



# 10th International Congress on Shwachman-Diamond Syndrome

## Congress Programme



**18-21 April, 2023**

Robinson College, University of Cambridge  
Cambridge, England

**Chairpersons**

Professor Alan Warren and Dr Johanna Rommens



UNIVERSITY OF  
CAMBRIDGE

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## Practical Information

### **Abstracts**

To save on paper for environmental reasons, we are offering the abstracts online only. Please follow the QR code below to find a copy of all abstracts online.

### **Speakers**

All speakers should ensure their presentation is loaded and checked during the break prior to their talk.

### **Poster Presenters**

The poster sessions will take place on Tuesday at 17:00 and Wednesday at 12:45. You should display your poster on arrival at the venue and take it down after the last session on Thursday. Please note your poster number (pages 14-15) and display your poster on the correct board. Velcro is provided.

### **Punting and Gala Dinner - Wednesday 19 April**

Punting is £10pp and will commence at 17:45 for those who reserved a space.

The Gala Dinner will take place at Gonville & Caius College at 19:00 for those who booked a ticket. Find further details on the website at the QR code below. Come to the registration desk with any questions.

Dress code: Smart casual.

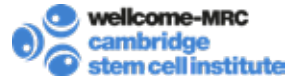
### **Wi-Fi**

Wi-Fi is available throughout the venue.

No password is needed.



# Generous Sponsors



## Award Winners



Many thanks go to the Quinlan Kohn Scholarship for supporting the following researchers:

Alex Faille, UK

Helen Reed, USA

Clarissa Gervasoni, Italy

Nic Robertson, UK

Vasileios Kargas, UK

Orna Steinberg-Shemer, Israel

Jane Koo, USA

Shengjiang Tan, UK

Apichat Photi-A, Canada

# Congress Chairpersons' Welcome

Dear friends and colleagues,

As chairpersons of the organising committees, it is our privilege to welcome you to the 10th International Congress on Shwachman Diamond Syndrome.

Much has happened since our last meeting in Houston in 2018, but we are grateful to be able to gather once more to continue the tradition of meeting to report outstanding new research and clinical developments from around the world relevant to those with an interest in SDS and related ribosomopathies.

SDS is a rare condition but it is the focus of research worldwide because of the impact it has on patients and their families, and the fascinating scientific problems associated with it. We are delighted to have assembled an outstanding array of speakers for the 2023 Congress, and the first Peter Durie Memorial Lecture, generously sponsored by SDS Canada. Johanna Rommens will give the lecture in memory of her much-missed friend and colleague.

We are proud to welcome you to the historic city of Cambridge for the 10th Congress, a city with a rich history of scientific discovery and progress. Boasting more Nobel laureates than any other University in the world, and with alumni like Isaac Newton, Charles Darwin, Philippa Fawcett, Rosalind Franklin, Stephen Hawking, and more, Cambridge is the perfect setting for a congress on scientific issues like SDS.

Scientific sessions will be held in the beautifully landscaped surroundings of 20th century Robinson College and its purpose-built conference centre, and we will gather for a gala dinner in the magnificent 16th century hall of Gonville & Caius College. A punting trip on the lovely river Cam and a chance to see some of Cambridge's outstanding sights are also on offer.

The Congress would not be possible without the involvement and sponsorship of the many outstanding organisations supporting people with SDS and their families. We would also like to thank all the invited speakers, chairs, and facilitators for lending their time and knowledge to help make the conference a success.

We welcome you to Cambridge for this fascinating and memorable event, and we hope you enjoy your stay.



**Alan J Warren**

University of Cambridge, UK



**Johanna Rommens**

The Hospital for Sick Children, Toronto

Co-chairs of the Organising Committee



## **SDS Programme 2023**

# Tuesday 18 April\*

11:00 Registration opens

## 12:00 Session 1: Joint Foundation Group/Science - Clinical & Research Advances for SDS

12:00	Welcome & Introduction to Family Session	Alan Warren
12:10	Welcome to Family Sessions	Julia Hawkins
12:20	Introductions from Families	Supporting Foundation objectives and immediate clinical/ research priorities
12:45	Introduction to SDS Research & Q&A	Alan Warren
13:15	Introduction to SDS Research & Q&A	Akiko Shimamura
13:45	Discussion	Working together, how to achieve goals
14:00	Break and refreshments	

## 14:30 Session 2: Future, Ongoing, Present & Past

Chair: Alan Warren

14:30	Alan Warren University of Cambridge	Welcome & Introduction
14:40	Johanna Rommens The Hospital for Sick Children	Peter Durie Lecture SDS: Looking back, and on toward the future
15:15	Alex Faille University of Cambridge	SBDS: Under the microscope
15:40	Peter Campbell Wellcome Sanger Institute	
16:20	Simone Cesaro Verona University Hospital	

## 17:00 Session 3 Poster Session: Patients, patient cohorts, and registries

Chairs: Adrianna Vlachos & Yves Pastore

17:00 **Drinks & Canapes Reception**  
**Poster session - see details on page 14**

18:30 Day 1 concludes

Delegates can order food from Robinson College's Red Brick Cafe Bar (open til 23:00) or find suggestions of local dining on our guide found at the link below.

\* Please note that this schedule may change from the time of printing. For updated schedules and resources for the congress, please follow the link from the QR code here:



# Wednesday 19 April\*

08:00 Tea & Coffee

## 08:30 **Session 4: Ribosomes: Structure & biogenesis** Chairs: Alan Warren & Victoria Cowling

08:30 Introduction to Session Alan Warren & Victoria Cowling

08:35 Sebastian Klinge  
Rockefeller University Principles of human pre-60S biogenesis

09:00 Vasileios Kargas  
University of Cambridge Regulating GTP hydrolysis during late cytoplasmic ribosome assembly

09:20 Vikram Panse  
University of Zurich Resolving a nuclear-mitochondrial targeting conflict during ribosome assembly

09:40 Ron Kopito  
Stanford University Ribosome UFMylation promotes 60S recycling and rescue from the endoplasmic reticulum

10:00 Victoria Cowling  
Beatson Institute for Cancer Research, Glasgow Ribosome production in T cells is co-ordinated by the RNA cap methyltransferase RNMT

10:20 Break and refreshments

## 10:50 **Session 5: Ribosome failures: Cell & organ consequences** Chairs: Alison Bertuch & Denis Lafontaine

10:50 Introduction to Session

10:55 Pierre-Emmanuel Gleizes  
Centre de Biologie Integrative, Toulouse 3' uridylation monitors exonucleolytic maturation of the 18S ribosomal RNA in the cytoplasm

11:10 Wei Tong  
Children's Hospital of Pennsylvania Regulation of Ribosome Assembly and Protein Synthesis during Hematopoietic Stem Cell Regeneration

11:25 Denis Lafontaine  
Universite libre de Bruxelles Mutations in the SSU-processome component C1orf131 cause ribosome biogenesis dysfunction leading to primary microcephaly

11:40 Timothy Olson  
Children's Hospital of Pennsylvania SBDS Deficiency in Bone Marrow Niches Impairs Donor Engraftment after Myeloablative Transplantation

11:55 Marc Raaijmakers  
Erasmus University

12:10 Kim de Keersmaecker  
KU Leuven Translatome analysis of somatic ribosomal protein mutations

12:25 Break

## 12:45 **Session 6: Lunch & Poster Session: Ribosomes, function and model systems** Chairs: Pierre-Emmanuel Gleizes & Ron Kopito

12:45 **Lunch Buffet**  
**Poster session - see details on page 15**



## 14:45 Session 7: SDS Phenotypes & natural history

Chairs: Johanna Rommens & Elizabeth Kerr

14:45	Introduction to Session	Johanna Rommens & Alan Warren
14:50	Elizabeth Kerr The Hospital for Sick Children	SDS: A Neurodevelopmental Disorder
15:10	Jane Koo Cincinnati Children's Hospital Medical Center, Dana Farber/Boston Children's Hospital	Neurocognitive outcomes in patients with Shwachman-Diamond Syndrome: a report from the Shwachman-Diamond Syndrome Registry and Study
15:20	Kezia Kite Great Ormond Street Hospital	Growth and supplementation in children with Shwachman-Diamond Syndrome
15:40	Lucy Fox Peter MacCallum Cancer Centre	
16:00	Yang Wan Institute of Haematology, Tianjin	Clinical characteristics of Shwachman–Diamond syndrome in China — result from Childhood Bone Marrow Failure Diseases Register of China Alliance for Blood Diseases(cBMFR-CABD)
16:15	Selected Abstract Orna Steinberg-Shemer	Genetic background and Clinical Characteristics of Congenital Neutropenias in Israel
16:25	Selected Abstract Petr Birke	The first Case of Acute Lymphoblastic Leukemia in a 7-year-old boy with Shwachman-Diamond Syndrome
16:35	Selected Abstract Helen Reed	Lymphoid Malignancy in Shwachman-Diamond Syndrome: A Case Series from the North American Shwachman-Diamond Syndrome Registry
16:45	Selected Abstract Clarissa Gervasoni	Study of angiogenic potential and metabolism in mesenchymal stromal cells derived from Shwachman-Diamond Syndrome patients and new therapeutic strategies
16:55	Selected Abstract Apichat Photi-A	Cellular and molecular mechanisms of the hematopoietic failure in Shwachman-Diamond syndrome
17:05	Session end	
17:45	Evening Social	17:45 - Punting Group departs Robinson College with Guide 18:00 - Punting on the River Cam 18:45 - All delegates wishing to go to Gala Dinner make their way to Gonville & Caius College
19:00	Drinks Reception & Gala Dinner at Gonville & Caius College	
22:00	Day 2 concludes	

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# Thursday 20 April\*

08:00 Tea & Coffee

## 08:30 Session 8: Genes, genotypes & phenotypes

Chairs: Akiko Shimamura & Pierre-Emmanuel Gleizes

- |       |   |  |
|-------|---|--|
| 08:30 | Yves Pastore<br>University Hospital Center Sainte Justine, Montreal | DNAJC21 mutations and bone marrow failure 3 overlaps SDBS  |
| 08:45 | Jean Donadieu<br>Hopital Armand Trousseau                           | EFL1 and SRP54   |
| 09:00 | Alison Bertuch<br>Baylor College of Medicine                        |  |
| 09:15 | Nic Robertson<br>University of Edinburgh                            | Mutations associated with Cartilage-Hair Hypoplasia impair ribosome synthesis                        |
| 09:30 | Lionel Blanc<br>The Feinstein Institute for Medical Research        | Revisiting the Hematopoietic Defects in Rps19 and Rpl5 Haploinsufficiency at the Developmental Level |
| 09:45 | Jutta Köglmeier<br>Great Ormond Street Hospital                     |  |
| 10:00 | Break and refreshments  |  |

## 10:30 Session 9: Clonal haematopoiesis in inherited marrow failure syndromes

Chairs: Jeffrey Lipton & Marc Raaijmakers

- |       |  |  |
|-------|--|--|
| 10:30 | Akiko Shimamura<br>Boston Children's Hospital/<br>Dana-Farber Cancer Institute | Biology and outcomes of clonal evolution in SDS: implications for surveillance |
| 10:50 | Alan Warren<br>University of Cambridge   | Convergent somatic evolution from early life in a Shwachman-Diamond syndrome   |
| 11:10 | George Vassiliou<br>University of Cambridge                                    | Causes and consequences of clonal haematopoiesis                               |
| 11:30 | Jean Soulier<br>Hopital St Louis   | Clonal hematopoiesis and leukemia in Fanconi anemia                            |
| 11:50 | Lucy Godley<br>University of Chicago   | Dynamics of clonal hematopoiesis   |
| 12:10 | Yigal Dror<br>University of Toronto  |  |
| 12:30 | Selected Abstract<br>Cornelia Zeidler  | Frequency of Somatic Mutations in SDS Patients                                 |
| 12:40 | Lunch buffet and additional poster viewings                                    |  |

**14:00 Session 10: Bone marrow challenges, transformation & leukaemia in SDS**  
Chairs: Timothy Olson & Lucy Godley

- 14:00 Stefan Meyer  
University of Manchester      Leukaemia in Shwachman-Diamond Syndrome - clinical features and management
- 14:20 TBA
- 14:40 Kasiani Myers  
Cincinnati Children's Hospital Medical Center      Updates and Emerging Therapies for High Risk Disease and malignant transformation in SDS
- 15:00 Carlo Dufour  
IRCCS Istituto Giannina Gaslini
- 15:20 Break and refreshments

**15:50 Session 11: Workshop / Panel Discussion & Q&A: Emerging practice for bone marrow surveillance in SDS**  
Chairs: Jeffrey Lipton & Stefan Meyer

- 15:50 Moderated Panel Discussion
- 16:45 Break and refreshments

**17:00 Session 12: Workshop / Panel Discussion & Q&A: Challenges & Considerations for bone marrow transplantation in SDS**  
Chairs: Carlo Dufour & Adrianna Vlachos

- 17:00 Moderated Panel Discussion
- 17:55 Break
- 18:15 Feedback meeting: Patient & family foundation groups
- 18:45 Day 3 concludes

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# Friday 21 April\*

08:30 Tea & Coffee

## 08:50 Session 13: Emerging & therapeutic approaches & strategies

Chairs: Valentino Bezzerri & Johanna Rommens

08:50 Intro & administration

09:00 Susana Navarro  
CIEMAT, Madrid      Lentiviral-mediated gene therapy for Diamond-Blackfan anemia:  
one step closer to the clinic

09:20 Selected Abstract  
Sabine Mellor-Heineke      G-CSF-Therapy in European Patients with Shwachman-Diamond  
Syndrome

09:30 Valentino Bezzerri  
Integrated University Hospital,  
Verona      Development of novel therapeutic approaches for SDS

09:50 Selected Abstract  
Shengjiang Tan      Fruit fly avatars for personalised medicine in Shwachman-  
Diamond syndrome

10:00 Maxim Rossmann  
University of Cambridge

10:20 Break and refreshments

## 10:40 Session 14: International collaboration, new research questions, summation & farewell

Chairs: Alan Warren & Julia Hawkins

10:40 Report on registries, recommendations & suggestions

11:00 New research questions & international collaboration

11:20 Julia Hawkins  
SDS UK      UK Registry & Tissue Bank

11:50 Alan Warren  
University of Cambridge      Summary, last words & farewell

12:15 Lunch

Food will be provided as takeaway option; delegates are welcome to stay at Robinson College or take their lunch elsewhere.

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## **Poster Sessions 2023**

# Poster Session Tuesday 18 April, 17:00

## Patients, patient cohorts, and registries

1	<b>Clarissa Gervasoni *</b> Tettamanti Center, Fondazione IRCCS San Gerardo Dei Tintori	Study of angiogenic potential and metabolism in mesenchymal stromal cells derived from Shwachman-Diamond Syndrome patients and new therapeutic strategies
2	<b>Nicholas Gloude</b> Rady Children's Hospital San Diego, UCSD	Immunological Function in Children with Shwachman-Diamond Syndrome
3	<b>Jane Koo*</b> Cincinnati Children's Hospital Medical Center, Dana Farber/ Boston Children's Hospital	Neurocognitive outcomes in patients with Shwachman-Diamond Syndrome: a report from the Shwachman-Diamond Syndrome Registry and Study
4	<b>Sabine Mellor-Heineke*</b> University Hospital Tuebingen, Germany	G-CSF-Therapy in European Patients with Shwachman-Diamond Syndrome
5	<b>Petr Birke*</b> Palacky University Olomouc, Czech Republic	Cellular and molecular mechanisms of the hematopoietic failure in Shwachman-Diamond syndrome
6	<b>Helen Reed*</b> aDana-Farber Cancer Institute/ Boston Children's Hospital	Lymphoid Malignancy in Shwachman-Diamond Syndrome: A Case Series from the North American Shwachman-Diamond Syndrome Registry
7	<b>Orna Steinberg-Shemer*</b> Felsenstein Medical Research Center, Petach Tikva, Israel	Genetic background and Clinical Characteristics of Congenital Neutropenias in Israel
8	<b>Shengjiang Tan*</b> University of Cambridge	Fruit fly avatars for personalised medicine in Shwachman-Diamond syndrome
9	<b>Daniel Turudic</b> University Hospital Centre Zagreb	A child with hemophilia A and Shwachman-Diamond syndrome
10	<b>Cornelia Zeidler*</b> Medical School Hannover	Frequency of Somatic Mutations in SDS Patients

\*Also presenting an oral presentation

# Poster Session Wednesday 19 April 12:45

## Ribosomes, function and model systems

11	<b>Marije Bartels</b> University Medical Centre Utrecht	Cytokine receptor-like factor 3 regulates platelet production via the Hippo pathway and tubulin modification, offering a new therapeutic target for myeloproliferative disease
12	<b>Alexis Bertrand</b> Imagine Institute/Inserm	Functional analysis of somatic genetic rescue in Shwachman Diamond syndrome
13	<b>Christian Boni</b> Azienda Ospedaliera Universitaria Integrata di Verona, Italy	Preclinical evaluation of ataluren as therapeutic option for patients with Shwachman-Diamond syndrome
14	<b>Seth Corey</b> Cleveland Clinic	SBDS-EFL1-EIF6 constitute a critical pathway that cannot be rescued by TP53 loss in Shwachman-Diamond syndrome vertebrate models
15	<b>Christine Hilcenko</b> University of Cambridge	Impact of acquired somatic mutations on eIF6 protein structure
16	<b>Alejandra Lagos Monzon</b> The Hospital for Sick Children	Aberrant early hematopoietic progenitor formation marks the onset of hematopoietic defect in SDS
17	<b>Angela Mercuri</b> Dept of Mother & Child, Azienda Ospedaliera Uni- versitaria Integrata	Prospective study of notch contribution on defective haematopoiesis of patients affected by Shwachman-Diamond Syndrome
18	<b>Apichat Photi-A*</b> The Hospital for Sick Children	Cellular and molecular mechanisms of the hematopoietic failure in Shwachman-Diamond syndrome
19	<b>Giuseppe Sabbioni</b> University of Ferrara	Shwachman-Diamond Syndrome: phosphoprotein pathway implication for discovering potential therapeutic targets
20	<b>Nuria Sánchez Puig</b> Universidad Nacional Autonoma de Mexico	Deviant conformational changes on an EFL1 disease mutant
21	<b>Dritan Siliqi</b> Institute of Crystallography - National Council of Research	Molecular mechanism of SDS: a combined molecular dynamics/ small angle X-ray scattering study of selected point mutations in the Shwachman-Bodian-Diamond Syndrome Protein (SBDS) and Elongation Factor-Like 1 (EFL1)
22	<b>Roberto Valli</b> University of Unsubria	An engineered plasmid system bearing the mutation c.258+2T>C of SBDS gene mimics the effects of the isochromosome i(7)(q10) in HeLa cells
23	<b>Kenichiro Watanabe</b> The Hospital for Sick Children	The aplastic anemia and leukemia-related protein SBDS interacts with the DNA repair protein KU80

\*Also presenting an oral presentation



## **Speaker and presenter information**



# Speaker and presenter information

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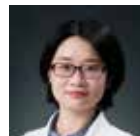
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# Robinson College



College Site plan

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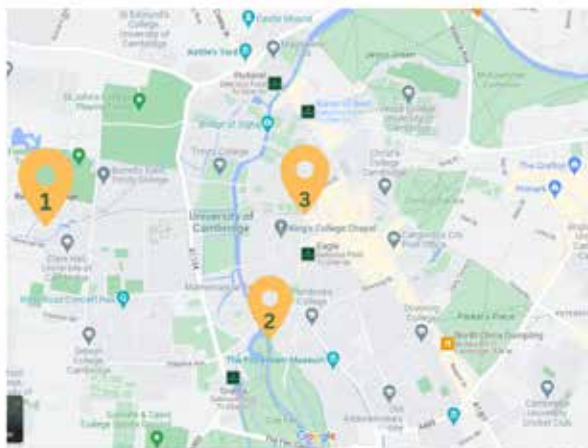


Main College Buildings

Look for and follow the signs to help you on your journey.

# Cambridge

-  **Robinson College**  
Congress Venue
-  **Scudamore's Punting,**  
Mill Lane Station
-  **Gonville & Caius College**  
Gala Dinner Venue



We hope you enjoy your time in Cambridge.

For details on transportation, dining and pub suggestions, and things to do, please follow the QR code to find the document with all the information you'll need during your stay!





**10th International Congress on  
Shwachman-Diamond Syndrome**

**Thank you for joining.**