnosis, how to diagnose and what to do after diagnosis. The guide was produced in consultation with Professor Ingrid Scheffer, Dr Sameer Zuberi and Dr Elaine Hughes, and our thanks goes out to them for their guidance. Our aim is for this guide to be available to all professionals right from primary care through to consultant neurology level.

Another important aspect of the congress was the private meeting we were able to organise in conjunction with the Dravet Syndrome Foundation Spain, Alliance Syndrome de Dravet in France and Dravet Italia. We were extremely fortunate to be joined by a fantastic team of experts in Dravet Syndrome including our own Professional Advisory Board and the research team from Swansea University who are undertaking one of our research programmes, alongside Drs Dravet, Nabbout, Chiron and Professors Guerrini and Scheffer. This was a wonderful opportunity to have so many experts in one room and be able to discuss research gaps and how to move forwards, particularly on a more clinical and practical basis. We would like to thank everyone who gave up their time to come along and share their expertise and ideas and we are delighted to have left the congress with some firm ideas and plans for future research, improving the lives of children and adults with Dravet Syndrome.

Chairperson Marie Baker with Dr Charlotte Dravet at ECE 2012

Special thanks also goes to all our volunteers who gave up time to help man the exhibition stand, Galia Wilson, Rachel Spray, Amy Thompson and Annabel Hughes.

DRAVET SYNDROME UK ONLINE

Don't forget to keep updated by regularly visiting our website www.dravet.org.uk

You can also find us onTwitter and Facebook

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GENETIC FACTORS AFFECTING MORTALITY, COGNITIVE FUNCTION AND CAUSES OF DRAVET SYNDROME

AUGUST 2012 UPDATE

Professor Sanjay Sisodiya

Dravet Syndrome UK are funding this 18 month research project that will use exome sequencing to help answer the following questions:

1) What underlies early mortality in Dravet Syndrome?

2) What causes cognitive impairment in Dravet Syndrome?

3) What is the cause of Dravet Syndrome?

The project aims to determine whether, in addition to a possible known SCN1a mutation, other genetic factors exist that determine these patterns.

Exome sequencing is still a developing technology, in the sense that it cannot be done everywhere by anyone, but needs to be undertaken carefully. Once technical issues are overcome, then analysis of the data is the next challenge, as this technology produces a vast amount of data that needs to be carefully examined to find any real causes of a given condition. However, the strategy can certainly prove successful.

With this study, we were keen to be particularly careful. One component of the study was to search for possible risk factors for the devastating outcome of SUDEP in Dravet Syndrome. Obviously, any DNA samples we have from children who have sadly succumbed to SUDEP are irreplaceable. We therefore wanted to undertake the sequencing in a very experienced unit.

There has been good progress along this and other lines. Firstly, the sequencing is going to be done at the Wellcome Trust Sanger Institute near Cambridge. The Institute is the country's leading sequencing centre (and indeed a leader on the world stage). They have enormous experience, and we are therefore confident that the samples will be handled carefully. We will also benefit from their experience in analysis.

Secondly, we have also secured collaboration with Professor Ingrid Scheffer from Australia, who has sent across additional samples for analysis. Thirdly there is at least one other group in Europe is working along similar lines and we have agreed to keep each other informed.

The samples are prepared, have been shipped to the Sanger and the process of exome sequencing has begun.

Processing and analysis will take 3-4 months, which is the expected time currently for such work. Of course I cannot guarantee this, but we may even have some results by the November meeting!

DRAVET SYNDROME UK ANNOUNCE NEW TRUSTEE

We are absolutely delighted to announce and welcome a new member to the Board of Trustees. Galia Wilson accepted our recent invitation to the board stating ‘since Arlo was diagnosed with Dravet Syndrome, DSUK has been an invaluable resource for me and my family. It's so nice to find a place that we all feel 'normal' and meet people who simply understand. I'm excited to join a fantastic team of Trustees where I can use the skills I developed working in Healthcare communications (before DS) to help this life-saving, and enhancing charity’.

So a huge welcome to Galia who is joining our team of trustees and committee members, all working hard to improve the lives of those affected by Dravet Syndrome.

New trustee Galia with son Arlo