SHWACHMAN DIAMOND SYNDROME UK

NEWSLETTER

SDS UK

WELCOME

Dear Friends,

Welcome to the late summer edition of the SDS UK newsletter. Our sincere apologies for the delay in sending this out to you and we thank everyone who has kindly contributed letters, stories, articles and updates.

We would especially like to thank our wonderful fundraisers for helping us to continue supporting our community, researchers and SDS projects, both current and future. This could not happen without your support, and we are truly grateful.

Please do get in touch (sds@sdsuk.org) if you would like to know more about the work we do and have planned; or feel you might be able to help us in our quest to make our SDS world a brighter, happier and, above all, healthier place to live. Together we are stronger.

Health and happiness wishes.

Andy, Heather, Julia, Kapil & Kim
SDS UK Trustees

INSPIRING READS IN THIS ISSUE:

USEFUL LINKS:

- SDSUK.ORG
- CONTACT - HELPFUL GUIDE FOR FAMILIES WITH DISABLED CHILDREN
- CEREBRA INFORMATION AND SUPPORT FOR PARENTS
- SDS ALLIANCE

CHAIRMAN OF THE MAB

PROF ALAN WARREN

Save the date....

10th International Congress on SDS that will be held at Robinson College in Cambridge from the 19th-22nd of April 2022

I hope you have all manged to stay well throughout this extremely challenging time.

I am pleased to report that there have been some exciting development on the research side. Some interesting papers have recently been published that provide insights into how the bone marrow cells in SDS patients adapt to the lack of functioning ribosomes, the machines that make all the proteins in our cells. The two studies, published in the journal Nature Communications (Kennedy et al 2021; Tan et al 2021), will improve our ability to monitor the health of the bone marrow in SDS. Our own study (Tan et al.) also reports the development of a robust SDS fly model. The SDS flies are very slow growing, but we were able to rescue the defects using the same strategies that we identified in the bone marrow cells from individuals with SDS.

Together, these studies provide a rationale for a drug development programme that is being driven by my lab in collaboration with a newly established spin-out company called SDS Therapeutics. These are very exciting and positive developments on the journey to find new treatments for SDS.

I am pleased to announce that we have set a new date for the 10th International Congress on SDS that will be held at Robinson College in Cambridge from the 19th-22nd of April 2022. Please put this in your diaries. More details to follow in due course!

I was pleased to recently host our first face to face adult clinic once again here in Cambridge. It was a great pleasure to meet some of you again. Thanks to the wonderful Natasha Jones, our dietician, whose valuable input is supported by SDS UK and to Phyllis Paterson, our clinical nurse specialist from Cambridge University Hospitals, in helping us hold this clinic.

I want to thank Kim Wright who is stepping down from SDS UK at the end of September, for all her dedication and hard work over the years.

Finally, I hope you are all able to get some rest and relaxation over the summer months.

Take care,
Prof Alan Warren

SDS CURE ROADMAP

WRITTEN BY JULIA HAWKINS
SDSUK CHARITY TRUSTEE

Toward a future where people with SDS live healthy and complete lives.

Toward a future where people with SDS live healthy and complete lives. SDSUK has led an effort to create a multi-year, multi-modality drug development plan, which will set up an organisation to make strategic investments in therapeutics readiness, clinical trial readiness and SDS-focused biotech startups.

SDSUK has partnered with Ethan Perlstein, Founder and CEO of Perlara, and has worked closely with Professor Alan Warren, PhD (University of Cambridge), Joan Mowery (President of SDS Foundation), David Grainger, PhD (Chief Scientific Advisor, Medicxi Ventures) and Yael Weiss, PhD (VP Business Development, Ultragenyx).

The team is now further engaging all researchers and clinicians and would love to involve patients and patient groups.

Information and Q&A sessions will take place during Q4, with the first session on 24 September at 5pm.
Please register here, we hope you can join us!

- Link to the first draft of the Roadmap
- Link to Summary, non technical version

SDS ALLIANCE SUMMER 2021 UPDATE

WRITTEN BY ESZTER HARS

Save the date....

The second annual virtual Global Fun Run/Walk to #CureSDS is in the works! We are aiming for the week of September 20th, 2021

Thank you SDS UK for inviting me to continue sharing the SDS Alliance's vision and progress with the UK SDS community! The work of SDS UK to support the UK SDS community is critical and inspiring, and we hope to be able to connect in person as soon as the pandemic allows. The SDS Alliance aims to supplement the efforts of local organizations with initiatives that go beyond a single country or region, in particular driving research and uniting the global SDS community. Here are some quick updates on recent and current projects:

- **Global SDS Awareness Week**. This past spring, we launched the inaugural annual Global SDS Awareness week. Our campaign included publishing several SDS family stories on our blog and social media channels. Don't miss Kim's story!
- **Adult SDS Patient Council**. Our international group of adult SDS patients is growing! The goal is not only to facilitate peer-to-peer support, but also to learn about what challenges adult patients face related to SDS and to come up with strategies to address them. Since the launch of the group in the fall of 2020, we have identified over 100 adults living with SDS. Contact us below if you would like to join.
- **Research tools for the SDS research community**. Science has made incredible progress in the last several years. I am sure most of you have heard about gene therapy and CRISPR! Our mission is to drive research so that our patients can get therapies and cures. We need to equip our scientific community with the tools they need to advance research into the clinic. The first critical piece of the puzzle in our strategy is to create a new mouse model for SDS. Read about the project we launched recently here https://www.sdsalliance.org/post/sds-mouse-launch and https://www.sdsalliance.org/post/mouse-model-meet-the-scientists
- Save the date! The second annual virtual **Global Fun Run/Walk to #CureSDS** is in the works! We are aiming for the week of September 20th, 2021. This year again it will be set up virtually so that anyone, anywhere in the world can participate. Follow our social media channels for updates.

Please don't hesitate to reach out to us with any questions or suggestions at connect@SDSAlliance or on Facebook.

Stay safe and healthy, everyone!

Eszter Hars, Ph.D.
President and CEO
Shwachman-Diamond Syndrome Alliance
www.SDSAlliance.org
www.facebook.com/SDSAlliance

CEREBRA AND CARDIFF UNIVERSITY PARTNERSHIP

Improving mental health support for individuals with rare genetic conditions

WRITTEN BY LOWRI ODONOVAN

Dear reader,

My name is Lowri, and I am a Research Associate working on a collaborative project between Cerebra and Cardiff University. I was grateful to write an article for the SDS UK Newsletter in December last year to introduce our work which aims to improve support for individuals diagnosed with a rare genetic condition associated with increased risk for developmental and mental health conditions.

Cerebra are a charity and support children with brain conditions and their families. Both Cerebra and the research team at Cardiff University (led by Professor Marianne van den Bree) recognise the need for better support for families with a child with a rare genetic condition, and specifically better mental health support.
In my last piece I wrote about some focus groups and interviews our team conducted to explore the challenges experienced by families and practitioners when trying to access and provide support for children. I also spoke about our project Steering Group, a group made up of parents and other key stakeholders from the health, social, educational and voluntary sectors, with whom we further discussed these challenges. SDS UK are represented in this group.

We concluded several key issues facing families and practitioners from our research (for example limited provision of early intervention/preventative support strategies to protect children's mental health) and now we are pursuing various lines of work to help address these problems. These include a dedicated Support Worker who will help improve children's quality of care and families' quality of life and for whom we hope to secure funding for before the end of the year. We are also co-producing a workshop about mental health in children with genetic conditions for carers and professionals to attend.

We hope these approaches will aid the accessibility of services for families via direct support provided by a Support Worker, but also by raising awareness of and informing parents and key professionals about mental health risks in children with genetics conditions.

Both areas of work are still relatively early in their development, but we look forward to taking them forward. If you would like to get in contact to discuss this project, please do! My email address is odonovanl@cardiff.ac.uk

Many thanks for reading!

Best wishes,
Lowri

CEREBRA
Working wonders for children with brain conditions

For Cerebra advice and support - parent guides click here

WELCOME TO HOLLAND.....

WRITTEN BY
EMILY PEARL KINGSLEY

"if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things about Holland..."

Welcome to Holland

I am often asked to describe the experience of raising a child with a disability – to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this...

When you're going to have a baby, it's like planning a fabulous vacation trip – to Italy. You buy a bunch of guidebooks and make your wonderful plans. The Coliseum, the Michelangelo David, the gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The stewardess comes in and says, "Welcome to Holland."

"Holland?!" you say. "What do you mean, Holland?" I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy.

But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to some horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.

So you must go out and buy a new guidebook. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.

It's just a different place. It's slower paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around, and you begin to notice that Holland has windmills, Holland has tulips, Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy, and they're all bragging about what a wonderful time they had there. And for the rest of your life you will say, "Yes, that's where I was supposed to go. That's what I had planned."

The pain of that will never, ever, go away, because the loss of that dream is a very significant loss.

But if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things about Holland.

Written by Emily Perl Kingsley

OUR STORY

WRITTEN BY LUCY CARMICHAEL

"Failure to thrive was a diagnosis that we were to hear again and again..."

Rob was born in the early 1980's. A time that there were less than a dozen surviving people with SDS and non older than In their early twenties. Weighing in at 5 1/2 lbs we were totally unaware of his tenacity, feisty attitude and a strong will to live.

'Failure to thrive' was a diagnosis that we were to hear again and again, but luck was on our side when Rob was 9 months old. Having spent all of those months in hospital we were lucky to have a new Senior Registrar who arrived one day on the ward. He examined Rob for over an hour and then told us that he believed he had seen similar observations in the child of a friend, and requested further tests to be done.

This proved correct, with a diagnosis of what was called Shwachman Syndrome, and so started Rob on Creon. This came as a powder in a large tub, which we were warned would burn his lips if you misjudged the amount you gave him. It turned all food to liquid and instantly separated his special milk in front of your eyes . Feeding became a race against the clock!...

Aged 3, this pint sized child started nursery. He made sure he was never left out of anything, was highly independent and insisted on being treated as a normal child throughout his education. Aged 6, Rob gained a brother, this was having had Genetic Counselling, not very advanced at the time, purely giving us the information that there was a 1 in 4 chance that we would have another child with SDS. This was not that helpful, but we went ahead regardless, as the importance of Rob having a sibling outweighed any complications that we already knew about and were currently coping with. His brother checked in at a healthy 7 1/2 lbs and has gone on, frequently to be introduced as Robert's 'Little, Big, Brother.'

Educationally it was never going to be a straight path as a result of his early hospitalisation and slow brain development. We very early on realised that Rob was severely numerically dyslexic. Times tables were mapped out on a large Lego base in differently colour bricks, almost still to no avail, telling the time and money were to become huge obstacles in Rob's life. Up to the age of 11 there were constant hospital admissions, appointments and procedures, but as he got older fortunately emergencies ceased to occur and he was lucky enough to spend his formative school years in the fells of Cumbria. Having achieved one A level he was accepted on a foundation course at West of England University, Bristol. He studied Social Housing living away from home and supporting himself.

Ironically, within the last six years, Rob's career took another turn. He has ended up working for the NHS in an administration capacity within a Gastroenterology Department, working in the hospital throughout the pandemic with not a day missed and never getting Covid. This job does come with advantages, as the nurses are fully aware of his syndrome, they are the first to drop off packets of Jaffa cakes onto his desk to 'fill him up.' His own hospital experiences have given him a good understanding of other patients.

Now in his late thirties , Rob continues forward with conviction and confidence in himself, which is what has made him the individual that he is.

TREASURERS UPDATE

WRITTEN BY ANDY OLDING

Thank you to everyone who has continued to fundraise and donate to SDSUK

The Charity is a small organisation, which has no employees and is entirely managed by volunteers. During the pandemic, this has been an advantage, as we have very low running costs and the fall in our funding has had less of an impact for SDSUK than for other charities.

The Trustees would like to thank everyone who has continued to fundraise for SDSUK, and in particular those who have raised money for us on Ebay, and using Amazon smile which provide a steady stream of regular income.

A particular thanks goes to Dan Cuffe and Caroline Hadfield who have raised over £4,500 for the Charity and completed the Bolton Ironman.

The charity has had some specific donations to support the development of a cure roadmap and a review of the SDS literature.

The Charity had £59,797 in cash, of which £9,819 is available after setting aside reserves and restricted cash.

In 2020, the Charity made a commitment of some £30,000 to the University of Cambridge to fund the patient registry. This will be paid over the next four years and has now been fully funded.

INCOME STATEMENT (1NOV 2020 TO 4 AUG 2021)

	£
Income	
Donations	27,270
Expenditure	
Cure roadmap and literature review*	(7,873)
Net Income	**19,397**

*funded by specific donations from Trustees

CASH AND FUTURE COMMITMENTS AS AT 4 AUG 2021

	£
Cash	59,797
Less restricted cash	(8,013)
Less reserve	(3,000)
Future commitments	
Cambridge 2021	(6,000)
Patient registry (2021to 2025)	(29,965)
Other	(3,000)
Cash less commitments	**9,819**

OUR FUNDRAISERS

some of the fantastic fundraising efforts to support SDSUK - if you have an event happening please do let us know!

Wigan 10k. Help Anna change the world! Make a donation now.

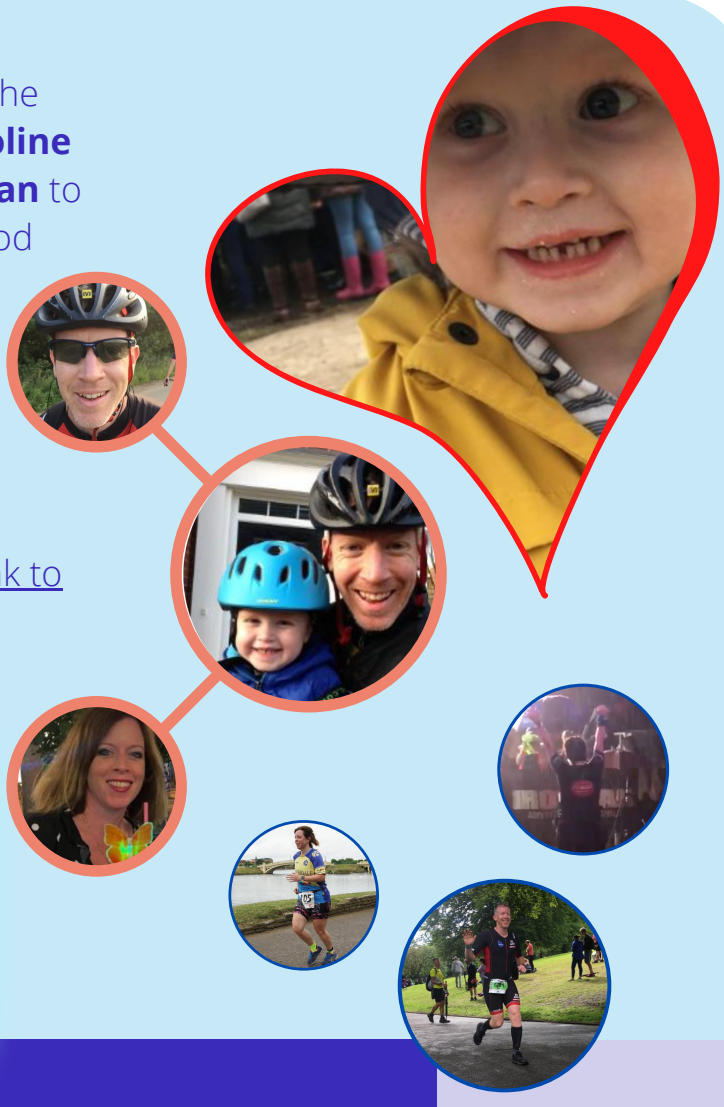
Anna Jeary is raising money for Charity. Sponsor them with

Congratulations Anna & Paul!!

Congratulations and a massive thank you from the SDSUK community to **Dan Cuffe** & his sister **Caroline** who **successfully completed the Bolton Ironman** to raise money & awareness of both SDSUK & Blood Cancer UK.

It was no mean feat completing the 2.4mile swim, 112-mile cycle & 26.2 mile marathon in July 2021 for such vital charities.

You can read Dan's "team story" by clicking this link to the Virgin Money Giving Page, Tri for Harry.



INSPIRATIONAL QUOTE

we want to leave you with this quote....goodbye for now!

the SDSUK team