

## Letter from the Chair of Trustees

Six months have flown by since our inaugural newsletter and I, and the Trustees of SDS UK, would like to take this opportunity to wish you all a happy & healthy 2020.

We have been busy concentrating our efforts and resources mainly on the following events and activities, details of which are expanded upon within this newsletter.

- Supporting the SDS International Congress & Family Meeting in Cambridge, 6-9<sup>th</sup> April 2020.
- Supporting a dedicated SDS UK Patient Registry.
- Supporting the SDS Adult Clinic.
- Supporting a Junior Doctor Training Day, 24<sup>th</sup> April 2020.

We will continue to keep you closely informed and updated of all our projects and work-in-progress via the website, newsletter and minutes of meetings.

As before, my sincere thanks is given to all those who have supported SDS UK, with a special note of thanks to Professor Alan Warren for his unwavering dedication to Shwachman-Diamond Syndrome, SDS patients and their families.

Last but definitely not least, thanks is given to our wonderful fundraisers and their sponsors who have worked so hard to raise vital funds for Shwachman-Diamond Syndrome. None of the above would be possible without this generosity. Fundraising remains a crucial activity to ensure the ongoing future, progress and success of the group – please help if you can.

After lots of careful thought I have decided to step down as chair of SDS UK after the Congress in Cambridge, I am resigning as Trustee. It has been a great pleasure working with SDS UK again and working with a superb group of fellow Trustees. In a short period of time the charity has accomplished many achievements and I am sure this will continue over the coming years.

I urge you all to help and support the Trustees of this amazing charity to enable them to continue to support research and continue to work with and support you – the patients and families.

Best wishes.

Keith



### Who's who:

- Keith Laughton is Chair and Trustee [keith@sdsuk.org](mailto:keith@sdsuk.org)
  - Andy Olding is Treasurer and Trustee [andy@sdsuk.org](mailto:andy@sdsuk.org)
  - Julia Hawkins is Trustee [julia@sdsuk.org](mailto:julia@sdsuk.org)
  - Kim Wright is Secretary and Trustee [kim@sdsuk.org](mailto:kim@sdsuk.org)
  - Kapil Gotecha is Trustee [kapil@sdsuk.org](mailto:kapil@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 & further information for our Medical Advisers can be found on the website [www.sdsuk.org](http://www.sdsuk.org)

## SDS Adult Clinic

Tuesday 26th November 2019 was a momentous occasion for us - our first SDS Adult Clinic, something recognised for many years as a great need by SDS patients and their families. SDS paediatric care has been comprehensively provided through the SDS clinics at Great Ormond Street & Leeds and through our own local hospitals; but once NHS deemed adulthood age was reached, a gaping black hole in care and support awaited.

I thank all of you who shared your stories and experiences, which helped show the authorities at Addenbrookes the suffering this lack of care and support has caused to so many and provide them with additional evidence required to work on preparing a commissioning report to present to the NHS authorities.

The determination, dedication and support of Professor Alan Warren, Phyllis Paterson and Callie Ansell (who until your stories were shared were also largely unaware of the huge struggles and suffering) has undoubtedly initiated and moved forward the clinic at a faster pace. For this, I thank them with all my heart. The introduction of Tasha Jones to the team, our truly caring and superb dietician, has already made a positive difference.

Yes, the clinic has still to be commissioned to secure the services of the additional specialists required but momentum is gathering and the Operations Team are working hard behind the scenes with Prof Warren and Phyllis Paterson to secure this.

The next adult clinic will be held on Thursday 20<sup>th</sup> February 2020 and we look forward to seeing all the young adults and their families together.



## Junior Doctor Training Day about Shwachman Diamond Syndrome (SDS) (fully accredited)

*Please pass on details of this Training Day to all your healthcare providers.*

24 April 2020

9:30-16:00

Phoenix Court, 2 Brill Place, London, NW1 1EL (across the road from The Crick)

Please join UK's leading SDS paediatric and adult clinicians and researchers as well as SDS patients and charity.

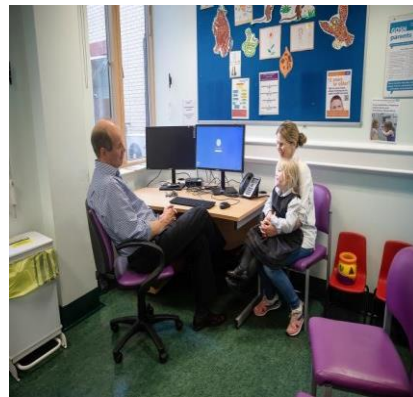
SDS is a rare genetic disease associated with a range of symptoms including poor digestion, failure to thrive, haematological abnormalities, reduced immune system, MDS, AML, developmental and psychological problems.

We are keen to raise awareness of the disease, especially to Junior Doctors, as we believe you may be the first to come across new cases and families who need to find the right care.

Please register using this link:

<https://www.eventbrite.co.uk/e/junior-doctor-training-day-shwachman-diamondsyndrome-tickets-89336912109>

<https://www.sdsuk.org/>



## Junior Doctor Training Day about Shwachman Diamond Syndrome (SDS) (fully accredited)

### Agenda

9:30 arrive for 10am start

#### Session 1: SDS and the paediatric MDT clinic

10:00 Julia Hawkins: Welcome and introduction to the day: SDSUK charity

10:15 Jutta Koeglmeier Clinical Presentation and Diagnosis of SDS: The Gastroenterologist's perspective

10:45 Phil Ancliff: Haematological complications

11:15 Natasha Jones: The importance of nutritional monitoring of children & adults with SDS

11:45 Coffee break

#### Session 2: Transition to adult care and the future of treatment

12:00 - 12:30 CNS Cambridge Transition to adult clinic

12:30 - 13:00 Alan Warren: Research and novel treatment strategies in SDS

13:00 - 14:00 lunch

#### Session 3: Patient and family perspective

14:00 - 14:15 Julia Hawkins: Parent's perspective of caring for a child with SDS

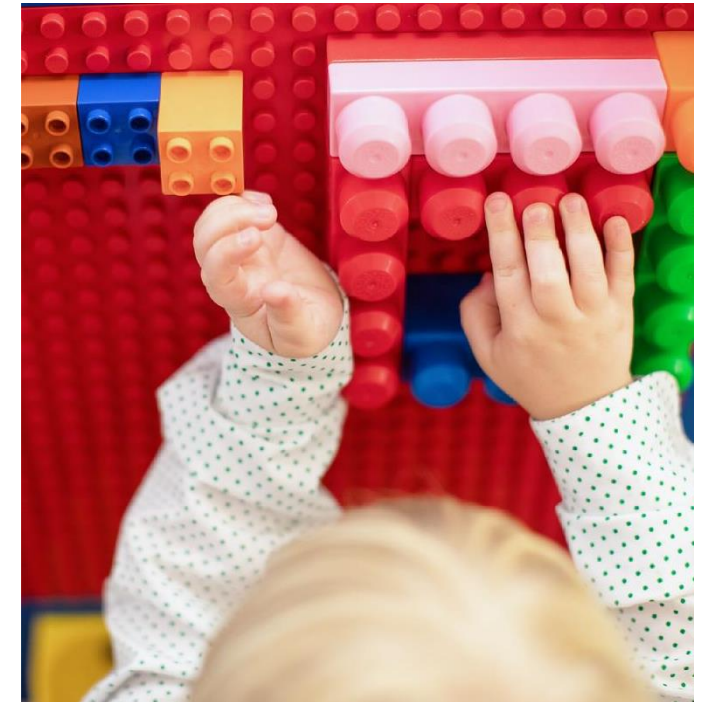
14:15 - 14:30 Kim Wright: Parent's perspective of caring for a child and young adult with SDS

14:30 - 15:00 Young adult's perspective of living with SDS

15:00 - 15:30 Group discussion

15:30 Julia Hawkins: Summary of the day and finish

<https://www.sdsuk.org>



## Letter from the Chair of the MAB

It has been a busy six months and we have much exciting news to share.

As some of you will know, the UK's first adult clinic for SDS is now in operation at Addenbrooke's Hospital in Cambridge, the first clinic taking place in November. Setting up the clinic has taken a lot of effort by many people over several years, but I would particularly like to thank SDS UK, Callie Ansell and Phyllis Paterson for all their help and support in making it happen.

SDS UK have also been crucial supporters of another very exciting project: the SDS Registry. Its aim is to collect data, including genetic material, from as many people with SDS as possible and make it available for research. It will be an invaluable resource for those studying SDS and a way in which everyone with SDS can make a contribution to understanding the condition and helping develop new treatments.

In October, my laboratory in Cambridge had the excellent news of the award of a £2.5m grant from the Medical Research Council to study how the function of ribosomes, the machines that make all the proteins in our body, is altered in SDS. We also want to understand how problems making ribosomes alters the function of the stem cells in the blood, so this is very important research for our community. I will be able to update you on the progress of this project in future newsletters.

Finally, I would like to invite you all to attend the International Congress on SDS that is taking place in Cambridge on 6-9 April (<http://www.sdscongresscambridge.com>). SDS families, together with all the leading SDS doctors and researchers from around the world will attend the Congress. This year will be the first time in fifteen years that the Congress will take place in the UK and so it is a once in a generation opportunity to attend. People with SDS can register for free while family members can register at a discounted daily rate. Monday, which has the special family sessions, the Peter Durie Memorial Lecture and the poster reception, and Wednesday, which has the clinical sessions and the gala dinner, might be the best days to come. I look forward to seeing you all there.

Best wishes,  
Alan

### **International SDS Congress Cambridge, 6-9 April 2020**

A banner link to the Congress can be found on the home page of the SDS UK website ([www.sdsuk.org](http://www.sdsuk.org))

We are holding a separate Family Meeting at the Congress for UK & International families on Monday, 6<sup>th</sup> April 12.00 – 2.30pm, please register your attendance for this by clicking on the link 'Family Meeting during International Congress' within the 'Upcoming Events' section on the website.

Below is a link address to a super quick survey for this meeting which will help us understand numbers and children coming, plus additional information to help the Trustees plan appropriately.

<https://juliahawkins777999.typeform.com/to/o8vmBk> (This survey is also included in the 'Upcoming Events' section of the SDS UK website)







## Fundraising News and the Stars behind them:

The Family of Nina Richardson would like to thank everyone involved in the fund raising activities that have been ongoing for the last few months, especially the efforts over the Christmas season which produced a fantastic result. Many people have been involved and assisted in the fund raising fun!

We have seen many locals express quite an interest in the SDS charity and how it supports families and those affected with the condition. Please continue to spread the word!



A huge thanks go to Ffi Jones (left) and all at Belcan in Derby for their “guess how many sweets in the jar” event along with a Christmas jumper day, a Christmas raffle, quiz and a dress down day which between them raised a fantastic £340. Even better news was to follow when Belcan, upon hearing of the amount raised, very generously doubled the money raised bringing the total to £680! A very big thank you to all involved!



*Ffi preparing  
The sweet jar!  
(she promised that she  
didn't eat any!)*

The Malt Shovel in Spondon ran several events including a 2<sup>nd</sup> hand book corner to buy books, a “guess how many sweets in the jar” event along with a similar “guess how many Lego bricks in the jar” which both created a lot of interested. There was a fantastic Christmas raffle with prizes donated from local businesses and residents of the village and a sale of Lego keyrings and donation bottle on the bar. Some of these activities are still ongoing and more are planned for the coming months. A very big thank you to our very good friends the landlady and landlord Karen & Mark Johnson and all their staff for their support and organisation of the events, their enthusiasm to help the charity was amazing!



*Nina showing off the  
Collection of items  
on the bar!*

Extra activities include ongoing charitable donations made via EBAY, with sold items donating 10% of the sale price to the SDS UK charity. You can find us on Ebay at 'biltomwife' & 'wakemeuponsaturday'

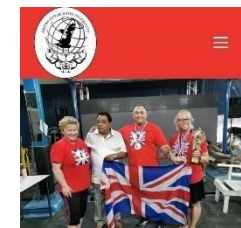
We have recently had some fantastic news from Marcus Griffiths of the British Powerlifting Federation who is hoping to raise money for Nina's charity in the upcoming British finals later this year, more info to follow. A huge thank you to Marcus and all involved for their efforts, I am sure Nina will be there to help cheer you all on!

A massive thank you to all involved for their support and for raising awareness of SDS, how it affects people and the good work that the charity has done for the people and families that suffer with this condition.

***total raised so far is a fantastic £1380.00!***

***A SMALL CHANGE CAN MAKE A BIG DIFFERENCE!***

***THANK YOU!***



*Marcus Griffiths 3<sup>rd</sup> from  
Left with staff from  
British Powerlifting  
Federation.*

## Adult Community News:



Hello, my name is Peter. I am 27 years old and have Shwachman-Diamond Syndrome (SDS). SDS has greatly affected my life physically, mentally, emotionally and socially. If it wasn't for SDS I would be like any other person in their 20's and be living a full and active life, unfortunately this is not the case.

### Useful Information:

A number of additions have been included on the SDS UK website within ['Upcoming Events'](#) and the newly named ['Useful Information'](#) section found under the link for Patients and Families – please do check these out.

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

In my childhood years I was an extremely active person and sport wasn't just my passion it was my life.

I loved everything about sport and still do, however I am no longer able to participate. I have had to overcome an array of setbacks and hurdles in all manner of ways.

I have unconditional love and support from my family although, I feel isolated and alone locked inside my own mind and body.

I can find it difficult to speak to family and unable to speak to friends about this condition, due to the rare, unrelated nature of this syndrome.

Generally, I have always felt as if I am a square peg trying to fit into a round hole.

I would like to gain awareness, understanding and ways and means both physically and physiologically of best living with and accepting my situation so that I may fulfil my life.

*'Thank you for Caring'*

