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CDKL5 UK News

VOLUME 1, ISSUE 1

MARCH 2013

Welcome to our 1st Newsletter

We are very pleased to welcome you to the first issue of CDKL5 UK News.. We have come such a long way in a short time, from the humble beginnings of a small band of like minded parents who were given some much needed support and guidance from Rett UK, to the fully fledged charity that we are today. Rett UK took us under their wing and gave us the opportunity to grow into the independent charity that we now are, and so our sincere thanks go to them and we hope to continue to work alongside them in future.

CDKL5 UK has been set up to raise awareness and vital funds for the global research effort into CDKL5. Run by parents, family and friends of those living with a CDKL5 affected child, we

hope to raise awareness and offer support throughout the UK and Europe whilst helping with research into this relatively unknown condition.



*We are growing from strength
to strength.*

Together we are Stronger!

In the last eighteen months we have gone from strength to strength, meeting new affected families, which is so important to us, and gaining some much

needed recognition within the medical community. This is something we hope to build upon in the months and years to come. Our aim is to make getting this diagnosis a little easier to deal with by having the support there for the family both medically and professionally.

We have gained some important support along the way, enabling us to really start to make a difference to those living with CDKL5. Their continued sponsorship and fundraising is deeply appreciated.

We have lots of things planned over the next year and we are excited by the prospect. We invite you to join us on this vitally important journey by continuing with your support and helping us really make a difference.

Meet our Board of Trustees

Our Board of Trustees is made up of dedicated, like-minded individuals who all share a vested interest in CDKL5.;

Carol-Anne Partridge is our Chair, and mum to Amber. She is currently studying for a BSc (Hons) in Social Work.

Sarah Skillicorn is our Vice-

Chair, mum to Gracie and a Community Staff Nurse.

Adrian Partridge, husband to Carol-Anne and dad to Amber

Amanda Black is mum to Olivia and a Haematology Nurse.

Teresa Newey is mum to Ellie.

Sarah Malins is mum to Evie.

Linda Bickerton is our Treasurer and is a Medical Practice Manager.

Claire White is auntie to Olivia.

And last but not least, Martyn Newey, dad to Ellie and Consultant Orthopedic Surgeon, is our Medical Advisor.



Our Mums from around the World raising awareness on the 'Today Show'

City of Dreams: NYC 2013

International Rare Disease Day saw our mums from around the world touch-down in New York City for the weekend of a lifetime. Cementing cyber friendships that have for some lasted years, by meeting face to face for the first time, with the amazing backdrop of the City that doesn't sleep. We gathered from as far afield as England, Ireland, New Zealand, and

Canada to meet fellow mums from the USA. We bonded with good food, good company and explored the city together through love, laughter and tears. But, of course, being the parent pioneers that we are, we couldn't pass up the opportunity to raise awareness, and so we found ourselves adorned in our bright green CDKL5 sashes outside NBC's The Today Show

one very chilly March morning. This gave us some much needed air-time live across the USA. Inevitably, the weekend ended far too soon and we found ourselves saying emotional goodbyes to our fellow CDKL5 sisters. With a reinforced sense of purpose we are all the better for our 'little weekend' away which highlighted just how important support really is.

Fabulous Fundraisers

Our families really have taken up the gauntlet with their fundraising efforts and for that we thank them as they are an integral part of CDKL5 UK.

They have for the past year been organizing local charity evenings, evenings of medium ship, baking, cooking, running, cycling and swimming all in the name of CDKL5. They have been and continue to be truly inspirational.

We look forward to a range of events planned over the coming months.

Saturday 30th March is the date of the "fire-walk", a joint venture between Rett Ireland and CDKL5 UK.

Friday 19th April sees a charity golf day and party night in Chelmsford, held by Evie's Angels on behalf of CDKL5 UK.

We also have two brave mums attempting the 5 x 50 challenge starting 31st March for sponsorship. This involves either running, jogging, walking, swimming.....5km every day for 50 days !

These are just a select few events - should you feel inspired to pick up the gauntlet yourself or to sponsor those that have please visit the web-site and have a peek.

Our first Gala Ball is scheduled for June 1st 2013.



"Extra, Extra" - Our first Gala Ball

2013 is the 50th Anniversary of Martin Luther King's "I Have A Dream" speech. To celebrate our dreams of making this world a better place for ill or disabled children, Martha Care, Cure Rett and CDKL5 UK have joined together to bring you an amazing Charity Event! With entertain-

ment, a 3 course meal, auction, live music and disco in fabulous surroundings, this will be the event of the Summer 2013. If you go to only one charity event, make it this one !

The event will be held at the Grand Hotel, Bristol on June 1st 2013. We promise an evening of grown up

glamour, glitz and retro, accompanied by an 18 piece band "Boulevard Swing". To find out more and to buy tickets, click on www.summerball.org.uk

CDKL5 Disorder International Registry Database



**We can
make a
difference
together**

The first cases of CDKL5-related disorders were only described in 2004. This is still a relatively new and uncharacterised condition with perhaps no more than 700 individuals in the world who are known to have a CDKL5 muta-

tion. Although the cardinal feature appears to be the onset of seizures within a few weeks to a few months of birth, it is also becoming clear as more case-series are published that there is some variability amongst affected children. Larger clinical studies are becoming available which will hopefully give a more comprehensive picture as time goes by.

The International CDKL5 Disor-

der Database was established in 2012 as a collaborative effort between researchers located in Australia and the International Foundation for CDKL5 Research. The database aims to collect information from families on their child's clinical presentation, including developmental history, seizure presentation, gross motor function, communication abilities as well as other medical problems and their management, including seizure control and use of therapies; and effects of having a child with CDKL5 on the family.

*Visit our
website
for
details
on how
to enrol*

Research

As this is our first year as a registered charity our top priority is to better understand CDKL5 and along with that the challenges faced by a family living with CDKL5. We are currently involved in discussions with medical professionals about the possibility of setting up a CDKL5 specific clinic. This will hopefully benefit the child and family but also go a long way towards medical

acknowledgement and interest into our very rare disorder. More details will be available in due course.

In the interim we are currently writing professional and family guidelines for CDKL5 which we hope to have translated for the European community.

There are 3 main areas of research into CDKL5 disorders,

namely genetics, molecular biology and characterisation of the clinical phenotype. At present the International Foundation for CDKL5 Research are involved through funding in all of the aforementioned areas. CDKL5 UK are looking towards targeting our research efforts once we have achieved our other initial goals and understand how we can make a difference.

**Research is
the key to
unlocking the
world of
CDKL5**

Personal Corner

Congratulations to our Chair Carol-Anne Partridge for being anonymously nominated and then subsequently chosen as a finalist in the 'Western Gazette Pride of Somerset Awards', as a Brave heart Hero. The award ceremony will take place on 17th April and will involve them making a short video of her which in her words "is all good awareness".

The criteria for nomination was strict and included only those who have responded to an immediate need and significantly affected the lives of others, and those who have gone on to make a positive difference to the lives of others.

We are sure you will agree with us that our Carol-Anne definitely fits the bill. So we hope you will join us in wishing her the best of

luck for the ceremony and we hope she has a fabulous evening as she deserves it. No matter what the outcome, she is already a winner in our CDKL5 community.



**Finalist 'Brave Heart Hero'
in the Western Gazette
Pride of Somerset Awards**



Our Official Sponsors

CDKL5 UK are proud to be one of the official charities chosen by the Leicester Tigers for 2012/13. Leicester Tigers are one of the most successful and best-supported Rugby Union clubs in England. Martyn and Teresa Newey are working really hard fundraising at various events the club have .



CDKL5 UK are proud to be supported by the Spire Hospital Leicester for 2012/13.

Money raised for CDKL5 UK through sponsorship and fundraising will be put to our key aims:

- Funding research worldwide for treating the symptoms, including epilepsy, and research into the genetic cause of CDKL5 and disseminating or publishing the useful results of such research for the benefit of the public.
- Supporting participation into clinical trials relevant to treating the symptoms of, or development of a cure for CDKL5, or participation in clinical trials for similar Rett-Like disorders and Rett Syndrome.

Raising public awareness of CDKL5 and promoting a greater understanding of the condition within the general public and the medical and social professions.

We thank you both for your continued support

What's Going On In Europe:

This year, the third annual European Rett Syndrome Conference will take place in Maastricht, The Netherlands on the 17th to 19th October.

This will be a great central place for our families from across Europe to meet and get to know each other. The Conference will be a meeting ground for world class clinicians and professionals in Rett Syndrome and so a perfect opportunity for CDKL5 families to meet and raise awareness whilst having the opportunity to meet the experts and give them our unique atypical perspective.

CDKL5 UK, are currently producing professional and family guidelines. These booklets will be available in a number of different

languages in the hope of bridging the gap between the UK and the rest of Europe. We have found through our social networking, that there is a gap in information for those families where English is not their first language. We hope that by compiling and producing these guidelines it will reduce the isolation that some families face, and also bring the CDKL5 community in Europe that bit closer. Further information will be available on our website soon.

Europe is supported by three foundations. One that is long standing, and was set up in Italy, they continue to fund research, and were instrumental in starting researchers to think about Mouse Models. They recently has funded a Protein Replacement Therapy project, which will last for three years,

The second, a organisation that has been set up in the Netherlands, they hope to financially support families, as well as fundraise for research.

Finally, Rett Syndrome and CDKL5 Ireland fundraise to support the research effort as well as families affected by Rett Syndrome and CDKL5.

Please visit www.cdkl5.org For the Italian Foundation and for the Netherlands visit www.cdkl-5.nl, and for Ireland www.rettssyndrome.ie



***COMING**

SOON*

ONLINE SHOP

Merchandise

available check

our Facebook

page for details

Focus on Therapy

We are often asked, “what can I do to help my child achieve head control; sit; walk etc”. Within the UK, as we have fabulous special needs schools, and therefore a lot of therapy is conducted within school. However, in the last couple of years, a specific therapy has been spotlighted by a couple of our families. Spider Therapy, is a intensive physiotherapy program, originally from Poland, but now available in the UK at a centre called FootSteps, which is based in Dorchester-on-Thames, near Oxford. By means of elastic ropes, the child is supported and therefore the therapists are able

to isolate various parts of the body, depending on what area they are focusing on for the child. They always work at the child's highest level, so if it is to achieve sitting, they will work to that, if it is to stand, then standing, and then to walking. In addition to the Spider, the child also undergoes intensive physio session, this maximizes the potential. Therapy at footsteps is offered for a three week block period, initially 2 hours a day, 5 days

a week. For further details please visit their website, www footstepscentre.com or contact us at CDKL5 UK for further information, or testimonials on its effectiveness with a child with a diagnosis of CDKL5.

Our very own Amber Partridge, has achieved independent walking within the last year aged nearly 8, and we and her parents UK couldn't be more proud at her determination.

“We never thought she would walk, but she proved us and them wrong thanks to intensive therapy”

Diary of Events

We have a number of events that are happening this year here is just some of them. As always further details of how to sponsor or further details are available on the website under events.

Karl's Wild Warrior Events— Karl is embarking on some pretty challenging assault courses all in support of Ellie Newey, and CDKL5 UK, so far he has completed one 10K event!

Paul's Challenge for Gracie Skillicorn—In support of Gracie, Paul a local firefighter in Winsford Cheshire, is setting himself a number of running and cycling challenges, including Northern Beast Assault Course followed by Run the Bridge.

5kx50 Challenge starting 31st March—Run, walk, swim, jog, cycle, or even row 5K for 50 days the second time out for this event. Why not join the team its not too late !

19th April—Charity Golf Day and Party Night, at Channels Golf Club in Chelmsford Essex in aid of Evie's Angels and CDKL5 UK

1st June—Summer Dream Ball, Grand Hotel, Bristol

Our justgiving page can be found at
www.justgiving.com/CDKL5UK

What else can you do?

Download the giveasyoulive app on your computer, every time you click, and buy goods online, you can raise money for CDKL5 UK at the same time. We are already achieving a good return. Just visit www.giveasyoulive.com and search for CDKL5 UK

You can also set up regular giving through Justgiving, details above, or give one off donations. Don't forget it is easy to set up a team, or a individual page for any event that you are planning. Justgiving has been the biggest success so far for us and we couldn't have achieved what we have without it.

Contacts

Peer Support

The trustees of CDKL5 UK are happy to provide peer support to families, and also give further information to professionals upon request.

For South West/Wales/Scotland
the rest of Europe

Carol-Anne Partridge
carolanne@cdkl5uk.org

For South East

Mandy Black
mandy@cdkl5uk.org

For North and Scotland

Sarah Skillicorn
sarah@cdkl5uk.org

For Midlands and East

Teresa Newey
Teresa@cdkl5uk.org

For Ireland

Paula Connolly
psconnolly@eircom.net

International Family Support Contacts

These are available from
www.cdkl5.com

Support Groups

www.facebook.com/groups/cdkl5

Facebook Page

www.facebook.com/cdkl5uk

Twitter

[Twitter.com/cdkl5uk](https://twitter.com/cdkl5uk)

Thank You

We would like to take this opportunity for the support that you have shown us since we began. We will continue to bring you updates on events; research; and latest news via our website. As we grow, we hope to bring more families together in the UK, and to continue to raise that vital awareness our community desperately needs. We have factsheets available, and if you would like some to distribute please contact Carol-Anne and she can arrange this. We have already embarked on a leaflet



CDKL5 UK

Fighting for the chance to flourish