

Letter from the Chair of Trustees

We hope everyone is having a good summer and enjoying the warm weather we have been having at times.

It is my privilege as Chair of SDS UK to introduce you to our inaugural newsletter which we intend to produce on a six-monthly basis and circulate via email and our website.

The current trustees have been working closely together for the past year and I feel we have made great progress in this short time. Our achievements to date have been to create a new website which includes news on research, access to MAB members and resource information which we hope you will find of great benefit. We have hosted a successful family day and are currently organising the family agenda for the next international congress, to be held in Cambridge next year. We will keep you closely informed and updated of all our projects and events.

My sincere thanks is given to those who have worked so hard in supporting SDS UK in their many different ways, with a special mention to our fundraisers. Fundraising is vital to the success and ongoing progress of the charity as it enables us to improve the services we can offer and the support we can give to patients, families and research.

I hope you enjoy this first edition of our newsletter!

Best wishes.

Keith

Who's who:

- For those not familiar with the Trustees here is a lovely photo of them.
- Keith Laughton is Chair and Trustee keith@sdsuk.org
- Andy Olding is Treasurer and Trustee andy@sdsuk.org
- Julia Hawkins is Trustee julia@sdsuk.org
- Kim Wright is Secretary and Trustee kim@sdsuk.org
- The Medical Advisory Board (MAB) consists of:
 - Professor Alan Warren (Chair), Dr Phil Anclif, Dr Peter Dale, Professor John Dodge, Dr Jonathan Kell, Professor Sally Kinsey, Dr Jutta Köglmeier and Phyllis Paterson.
- Contact details and further information on the MAB can be found at the website www.sdsuk.org



SDS Family Day

This April saw the first Family Day with members of the MAB in attendance. The day was a great success with presentations from the Trustees and MAB. It was chance for families to get together and share experiences and information. For the young adults with SDS it was an opportunity to get together and hold an informal workshop/brainstorming on the difficulties they have faced and how things could be changed to make life better, they kindly shared this with the attendees in a very candid and insightful way. For all families a lot of thoughts were exchanged and the evaluation forms that were completed were encouraging and positive. It would be nice if we could 'map' out our families across the UK to establish some informal regional get togethers, or just know who is around locally to meet up for a coffee.

See below some photos of the day - from left to right Professor Dodge explaining the background to the discovery of SDS, The MAB and Trustees and finally Professor Warren discussing SDS and the latest research.



Further to the updates given by Professor Warren into the current ongoing research, we have the following update from Trustee Julia Hawkins. 'We have set up a mouse model working group bringing together researchers from Canada, Holland and Sweden, UK and US who have all created different SDS mouse models. It's very difficult to recapitulate SDS in mice and this group is aiming to collaborate to progress this aspect.'



Letter from the Chair of the MAB

It is my great pleasure as Chair of the SDS UK Medical Advisory Board to contribute to this inaugural newsletter. Together with the amazing efforts of the Trustees, I think we have taken some tremendous steps forward over the last year.

Importantly, I am very excited to report that we will host the UK's first dedicated adult SDS clinic here in Cambridge at Addenbrookes Hospital in November this year.

On the research side, with the support of SDS UK, we have initiated an exciting new study that will help identify new biomarkers to guide the treatment of patients with SDS. With the help of the international advisory committee, I have also begun to put together an exciting list of speakers for the international SDS congress at Robinson College in Cambridge on April 6-9th 2020. I am pleased to report that the first Peter Durie Memorial Lecture will be delivered by Dr. Johanna Rommens, University of Toronto, who first identified mutations in the SBDS gene as the predominant cause of SDS.

Two important new research studies have been published this year. In a great collaboration between my own group, Dr. Patrick Revy and Dr. Jean Donadieu in Paris, we reported the identification of new SDS gene and an animal model that will potentially help guide the development of new treatments (Tan et al Blood 2019). A second paper from our group (Kargas et al eLife 2019) provides a better understanding of the function of the SBDS protein that is deficient in the majority of patients with SDS. The SBDS protein is dedicated to making ribosomes, the machines that make all the proteins in our bodies.

Thank-you for all your support and fundraising that will drive research and the much-needed advances in the care of patients with SDS.

Have a great summer!

Best wishes, Alan

International SDS Congress Cambridge April 2020

The dates for the 10th International Congress on Shwachman-Diamond Syndrome have been announced!

Save the date for 6th-9th April 2020. Professor Warren wants this conference to be inclusive for patients/families and we will be working with him, and the conference organisers, to ensure that the UK hosts an effective event for researchers, clinicians and patients/families.

Further information will be communicated via the next newsletter and the SDS UK website and it would be great to see as many of you there as possible. Your feedback is important to ensure the family aspect of the congress is correctly planned and catered for - are you interested in attending (with or without children); would you prefer to attend the entire conference or a meeting within the conference to get feedback on the latest research and outcome of the sessions that were held 6th to 9th April? Would you like to combine the meeting with a family activity?

Please contact Becky in the first instance at dauntseybecky@aol.com





We thought it would be a nice idea in each edition to include a contribution from our international SDS friends, for this edition the US group has kindly offered to give us an insight into their operations.

Greetings from the U.S.

I'm delighted to hear that you are launching a newsletter. It's such a great way to stay in touch with everyone.

Our group, SDSF, has existed a lot longer than your group, but it's great that you have a family support group for your families in the UK. We thought you might like to hear a few of the things that we do to help and advocate for our families. Our group is governed by a Board of Directors. All members are parents of SDS patients, except for our newest member, who is an SDS adult patient. Each board member is involved in a specific area of helping our group. These duties include fundraising, public awareness, communications, family education, and special projects.

In addition to our website, we have a private Facebook page where over 400 adult patients and parents can share experiences, exchange ideas and sometimes ask advice in a secure environment. We produce a quarterly newsletter which is emailed to over a thousand subscribers, including several physicians.

We sponsor a bi-annual, week-long family conference, where the entire family can enjoy themselves, get to meet other families, and hear from some of the SDS medical experts. We strongly feel that when a family member suffers from a chronic illness, it affects the entire family. We offer family support through our support page, helping to find doctors, our bi-annual conference mentioned above, and giving general help; answering questions that parents and patients have to give them peace of mind.

Each December, we have an exhibit at the American Society of Hematology Congress. This meeting is attended by about 25,000 hematologists and researchers from all over the world. We speak to as many doctors and researchers as we can to spread the word about SDS. Of course, we also sponsor and support the biannual International Scientific Congress on SDS, which will be held in April 2020 in Cambridge England. Along with this, we fund research projects that involve various aspects of SDS.

If I had a specific message for you, it would be: No matter how few there are in your group, every SDS patient and their family is important, and deserves as much attention and support as diseases affecting larger numbers of patients.

Keep up the good work. We'll look forward to hopefully seeing some of you at the Scientific Congress next year.

My very best to all of you, Joan Mowery

Fundraising News and the Stars behind them:



Fundraising ball. Saturday 9th November 2019 Peterborough

Sue Kilby's grandson was diagnosed with SDS at the age of 9, he's now 23. Like all families who get this very frightening and devastating news, it makes you feel helpless to say the least. At that time 14 years ago, SDS was practically unknown to the medical profession, so Sue and her niece decided over a cup of coffee to see what they could do to help. Not only for her grandson but for all the children who suffer with SDS.

So they started fundraising to raise the much-needed money for research. Over the years as a family they have done car boot sales, tombolas at different functions, bingo nights, baking and selling cakes and of course their charity ball!

They are now in their remarkable 14th year of fundraising and have raised over £110,000 to date!

The money was initially split between Great Ormond Street Children's Charity and Addenbrookes, purely for research into SDS. Now all of their fundraising goes to Addenbrookes Hospital under the care of Professor Warren for SDS research.

The theme for the Connor Wright Shwachman Diamond Project Ball this year is *Blame it on the* **BOOGIE**. The venue is the Holiday Inn West in Peterborough and the ticket price includes: Arrival drink and 4 course meal, Band, Disco, Gift Tree, Auction, Silent Auction, Games to raise money, Candy cart and **lots of FUN**.

Please support this event in any way you can, if you are unable to attend perhaps you could donate a gift/auction prize or make a donation. If you are able to do this, please contact Sue @ jkilby585@aol.com

Daniel Taylor, on 6th July 2019 Daniel and his friends hiked 50k in the Brecon Beacons to raise funds for SDS UK in memory of his beloved brother Tom. To date Daniel has raised an amazing £2242.81. You can read Daniel's story via the 'Support our Fundraisers' link on the website (www.sdsuk.org).

Georgina Surguy, ran the London marathon for SDS this year and has raised a staggering £9,699.00 to date. Congratulations! You can read Georgina's story via the 'Support our Fundraisers' link on the website (www.sdsuk.org).

Huge thanks is given to everyone who has fundraised for, donated to and supported Shwachman-Diamond Syndrome and SDS UK.

Connor Wright Shwachman Diamond Project
Presents

*Blame it
on the*
BOOGIE

GREATEST 70s PARTY OF ALL TIME

9th Nov 2019
7pm - 1.30am
Holiday Inn West
£55 per ticket

Enquire
Sue : 07894253931
Rachel: 07944939740

Adult Community News:

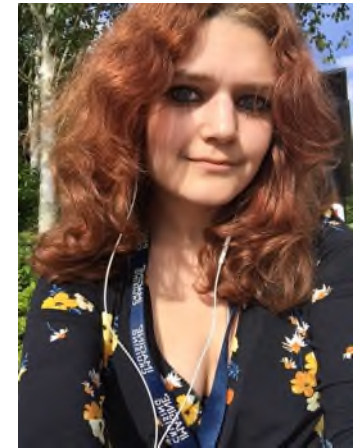
Hello, my name is Sophie and I am a 21 year old with SDS, I was diagnosed when I was 18 months old. I have the usual SDS gremlins such as blood, bone and digestive problems, but I would say my main day to day issue is chronic shoulder pain.

I remember at the SDS Family Day in April there were some concerns from parents with younger children around issues from schooling to travelling abroad. I attended a main stream school, it was at times a struggle and my teenage years were quite difficult. However, I did leave school with the GCSEs I needed to go to college and study accounting and business and I currently work as a sales administrator at Imagine Cruising.

Regarding travel, apart from the odd unplanned incident, such as developing chicken pox halfway across the English Channel en-route to France... things have been ok and I have visited North America, Cuba and The Seychelles – so it is possible. One useful travel aid we had when I was a child was powdered antibiotics that we took with us when travelling which could be reconstituted if required.

Part of the idea of this section is to gather the young and older adult SDS sufferers together, we have set up a Facebook page just for us, if you would like to be included please message Sophie Hazell or email sophieindauntsey@aol.com - particularly if you are another female as we seem a bit thin on the ground! 😊

We are currently planning a meet up in London, which hopefully will involve more than a visit to a pub (hence the need for more women!), if you would like some information please do not hesitate to contact me.



Useful Information:

A number of additions have been included on the SDS UK website within the 'Resources' section – please do check these out.

Our special thanks is given to Great Ormond Street Hospital for preparing the SDS 'Educational Psychology Information Leaflet'

Finally, please do share your stories, thoughts, ideas and any useful information you may have. Our next newsletter is planned for February 2020 and if you would like to contribute, please contact Becky Guy (Editor) at dauntseybecky@aol.com

'Health and Happiness wishes to you all'

