Shwachman Diamond Syndrome UK

NEWSLETTER

Welcome!

Dear Friends,

Welcome to the summer edition of the SDSUK Newsletter. We extend our sincere thanks to everyone who has kindly contributed.

We hope you are having a wonderful, relaxing summer - despite the inclement weather of late!

SDSUK has had a very eventful year and we wanted to use this opportunity to share our latest news with you.

Whilst it has been a very happy year in many ways, it has been a year of great sadness too with the heartbreaking loss of two of our precious family community, Ash (Pottle) and Rycroft (Foster). Our deepest sympathies and condolences are with their families, friends and loved ones.

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INSPIRATIONAL QUOTE:

'The best way to predict your future is to create it' - Abraham Lincoln

We are reminded of our mission to support SDS families and facilitate new treatments for patients:

In April, Prof Alan Warren hosted the 10th International Congress on SDS and welcomed the world's leading SDS researchers and clinicians to Cambridge, to progress research and scientific and clinical collaboration. It was fantastic to meet the many families who travelled great distances to take part in the discussions – all with the shared goal, to 'find a cure for SDS.' We are so grateful and proud to be part of this global community of patients, families, clinicians and researchers.

Last year SDSUK joined with five other bone marrow failure disorders as a member of the 'Together for Healthy Marrow Alliance.' This year heralded the start of a National Lottery funded collaboration project with our partner charities, 'Better Together for Healthy Bone Marrow.' This vital project will help us respond to the challenges faced by people with rare bone marrow conditions by working together to deliver mental health and wellbeing support to people in our community. Please see below for information about the project, what is available, and how to access it.

We have a number of exciting events coming up:
Runners for SDSUK are taking part in:
The Loch Ness Marathon on 1st October 2023 &
The Royal Parks Half Marathon on 8th October 2023

We send our thanks and best of luck wishes to everyone taking part.
Please do support our brilliant runners.

We send our immense thanks to all our wonderful fundraisers and their sponsors, who have worked so hard to raise funds for SDSUK. None of the work we do would be possible without this generosity. Fundraising remains a crucial activity to ensure the ongoing future, progress and success of the group. Your support is truly appreciated.

Finally, we can't wait to meet you at our Family Fun Day at the Hatton Estate near Birmingham on 19th November 2023. Our family fun day at Hatton Estate will include all day access to Hatton Adventure World and activities on the daily program, plus our own exclusive space and parking.

And if any of you would like to become involved as a Trustee or volunteer in SDSUK, we'd love to hear from you! Please email us sds@sdsuk.org

All our love and best wishes for your health & happiness.

Your team at SDSUK

SDS UK MAB

Read about the exciting and positive developments in SDS!

The next Congress will be in 2025 in Cincinnati, Ohio, USA

Chairman of the MAB

Professor Alan Warren

I hope this message finds you all well!

We were thrilled to host the 10th International SDS Congress at Robinson College here in Cambridge in April. The meeting was a great success with over 150 attendees including families, scientists and clinicians from around the globe, including China, Australia, Canada, USA, France, Spain, Belgium, Italy, The Netherlands, Germany, Czech Republic, Switzerland, Ireland, Israel, Japan, Mexico and of course the UK, with many new people joining us for the first time. Thanks to all the families in particular, as without your support and incredible fundraising, this event would not have been possible.

Some highlights of the meeting for me (apart from punting on the river Cam and the wonderful dinner at Caius College!), included meeting so many inspiring SDS families; the enthusiasm of the many new young investigators I met which brings so much hope for the future; setting up of many new international collaborations; important new insights into the basic biology of ribosome assembly; updates on gene therapy trials from Spain that are currently ongoing for the treatment of related marrow failure disorders which may find future application for SDS; trying to reach consensus on the nature and frequency of bone marrow surveillance and timing of transplant in SDS; updates on the mechanisms of progression to blood cancers in SDS. Lots more work still to do, but as a community we have made huge strides since the first SDS Congress back in Verona in April 2001!

There was also exciting news that a new clinical trial for SDS patients has just started in Italy, led by Dr. Valentino Bezzerri and Dr. Marco Cipolli. The trial builds on the work of this group to develop "translational readthrough-inducing drugs" that aim to improve expression of the SBDS protein.

Dr. Ron Kopito from Stanford University in the USA gave an inspiring scientific talk at the congress and we later learned at the Caius College dinner how he had been inspired from an early age by Dr. Harry Shwachman, one of the first physicians to describe what later became known as Shwachman-Diamond syndrome. Dr Kopito's story highlights the importance of finding great mentors, particularly for younger researchers as they try to get established.

We look forward to the next International SDS congress which will be hosted by Dr. Kas Myers in Cincinnatti in 2025!

Hope you will have some time to enjoy the current spell of good weather!

Take care,

Prof Alan Warren

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

SDSUK - Treasurer's update

SDSUK Treasurer - Andy Olding

SDSUK raised some £31,100 since the start of its financial year in November. Set against this were expenses of £9,600 which included payments to support the SDS International Congress in Cambridge. The Charity received specific donations to enable it to host a dinner in Cambridge for the International Family groups.

The Charity is a small organisation, which as it is entirely managed by volunteers, has very low running costs. We would like to thank Mark Irvine and Sophie Olley who organised a golf day, kindly sponsored by Richard Hawkins from RTH Staircases, that raised over £14,000 for the Charity. The Trustees would also like to thank everyone who has continued to fundraise for SDSUK, and in particular those who have raised money for us on Ebay and EasyFundraising which provide a steady stream of regular income. Amazon Smile has ceased its charity support programme.

We are delighted that so many people are committed to raising funds for SDSUK and we have teams running in the Loch Ness Marathon and Royal Parks half-marathon in October.

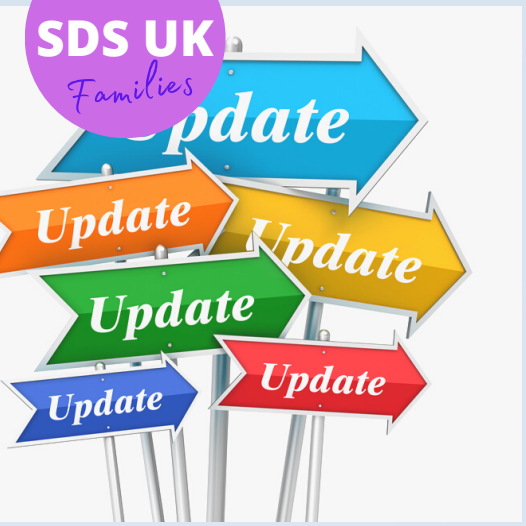
The Charity had £84,283 in cash, of which £44,597 is available after reserves and cash that has been set aside for specific purposes.

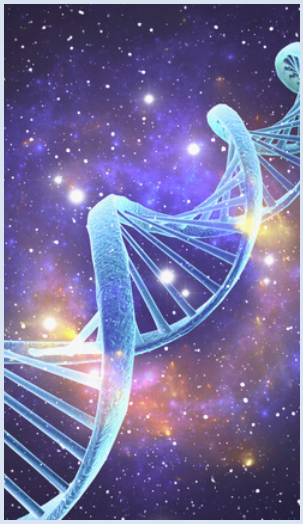
CASH AND FUTURE COMMITMENTS AS AT JULY 2023

	£
Cash	84,283
Less ring fenced cash	(3,717)
Less reserve	(3,000)
	77,566
Future commitments	
Patient registry (2023 to 2025)	(29,969)
Other	(3,000)
Cash less commitments	**44,597**

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 coming years and has now been fully funded.

SDS UK Families





Congress update - family friendly digest!

Report from the 10th International Congress on Shwachman-Diamond Syndrome, Cambridge, 18-21 April 2023

by Claudia and Richard Foxon

Dear Families and Friends of SDS-UK,

We hope that you and your families are well. In April this year we had the opportunity to attend the 10th International Congress on Shwachman-Diamond Syndrome in Cambridge. Both Richard and I have a background in biomedical research and we are very grateful to SDSUK for having given us the opportunity to attend the conference. It is our privilege to write here a brief summary of the congress, in simple terms, so that everyone can have a picture of the current research and findings in the field of SDS. We are reporting here areas that, hopefully, are relevant to us as families, parents of patients and patients affected by SDS. During the congress, there was a lot of in-depth research that is naturally relevant to scientists, but it was not at the stage where it has any direct relevance to us as families, at the present moment.

Top researchers and medical professionals, all working in the field of SDS, have gathered from all over the world to share their current research and findings, as well as share guidelines for surveillance of SDS.

There are several aspects that have been shared and discussed in the congress and we will look at them here one by one.

Nutrition

Dr Köglemeier, and Dr Kite (GOSH, London) have discussed in detail that Shwachman patients are prone to nutritional problems, not just through pancreatic insufficiency, but also due to antibiotic courses leading to poor appetite, infections, tube feeding, and vomiting especially related to chemotherapy/immunotherapy.

Shwachman children will grow according to their own growth curves which will often be on the low end of normal expectancy. Overfeeding to boost the child onto a higher growth curve is counter-productive. There are sets of growth curves specifically for Shwachman children, so paediatricians are not constantly comparing SDS children to unaffected children.

Creon 10,000 is the main supplement to replace digestive enzymes and is best given as a capsule, so early training of children to take capsules is important. Before that, the capsule can be opened, and the beads mixed with fruit paste. The beads are made from pig pancreas, which presents problems to some religious groups. Individual requirements should be discussed with gastroenterologists and dietitians.

Vitamins A+D are also often prescribed as supplements and there is a new version of these vitamins encapsulated like the Creon enzymes to protect them from stomach acid. However, these are much more expensive than the gelatin vitamin capsules so they are unlikely to be prescribed unless there are problems with the absorption of vitamins in gelatin.

A balanced diet, which also includes essential walnut oil as well as plenty of antioxidants (e.g., fresh berries), has been recommended.

Cognitive issues

Dr Kerr, a Canadian Neuropsychologist, discussed that neurocognitive issues are common in children with SDS, but that there is no recognisable pattern as to who will be affected or how. Common symptoms are slow learning, delayed development, attention deficit and impaired social skills. Parents should ask their paediatricians for a full assessment if they suspect similar problems. However, since these features are not unique to Shwachman children, there is every expectation that current support programs should be equally effective in supporting Shwachman children.

Haematology

Considerable progress was reported on the identification of the timing and range of genetic changes (called somatic variants) that are acquired by the bone marrow cells of patients with SDS (Prof Warren and Dr Shimamura). These genetic changes are as a consequence of the normal ageing process that happens in all our bone marrow cells. However, this kind of research is informing surveillance strategies that may ultimately be used to better stratify patients in terms of their risk of developing bone marrow disease, but a lot more work needs to be done before definitive recommendations can be made based on such data. Some consensus was reached on the frequency and types of surveillance that should form best practice in the clinic.

Novel Therapeutic Treatments

Several novel possible treatment strategies were discussed.

1. Dr Bezzerri, an Italian scientist working in Verona, presented a study on a drug called Ataluren which allows ribosomes to ignore some premature termination sites. In this way mRNA transcripts from the SBDSgene carrying the 183-184 TA>CT mutation could then be read to complete a full SBDS protein of 250 amino acids rather than finishing early due to the mutation mimicking a termination signal at amino acid 62. Ataluren is being used to treat a similar problem in Duchenne muscular dystrophy in Europe and is now being trialled on three SDS patients in Italy. As half of all known SDS patients have a copy of this 183-184 TA>CT mutation, this could potentially be of immense help.

2. The other common SBDS gene mutation is 258+2T>C, that causes a gene-splicing defect, but which appears to allow the synthesis of a small, but sufficient amount of SBDS protein to allow survival. Dr Cipolli and other Italian scientists are currently working on drugs to correct this splicing defect, although this work is still at the laboratory stage and it will be some years before a safe therapy will be ready for trial.

3. Shwachman patients are deficient in a protein called SBDS that is indispensable for the formation of ribosomes. Ribosomes are essential organelles present in cells that make proteins in our bodies. In Cambridge, Prof. Warren and his team are currently developing a molecule that can bypass the lack of SBDS protein, thus allowing the production of ribosomes even in Shwachman patients. This approach has the advantage that it could potentially help every SDS patient, rather than being gene mutation specific (as seen previously in points 1 and 2 by the Italian groups). However, there are reports that SBDS protein has other roles in the cell such as telomere protection (cellular ageing), which will not be assisted by any therapy that only corrects the ribosome biogenesis fault, but this is a minor concern compared to restoring normal levels of protein synthesis in the cell.[AW1]

Registry and future international events

Many countries are keeping a registry of their Shwachman patients (e.g., Canada, Germany, Holland, Italy, the UK, and the USA) detailing as much medical data as the patients are happy to allow. This information can be used to compare different treatment strategies and outcomes to better inform future clinicians. Notably, these registries have highlighted that Shwachman patients are unusually sensitive to the drugs used to prepare a patient for bone marrow transplant and shown that much weaker drug regimens have a better outcome.

The power of these registries is related to their size (patient number) and the depth of medical data recorded. While it is useful to compare results from the various registries to validate tentative conclusions, it has not been possible so far, to create a super registry of all the individual country registries, due to formatting differences between the registries and legal concerns over the confidentiality of the data.

At the closure of the Congress in Cambridge, it was announced that the 11th International Congress on Shwachman-Diamond Syndrome will take place in the Spring of 2025 in Cincinnati, USA.

Psychological support for our community made available through the BTFHBM project

Mental health and wellbeing support to people

Lesley Howells
Lead Clinical Psychologist
for Better Together

Managing Stress and Uncertainty

Free 12 week course for people with rare bone marrow conditions and their families.

Starts 23rd May

Other emotional wellbeing provisions on the horizon:

- Mindfulness courses
- Where Now? courses
- Potential to access counselling services

Sign up
www.theaat.org.uk/managing-stress

Ways you can get involved

SUPER RARE
BUT NOT ALONE

SUPER RARE is a fundraising campaign that runs February and March.

This year £3469 was raised for SDS UK.

THANK YOU!!

Community reference group

We are looking for another volunteer to be part of a community reference group as part of our Better Together Project. Please let us know if you would be interested!

The community reference group comprises a selection of volunteers who are affected by rare bone marrow conditions and the main purpose of the group is to help us to identify the unmet needs within the community which charities can potentially address. You will do this by sharing your story and your experiences as a patient or carer of someone affected by SDS.

Can you help? Share your voice

SDS UK Families

AUGUST 2023

To find out more contact the Better Together delivery team.

Claire
Project Manager
cbaggott@theaat.org.uk

Vicky
Community Engagement Manager
vburford@theaat.org.uk

Hannah
Project Officer
hmitchell@theaat.org.uk

Becky and Dan
SDS UK

Meet the new Emotional wellbeing Coordinator for SDSUK

Hi, I am Heather and I'm an SDSUK Trustee and the new SDSUK Emotional Wellbeing Coordinator for the Better Together for Healthy Bone Marrow initiative.
If you have any wellbeing concerns please get in touch with me at heather@sdsuk.org to discuss your needs and how the initiative can help you.
The courses mentioned below are available to our community so if you need us reach out - all contact is secure and confidential.

Managing stress and uncertainty course with Lorena Tuasis - 12/09/2023 19:00:00

Join us for our twelve-week course that helps you deal with the stress and uncertainty of living with aplastic anaemia and other rare bone marrow conditions. Wi...

Mindfulness Course with Lesley Howells - 28/09/2023 19:00:00

Mindfulness helps us find an extra energising dimension to our lives, so join us for our eight week

SDS UK Families

Community news

In this section read news from our community, ways to get involved, fabulous fundraisers, how to sign up to our families' day and more!

SAVE THE DATE

Join us at the Hatton Estate, 19th November 2023,Our family fun day at Hatton Estate will include all day access to Hatton Adventure World and activities on the daily program, plus our own exclusive space and parking.

SDSUK family DAY

Hatton ESTATE

click here to find out more

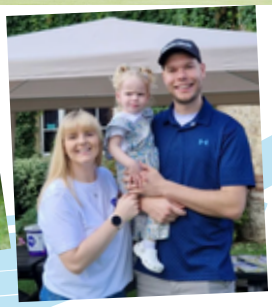
Congratulations

Congratulations to proud grandparents Christiane & Thomas, Mum Jana, who has SDS, welcomed Felicia Estelle on 18.06.23

SDS UK Families

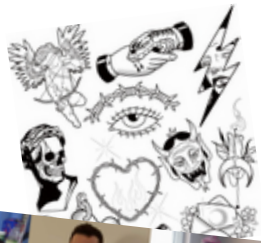
Fantastic fundraisers

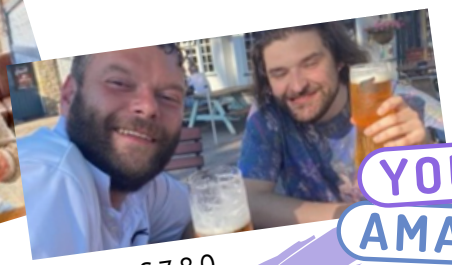
We now have a JustGiving checkout - 100% of donations go to SDSUK



SOPHIE, CHARLIE, OLIVIA AND FAMILY RAISED A HUGE £16,020 AT THEIR CHARITY GOLF DAY!

WELL DONE



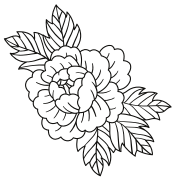


YOU'RE AMAZING

ADAM RAISED £780 IN HIS SOBER UP CHALLENGE

Thank You

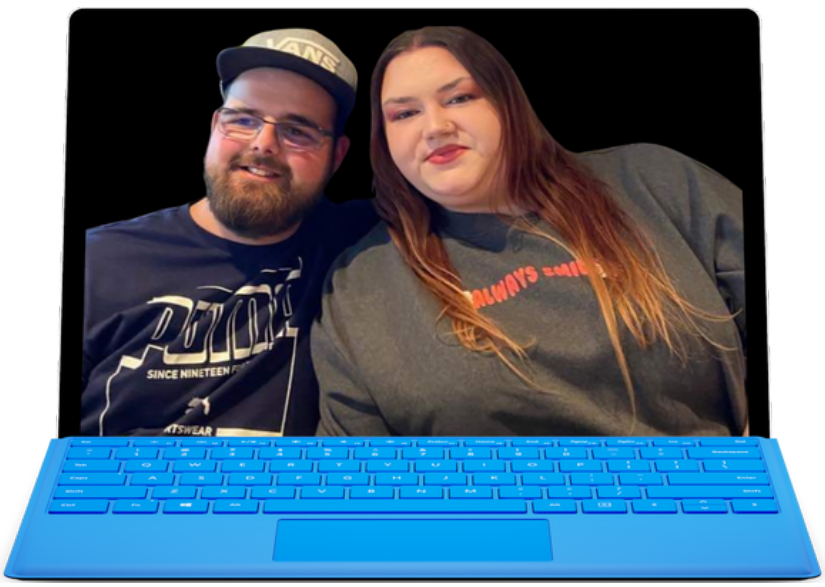
STACEY WALKER CONTINUES TO FUNDRAISE IN MEMORY OF ASH WITH HER FABULOUS TATTOO'S



£3,469 RAISED BY SDS SUPPORTERS IN THE SUSPER RARE CAMPAIGN

CONGRATULATIONS

TO CONNOR & KIRSTY



Their story is unbelievable:-

They met through online gaming 9 years ago.

Kirsty liked Connors gaming tag so they then started playing the same game & chat. This went on for a couple of years, they both then decided to exchange phone numbers & started chatting & texting. This carried on for 4-5 years until they both got very brave and decided to meet in person. Kirsty lived in Manchester, Connor lived near Peterborough. One Saturday Connor went to Manchester to meet up with Kirsty and here we are.

Kirsty came back to Connors home & has started a new life here. They got engaged recently & are like 2 peas in a-pod, they are so happy , spend their time having fun & laughter and even work together in the family business.

Hope that story is an inspiration to other SDS children /adults.

Great work Charlotte

DFN Project Search is a supported internship programme for young adults with a learning disability and/or Autism

featuring our very own Charlotte - SDS patient

Charlotte is working as a housekeeper on a maternity ward, making tea, juice and toast, cleaning and making beds. She would like to rotate to the children's ward next as she has a childcare qualification. Charlotte likes the opportunity to explore new areas of the hospital, meet the interns and make new friends.

DFN is a project by Mid Yorkshire NHS teaching trust partnering with Highfield School, Wakefield Council, HFT Supported Employment Agency, Pennine Camphill Community, Wakefield College, Kirklees Council and Kirklees College to give young adults with learning difficulties the chance to gain valuable work experience, in a programme called Project SEARCH.

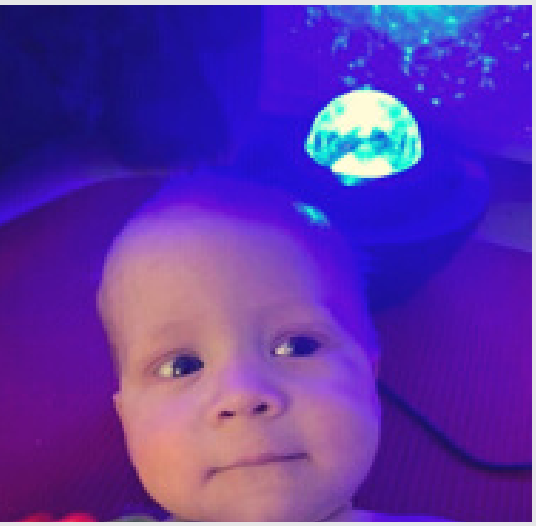
Do you want to know more about Project SEARCH - how to apply and more

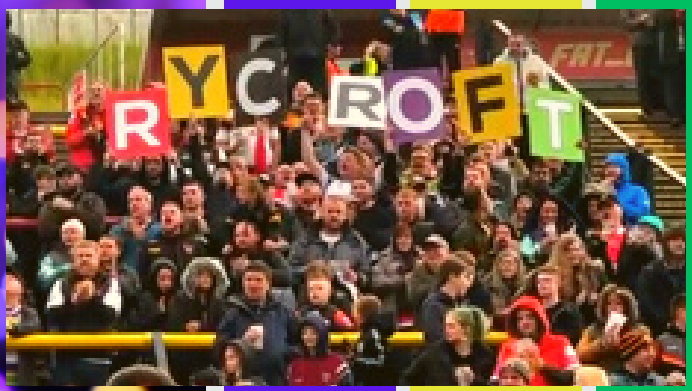
- follow this link: https://www.midyorks.nhs.uk/project-search/

Although Charlottes interview didn't make the one show cut - she is a shining star in our eyes.

You can watch her interview (approx 3:17 mins in) on YouTube here:

https://www.youtube.com/watch?v=hEZ73qkdIQQ





Rycroft Arthur Kenneth Alport Foster, 2022-2023

Rycroft was named after his great-grandfather, but it was his great-grandmother of whom he came to remind me most. Like Rycroft, her medical condition impacted her significantly, and caused her no little pain. But throughout her life, like Rycroft, she took delight in other people, on sight breaking into a smile like the sun parting the clouds.

And that was Rycroft too. Even in dark times, that smile would break across his face and you could stand there, warmed by its glow.

Rycroft liked to shout, a joyful, guttural roar. If you recall the episode of Blackadder where Hugh Laurie as Prince George is being trained in oratory, you will have a sense of it. He was a vital little boy and never more than when he was shouting.

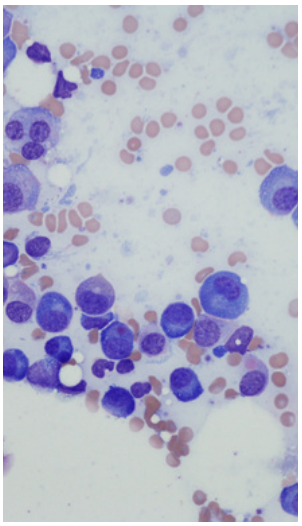
Rycroft liked to crawl. His condition kept his body small, and made it very difficult for him to raise his head from the floor. So he developed a manner of crawling on his back, a cross between butterfly and inverted breaststroke that was surprisingly speedy. A&E was one of the few hospital departments Rycroft never visited, but there were a few close calls at times where he would make an unexpected break for freedom at a height.

And Rycroft was a world champion sleeper, the Messi of sleepers. Give him a darkened room and he could be off in moments. BunBun, his favourite toy, was his loyal lieutenant in these endeavours. He would pull BunBun over his face, shading his eyes and signalling to anyone in the room that their presence was no longer required.

Goodbye Rycroft. Thank you Rycroft. We love you Rycroft.
We will always love you, Rycroft

By Andrew Foster, Rycroft's dad

AUGUST 2023



Do you know how to donate Stem cells?

September is Bone Marrow Awareness month

There may come a time when our loved ones with SDS need a bone marrow transplant and donors like you are vitally needed. Here in the UK there are a number of ways you can donate stem cells, each registry has slightly different criteria to meet but don't worry, you only have to sign up to one and they will anonymously share information.

Click on the links to the registry that fits you best- they are jammed packed with useful information and helpful advice explaining how you could potentially help save a life and scroll down for some quick facts.

To start its as simple as a cheek swab!

BRITISH BONE MARROW REGISTRY

www.bbmr.co.uk.

- male blood donors who are between 17 and 40 years old
- women who are aged between 17 and 40 and from black, Asian, minority ethnic or mixed (BAME) backgrounds

DKMS
WE DELETE BLOOD CANCER

www.dkms.org.uk

- aged between 17 (minimum age) – 55 (maximum age)
- in general good health
- Body Mass Index (BMI) under 40
- not already registered on another bone marrow registry
- UK resident at a permanent address for two years minimum
- haven't recently requested a swab online from DKMS.

ANTHONY NOLAN
saving the lives of people with blood cancer

www.anthonynolan.org

- aged between 16 – 30 to join their stem cell registry.

SDS UK Families

Stem cell donation quick points

HEATHER'S DKMS SWAB

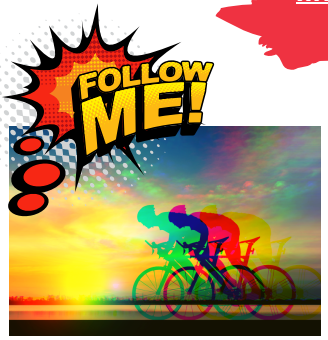
I didn't realise signing up to the bone marrow registry was as simple as taking a cheek swab! Watch my video and see just how easy it is

CLICK HERE

- signing up is super simple - its just a cheek swab
- you can opt out at any time
- HLA (Human Leukocyte Antigen) matching is how patients and donors are matched
- you only donate your stem cells if you're a match
- 90% of stem cell donations are peripheral blood stem cells (PBSC) and are taken straight from the blood stream!
- only 10% of stem cells are collected directly from the bone marrow in your hip bone while you're under a general anaesthetic
- costs are covered - loss of salary, travel costs, companions hotel stay and psychological support is offered

Take on the Swabtember Challenge with DKMS

https://www.dkms.org.uk/get-involved/events/swabtember-challenge-2023



SDSUK & BTFHBM team member Dan Cuffe is CyclingForSwabs! an epic challenge that will see him cycle through 12 countries to increase awareness of stem cell donations in the fight against blood cancers whilst cycling across Europe!

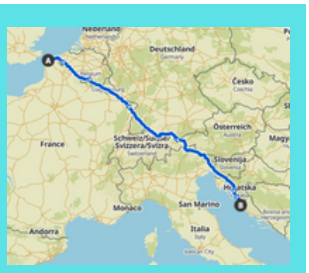
WOW!

LEG 1

Preston to Dover
486km

LEG 2

Calais France to Zadar Croatia
1,700km



SDS UK Families

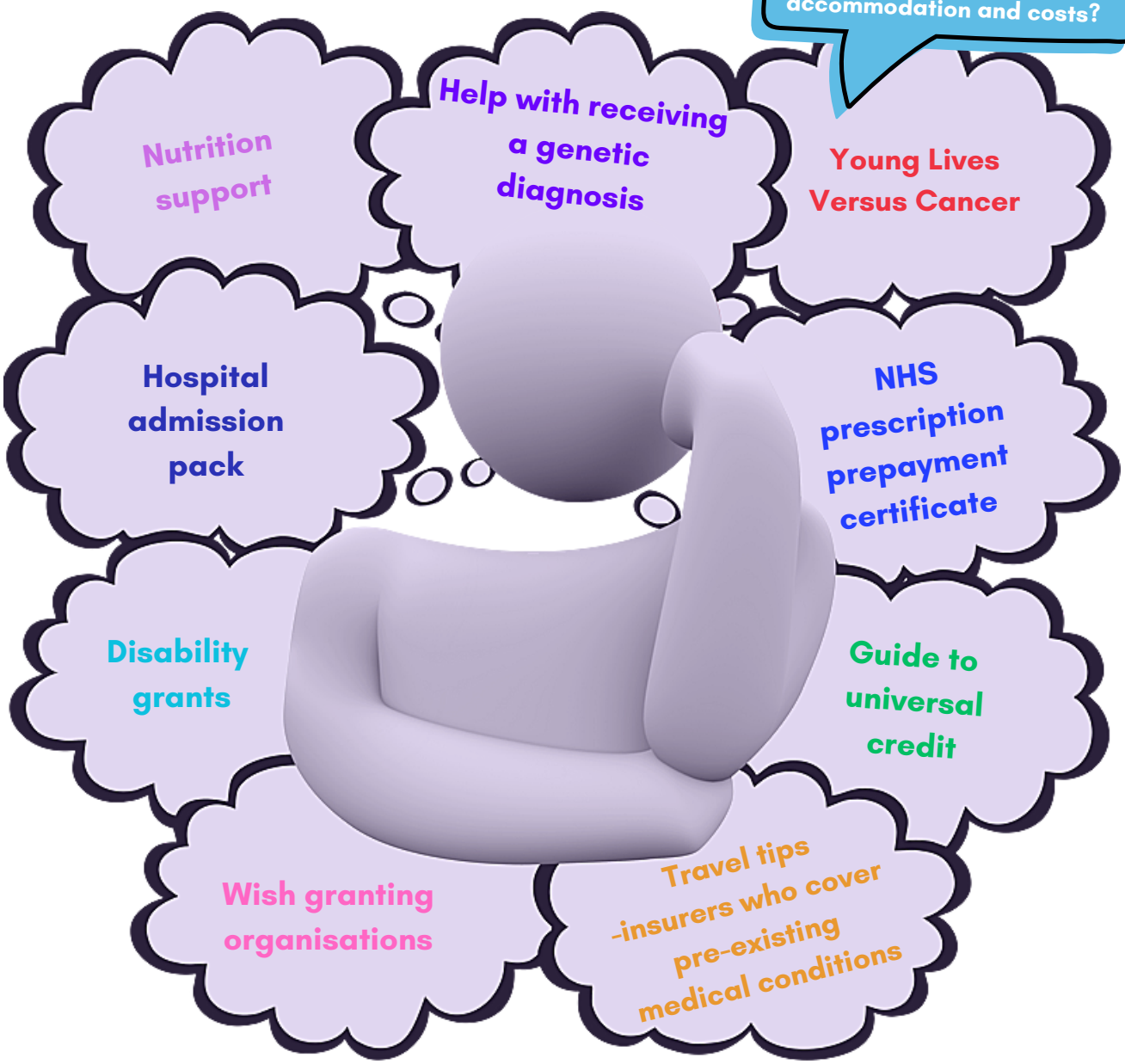
AUGUST 2023

Useful Information

HELP!

Please check out our useful information page at www.sdsuk.org/read-me its full of helpful advice

did you know that if you have to have a hospital stay for a bone marrow transplant they may be able to help with accommodation and costs?



AUGUST 2023

BULLETIN BOARD

JOIN US

TO DO LIST!

Look at www.sdsuk.org /read-me

Look in to stem cell donation

CHECK OUT EASY FUNDRAISING!

Check in on a friend

Download fundraising pack

Book my place

TAG US ON SOCIAL MEDIA

Diary Date
19.11.23
Hatton Estate
SDSUK Family Day

Family Fun Days

Family Fun Days are properly inclusive events that really inspire loyalty and build relationships in the happiest of Away-Day environments. And at Hatton...

HOW TO FIND US

5 minutes away from both the Midlands Motorway network and Warwick Parkway, with it's frequent train service to London and Birmingham; 15 minutes from Birmingham Airport and the NEC. A great location for the 21st Century hospitality.

HATTON ESTATE
HATTON, WARWICK CV35 7LD

TEL: 01926 844320
EMAIL: ENQUIRIES@HATTONESTATE.CO.UK