

Shwachman-Diamond Syndrome

VOICE of the PATIENT REPORT



The official report from the Shwachman-Diamond Syndrome
Externally-Led Patient Focused Drug Development Meeting
held online and in Cincinnati, OH, on June 4th, 2025.

This report was prepared by the Shwachman-Diamond Syndrome Alliance (SDS Alliance) as a summary of the input shared by families and caregivers living with SDS during an Externally Led Patient Focused Drug Development (EL-PFDD) meeting, conducted as a hybrid meeting online and in Cincinnati, OH, on June 4th, 2025. SDS Alliance is a 501(c)(3) nonprofit organization serving the global SDS community to improve and save the lives of people affected by SDS by focusing on research and therapy development. More information at www.SDSAlliance.org/pfdd.

Authors and collaborators: This report was prepared and submitted on behalf of the SDS Alliance by Eszter Hars, Ph.D., Founder and CEO, in collaboration with Vanessa Merker, Ph.D., Massachusetts General Hospital and Harvard Medical School. Consulting Partners include Larry Bauer, RN, MA, and James Valentine, Esq. from Hyman, Phelps & McNamara, P.C. Community partners included the Shwachman-Diamond Syndrome Foundation.

Disclosures and funding: SDS Alliance was responsible for funding the EL-PFDD meeting and receives funding from the public, foundations, funding agencies, and life science companies in the form of unrestricted and restricted grants and sponsorship of programs and events.

Major funding was provided by the Chan Zuckerberg Initiative (Rare as One program), and the Patient-Centered Outcomes Research Institute (PCORI) through a Eugene Washington PCORI Engagement Award (EASO-42419). In return for financial support, these organizations were acknowledged on the meeting webpage. They did not have any input in the design, planning, coordination, or execution of the meeting or in the writing of this report.

James Valentine, Esq., and Larry Bauer, RN, MA, are employed by Hyman, Phelps & McNamara, P.C., a law firm that represents patient advocacy organizations and companies that are developing therapeutics and technologies to advance health.

Technical services: A/V and production support provided by Syndikast. Transcripts were prepared by HomeRow.

Report version date: May 12, 2026. **Published:** May 12, 2026

Revision statement: This document was not revised or modified after May 12, 2026.

Statement of use: The SDS Alliance has the necessary permissions to submit the “Voice of the Patient Report for Shwachman-Diamond Syndrome” to the FDA. Linking to this resource on the FDA website does not violate the proprietary rights of others. Permission to link from the FDA website is granted by the SDS Alliance.

© 2026 Shwachman-Diamond Syndrome Alliance Inc. This report is licensed under a Creative Commons Attribution 4.0 International License (CC BY 4.0). It may be freely shared, cited, and adapted with appropriate attribution. We respectfully request that the SDS Alliance be acknowledged if any part of this report is used or adapted. Organizations and companies intending to use this report for commercial purposes are warmly invited to contact us at patientvoice@SDSAlliance.org — we welcome the conversation.

Point of contact: Please contact Eszter Hars, CEO, Shwachman-Diamond Syndrome Alliance, or other members of the SDS Alliance team at patientvoice@SDSAlliance.org for questions related to this report.

Suggested citation: Hars E, Merker V. (2026). Voice of the Patient Report for Shwachman-Diamond Syndrome. SDS Alliance. <https://doi.org/10.5281/zenodo.20126868>

Acknowledgments: SDS Alliance extends our deep gratitude to everyone who helped to plan and implement the EL-PFDD meeting for SDS. We are grateful to have had this opportunity to ensure that patient and family perspectives are considered in drug development and regulatory processes, as well as in other aspects and stages of clinical and comparative effectiveness research.

Thank you to the many staff members from the US Food and Drug Administration who made time to attend the meeting or watched the meeting recording. Thank you to Dr. Martha Donoghue, Associate Director for Pediatric Oncology and Rare Cancers in the FDA's Oncology Center of Excellence, Office of the Commissioner and the Acting Associate Director for Pediatric Oncology in the Office of Oncologic Diseases, Center of Drug Evaluation and Research (CDER) for providing the welcome remarks and a thoughtful framing for the important role patients and families play in drug development. Thank you to Ethan Gabbour from the FDA's Patient-Focused Drug Development Staff, who guided us through the planning process and the preparation of this report.

Thank you to the many representatives from advocacy and professional organizations, pharmaceutical companies, federal agencies, and clinical and research centers from across the world who attended this meeting and who are working to ensure that we find a cure for SDS. We extend our appreciation to the investigators working in labs all around the world, striving toward a better understanding of basic and translational SDS science to move us closer to future clinical trials.

Thank you to Dr. Kasiani Myers, MD, Professor of Pediatrics in the Division of BMT at Cincinnati Children's and Co-Director of the North American SDS Registry for providing a concise and comprehensive clinical overview of Shwachman-Diamond Syndrome, and Dr. Alyssa Kennedy, physician scientist and assistant member at St. Jude in the departments of Hematology and Pathology, for providing a timely and compassionate overview of current and future treatment options and directions.

Thank you to Vanessa Merker, Ph.D., Massachusetts General Hospital and Harvard Medical School, for her support in preparing the pre-meeting survey, presenting a brief recap of the meeting at the end of the meeting, and her partnership in creating this Voice of the Patient report. Thank you to Grace Lynch, MS, for her careful review of the manuscript, drawing on her insights from working with our community on a qualitative research study (www.sdsalliance.org/lens), and her help on site at the meeting. Thank you to Ashley Ferreira, SDS Alliance, for her help with meeting execution, and the volunteers who ensured that the meeting ran smoothly.

Thank you to James Valentine and Larry Bauer from Hyman, Phelps and McNamara, whose assistance in planning and moderating the EL-PFDD was invaluable. Thank you to the Syndikast team for their production planning and all the behind-the-scenes work to ensure our meeting was a success.

Thank you to the SDS Foundation, our community partner, for helping us reach as many members of the SDS community as possible to ensure that everyone can contribute their voice. Special thanks to John Wall for the excellent communication and facilitation, and for helping run the microphones to patients and families so they could be heard.

And most importantly, we thank all the members of our worldwide community whose lives have been directly impacted by SDS, and who participated in the EL-PFDD meeting as attendees (in person or online), panelists, and who took the time to submit a written comment or fill out the pre-meeting survey or live polling. This includes the community of caregivers who shared their lived experiences, and adults living with SDS. This meeting was about you and for you and would not have been possible without each and every one of you. Thank you for making your voices heard.

Table of Contents

Executive Summary.....	4
Shwachman-Diamond Syndrome Overview.....	6
Meeting Overview.....	9
Topic 1: Living with SDS - Symptoms and Daily Impacts.....	12
Neutropenia, Immune Dysfunction, and Chronic or Severe Infections.....	14
Risk of Leukemia (MDS and AML).....	16
Digestive System Challenges and Exocrine Pancreatic Insufficiency.....	16
Skeletal and Orthopedic Issues.....	17
Fatigue and Stamina.....	18
Cognitive and Neurodevelopmental Impacts.....	19
Quality of Life and Mental Health.....	19
Topic 2: Patient Perspectives - Current & Emerging Treatments.....	21
Overview and Live Polling.....	22
Perspectives on Current Treatments.....	23
Priorities for Future Treatments.....	27
Risk-Benefit Considerations.....	32
Drug Formulation Considerations.....	32
Summary and Future Direction.....	34
Appendix.....	35
Agenda.....	36
Speakers.....	39
PFDD Discussion Questions.....	42
Meeting Recording and Photography.....	43
Live Polling.....	44
SDS Documentary Film: “Until There’s A Cure”.....	46
Patient Posters.....	47

Executive Summary

This Voice of the Patient (VoP) Report summarizes the experiences and perspectives of caregivers and patients living with Shwachman-Diamond Syndrome (SDS) shared during the Externally-Led Patient-Focused Drug Development (EL-PFDD) meeting on SDS, held online and in-person in Cincinnati, OH, on June 4th, 2025. The meeting was convened to share, document, and better understand the lived experience of SDS, the burden of disease, patients' perspectives on current and emerging treatments, and the community's priorities for future drug development.

Topic 1: Living with Shwachman-Diamond Syndrome (SDS)

Patients and caregivers described SDS as a multisystem, lifelong disorder that has profound physical, emotional, developmental, and social impact — not only on the person diagnosed, but on their entire family. Chronic infections disrupt schooling and careers. Feeding difficulties and poor growth shape children's early years. Skeletal complications may limit mobility and require repeated surgeries. Cognitive and neurodevelopmental challenges go unrecognized and underserved. And all of it is largely invisible, leaving families to constantly explain the disease to their networks and even healthcare providers.

The risk of serious infections shapes everyday decisions, especially in childhood — from where families travel, to whether a child can attend school, to what career parents can pursue. As Nicole, mother of ten-year-old Roman, put it: "SDS doesn't just steal health — it robs childhoods, careers, and stability."

Above all, the fear of leukemia is ever-present. Patients and families described it as an acute stress they live with every day. Patients also fear the bone marrow biopsies used for assessing if there is progression to leukemia. As Cresta, an adult living with SDS, put it: "It causes me anxiety to wait weeks for the results and wonder if it will be positive or negative and if I will be facing death soon."

Topic 2: Patient Perspectives on Treatments

Participants described treatments that have improved lives — but significant gaps remain. There are no therapies that address the underlying cause of SDS. Available treatments target symptoms as they arise, without options to prevent them, and many symptoms have no treatment at all. The need for disease-modifying therapies that can prevent complications is high and urgent.

Patients and families consistently prioritized reducing the risk of leukemia; developing alternatives to transplant, or making it safer; treatments that patients — especially children — can tolerate; improving growth and reducing skeletal issues; and enhancing the ability to participate in school, work, and everyday life. They are asking for treatments that are safe and effective — where safe, for

this community, means above all else not increasing the risk of leukemia. Leukemia in SDS is almost always fatal; Hematopoietic stem cell transplant (HSCT) is the only option to prevent it if used before malignant transformation, but it carries an enormous treatment burden and significant risks. As one parent put it: “Yes, what we’re doing is helping. But I want that fear removed — that this could turn into something worse. Why is transplant our only hope?”

Patients and families emphasized that the formulation — how a treatment is delivered — can have a profound effect on its utility. Many SDS patients have a history of regular injections, procedures, and complex medication schedules, and may struggle to adopt additional ones due to medical trauma. As Maria, mother of eleven-year-old Felicity, put it in the context of her daughter’s daily G-CSF injection: “Half the benefit without the fear and trauma we face every day? Sign us up.” Oral formulations may be more feasible in the SDS pediatric population than typically assumed — as many patients already swallow multiple capsules with every meal starting in childhood — but there are also many children with SDS who are tube-fed or have oral aversions.

This report documents what patients and families have experienced and shared — and what they are asking the research and regulatory community to help them with. As a longtime SDS mom who has cared for her child for over 50 years has summed up: “I hate transplants. I hate Neupogen. I hate it all. But there is progress. Keep pushing.”

Shwachman-Diamond Syndrome Overview

Shwachman-Diamond Syndrome (SDS) is one of the most serious inherited bone marrow failure disorders, yet it remains underdiagnosed and undertreated. A substantial proportion of patients will develop leukemia over their lifetime – 30% by age 30, and it continues to rise thereafter. Once leukemia develops in SDS patients, outcomes are extremely poor. The window of opportunity for treatment is narrow, and missing it is often fatal.

SDS is a rare genetic disorder that affects thousands of people in the US and globally. Many patients remain undiagnosed due to the variable presentation of symptoms and challenges accessing the right genetic testing or expert healthcare providers.

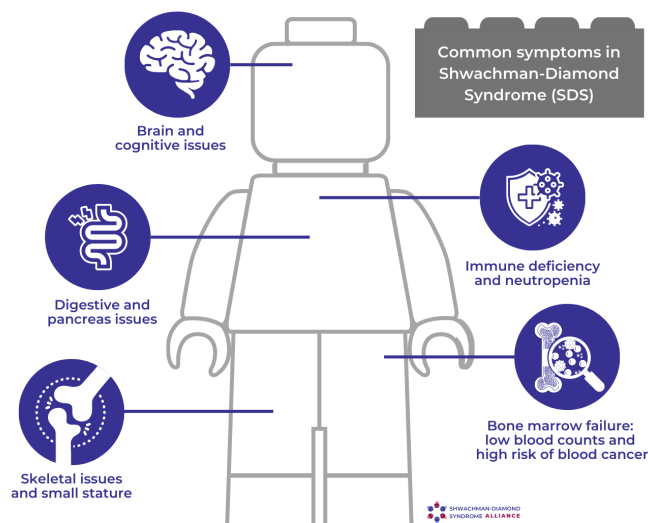
Dr. Kasiani Myers (Cincinnati Children's Hospital) provided a clinical overview of SDS, and Dr. Alyssa Kennedy (St. Jude Children's Research Hospital) shared an overview of current treatment options and future research directions.

Symptoms

SDS impacts many organ systems and significantly increases the risk of leukemia. A significant proportion of SDS patients develop acute myeloid leukemia (AML), myelodysplastic syndrome (MDS), or severe bone marrow failure, often necessitating a hematopoietic stem cell transplant (HSCT) – a high-risk procedure with significant potential for serious complications. The outcomes for AML in SDS patients are particularly poor.

Signs and symptoms of SDS include:

- **Bone marrow failure**/low blood counts and/or marrow cellularity
- **Clonal hematopoiesis**, often with TP53-mutant clones that cause a highly elevated risk of developing MDS/AML
- **Immune deficiency**, most commonly **severe chronic neutropenia**, putting patients at risk for frequent or serious



infections, including sepsis, often requiring hospitalization.

- **Digestive issues**, including pain and malnutrition due to **exocrine pancreatic insufficiency (EPI)**, typically apparent in over 90% of patients within the first 6 months of life. SDS is the second most common genetic cause of EPI.
- **Failure to thrive** and/or low birth weight
- **Elevated liver enzymes** with unknown cause and impact, typically peaking around 12 months of age. Some patients develop liver fibrosis.
- **Skeletal problems** due to bone deformation and growth issues, often affecting the knees and hips. Some patients have low bone density. Some infants are born with a narrow rib cage, which can cause life-threatening breathing problems. Short stature is common.
- **Oral health** issues and problems with teeth are also common.
- **Low tone** and mobility issues in early childhood, as well as feeding challenges, are common. Feeding tubes may be needed.
- **Cognitive issues** resulting in learning and behavioral challenges at home and school. People with SDS often have speech delay, executive function issues, ADHD, and may have depression/anxiety. Some patients are on the autism spectrum, while others can live independently. Some people with SDS have no cognitive issues. Structural changes in the brain (white matter, gray matter, and their connections) have been reported.

The severity of symptoms varies widely from patient to patient, causing frequently missed or delayed diagnoses, putting patients at risk for unnecessary suffering and life-threatening complications.

Prevalence, Cause, and Biology

The prevalence of SDS is estimated at 2,000-3,000 people in the US, based on an incidence of 1:76,000 to 1:170,000 in population studies and a shortened life expectancy (median in the mid-40s). Exact numbers are not known due to difficulties with diagnosis and tracking, and SDS patients are likely underdiagnosed due to a lack of awareness and access to (pre-symptomatic) testing and screening.

Most cases of SDS are caused by pathogenic variants in the essential gene **SBDS**, typically inherited from unsuspecting carrier parents in an autosomal recessive pattern, or caused de novo by gene conversion/recombination with an adjacent pseudogene, **SBDSP1**.

The essential SBDS protein plays a critical role in **ribosome biogenesis** by catalyzing the displacement of eIF6 from the large ribosomal subunit, thereby allowing the large and small ribosomal subunits to join and form a working ribosome. Ribosomes are large protein complexes that translate the genetic information encoded in mRNA into proteins, playing a key role in the

central dogma of molecular biology and being fundamental to life. In SDS, there are not enough working ribosomes to meet the cells' protein-synthesis demand.

Emerging evidence suggests that the highly increased risk of developing leukemia is due to “reduced fitness” of hematopoietic stem cells (HSCs), which creates selection pressure favoring cells that acquire maladaptive somatic mutations in genes such as TP53 (also known as the ‘guardian of the genome’). The short-term fitness advantage conferred by TP53 mutations increases the risk of malignant transformation.

Current treatment options

Currently, there is no targeted therapy for SDS. The only treatment for serious hematological complications – bone marrow failure, MDS, and AML – is hematopoietic stem cell transplant (HSCT). HSCT is rarely successful once AML develops, and carries significant risks even when performed before malignant transformation, precluding its use as a preventive measure. It remains a treatment of last resort.

Day-to-day quality of life has improved meaningfully due to advances in symptomatic treatment options, including pancreatic enzyme replacement therapy (PERT) to manage exocrine pancreatic insufficiency, granulocyte-colony stimulating factor (G-CSF) to improve neutropenia, and others.

Patients and families live with ongoing anxiety about life-threatening complications and the burden of invasive surveillance to monitor for bone marrow and clonal changes that may trigger HSCT.

While there is no targeted therapy for SDS yet, the patient community is eager to facilitate and accelerate therapy development – in particular for the priorities they have communicated in this meeting. In a recent publication¹, SDS Alliance has summarized many of the tools and infrastructure available to support this work.

¹ Hars ES, McReynolds LJ. **From Challenge to Opportunity: How Shwachman-Diamond Syndrome Became a Promising Target for Therapy Development.** Clin Pharmacol Ther. 2024 Dec;116(6):1377-1380. doi: 10.1002/cpt.3393. Epub 2024 Jul 22. PMID: 39039619; PMCID: PMC11567794.

Meeting Overview

This Voice of the Patient Report summarizes the input shared by patients, caregivers, clinicians, and researchers during the Externally-Led Patient-Focused Drug Development (EL-PFDD) meeting on Shwachman-Diamond Syndrome, held online and in-person in Cincinnati, OH, on June 4th, 2025. The meeting was convened to better understand the lived experience of SDS, the burden of disease, patients' perspectives on current and emerging treatments, and the community's priorities for future drug development.

The focus was on patients and their caregivers (primarily parents) to learn firsthand from the community about their lived experiences and priorities. Disease experts provided a brief overview of the clinical and research landscape, and an FDA representative provided an introduction to frame the meeting. Only patients and caregivers were invited to participate in the discussions and live poll.

The meeting was structured around two discussion topics:

Topic 1: The Burden and Daily Impact of SDS

Topic 2: Patient and Caregiver Perspectives on Current Treatments and Desired Future Therapies



Patients and caregivers provided their perspectives in various forms, including

- **Prepared statements** on two panels with five [patients or caregivers](#) each
- **Moderated discussion** and **live polling** with patients and caregivers in the audience, in person and online. See questions in the [Appendix](#).
- **Live call-in** options during the meeting for the remote audience; **phone** for voice and text messages, plus dedicated **email** addresses for comments before, during, and after the meeting.
- **Patient posters** displayed at the meeting, highlighting their experiences, perspectives, and humanity. See [Appendix](#).
- **Documentary film** to give a voice to patients and families who could not join, and to show the impact of SDS on people in their homes. It highlights the experience of four families. It was premiered at the meeting. See [Appendix](#).
- **Online surveys**, covering the same or similar questions as the live polling, were administered before and after the meeting to prepare the community and collect additional insights. The results have helped guide the meeting and this report. The full analysis will be published separately and available on our website at www.SDSAlliance.org/pfdd or upon request.

Objectives

The goal of this meeting and report was to document and share the profound impact SDS has — not only on the lives of the person diagnosed, but on entire families — who suffer because of a lack of safe, effective therapies. We are sharing the perspectives of SDS patients and caregivers with the FDA and other stakeholders on the impact of their condition and treatment preferences, as they know better than anyone the successes and unmet needs in the current diagnostic and treatment landscape. Learning what is most important to patients and their families provides critical information for clinicians, researchers, and drug developers about how to improve the lives of individuals and families living with SDS.

Our specific objectives were to share the impact SDS has on individuals' daily life with the FDA and other meeting attendees, including:

- Symptoms and daily impacts, including in early childhood development, adolescence, and adulthood
- Variable manifestation, highlighting the wide range of disease severity and symptoms among those living with SDS
- Current treatments and unmet needs: What constitutes a meaningful improvement for patients and families, and what kinds of risks are they willing to take to achieve it

We aim to offer FDA, industry, and other stakeholders deeper insights into disease manifestations, progression, and the weighing of risk and benefit by people with SDS and family caregivers. We aim

to provide industry with resources to improve clinical trial design, meaningful outcome measures, endpoints, and biomarkers to aid in the development of treatments for people with SDS.

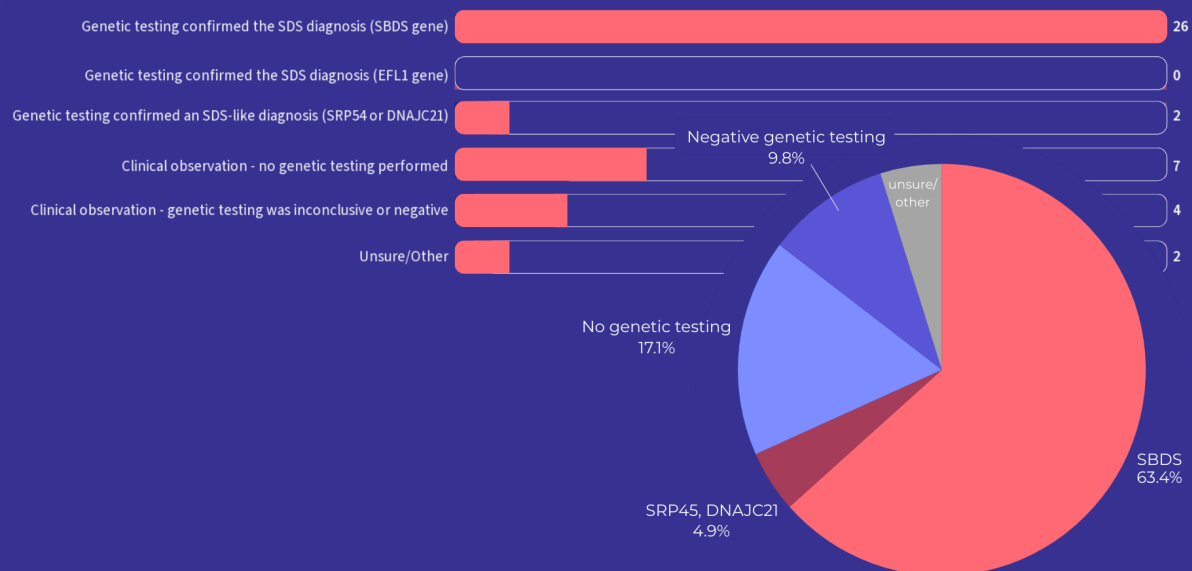
Meeting participants

The meeting participants were primarily SDS patients and their caregivers, with a few clinicians and researchers, as well as remote observers from the FDA.

Only patients and caregivers participated in the panels, discussions, polling, and surveys. Live polling showed about five times as many caregivers as patients in the audience, primarily from the US, with a concentration in the Eastern Time Zone. The large majority of the patients had a genetically confirmed diagnosis of SDS due to biallelic mutations in SBDS, and there were slightly more males than females (consistent with the literature). The complete set of polling questions is available in the [Polling section of the Appendix](#). Results are available upon request.

Polling question 5C:

How was the SDS diagnosis established?



Topic 1: Living with SDS - Symptoms and Daily Impacts



The morning session focused on the symptoms associated with Shwachman-Diamond Syndrome (SDS) and how they impact people with SDS and their families on a day-to-day basis. Across the prepared statements and open discussion, patients and caregivers described SDS as a multisystem, lifelong disorder that causes significant physical, emotional, developmental, and social challenges. Many emphasized that SDS affects “every organ” and that its impacts evolve and compound over time. Despite its severity, SDS often remains an “invisible disability,” leaving patients and families to navigate a condition that others can’t see or understand.

In the live polling, when asked **which SDS-related health concerns the patient has ever experienced**, the most common issues selected were:

1. Pancreatic Exocrine Insufficiency
2. Small Stature
3. Neutropenia

When asked **which of these health issues are the three most troublesome**, participants reported:

1. Neutropenia
2. Pancreatic Exocrine Insufficiency
3. Bone Marrow Failure

SDS related **health issues negatively impact many specific activities of daily living**. Poll participants report the following issues most frequently:

1. Attending school or having a job
2. Participating in sports and other recreational activities
3. Socializing

Notably, despite the above-mentioned impacts on daily living, participants were most **worried** about the future related to leukemia, life-threatening complications, and the development of new issues. They reported the following issues of SDS as worrying them the most, by a large margin:

1. Stress of not knowing if/when leukemia would develop
2. Reduced life expectancy/shortened life span
3. Symptoms (other than leukemia) may develop

Polling question:

What worries you most about SDS's impact in the future? Select TOP 3.



Neutropenia, Immune Dysfunction, and Chronic or Severe Infections

Patients and caregivers consistently described chronic, severe neutropenia and recurring infections as among the most disruptive and frightening aspects of living with SDS, day to day. Many described frequent, severe, and even life-threatening infections — including cellulitis, sepsis, and pneumonia — that required hospitalization or strong and prolonged antibiotic treatment. Even typical childhood illnesses could be dangerous. As one adult with SDS described being “constantly sick” as a child because “sicknesses last longer and are harder to recover for me.”

Joyce, a high school student living with SDS, described what a common illness means for her:



A normal case like the flu for any normal individual can lead to a possibly fatal and dangerous infection to me. While my friends would stay home from school, eating their favorite soup, and watching their favorite TV shows, I would need to go to the emergency room every time I had a fever, which, quite honestly, is terrifying.

The frequency of infections and related hospitalizations poses a huge burden on patients and caregivers. One adult with SDS described how he “was in and out of the hospital almost every three months [with infections and illnesses]. My mom could set a calendar by it.”

Another adult with SDS described how her “biggest health difficulty is getting recurrent pneumonias” because they lead to “losing time, which is so precious”.

Repeated and extended hospitalizations significantly disrupt both patients’ and caregivers’ lives, interfering with their ability to attend work and school and to socialize with their peers. Multiple parents described leaving work to manage their child’s SDS, leading to disruptions in their careers and significant loss of income. As Nicole, a mother who left an executive track job to care for her child with SDS, aptly summarized: “SDS doesn’t just steal health—it robs childhoods, careers, and stability.”

The significant risks of infections and the burden of hospitalizations lead caregivers to develop a pervasive sense of anxiety. Families described living in “constant fear of fevers, test results, new symptoms, of the unknown”.

Patients and families shared that the constant vigilance required to minimize exposure can lead to social isolation, as they avoid public places and large gatherings to protect their child from infections. As one mother described, “We’re incredibly careful going out in public. We avoid anybody that’s sick. No one comes to our house if they’re sick.” Another parent expanded on this, noting that when they isolated to prevent infections for their daughter, it impacted the whole families’ socialization: “That was really a tough time for not only her but [...] her other sibling, but it was also hard on us because we couldn’t go out and do normal life things with other people.”

A longtime member of the SDS community (who was originally diagnosed by Dr. Shwachman himself) described her “biggest health difficulty” as recurrent pneumonias — at one point every three months. The antibiotics required to treat them came at a cost: “Many of which were ototoxic, so I have hearing loss.” The infections nearly derailed her career: “I almost lost many jobs because of it...I was working 80-hour weeks in nursing school, and having to lose a week of nursing school puts you in danger of failing.” She described the broader impact as:



We always feel like we have a disadvantage to start with, and we’re always playing catch up.

She added: “And then there’s the ‘well, you don’t look sick’. That’s an everyday thing.”

Risk of Leukemia (MDS and AML)

Patients and families consistently identified the risk of leukemia as one of their greatest fears and a persistent source of anxiety. As Cresta, an adult living with SDS, shared, “I have fear every day that my TP53² clone mutation will develop into leukemia or myelodysplastic syndrome.”

Similarly, one parent of a child with SDS shared,



On top of the neutropenia, we know that he has the predisposition towards leukemia, and that's something that we live with in the back of our mind every single day. All we can do is monitor, we pray, but that's all that we can do for now.

The stress of not knowing if and when leukemia may develop, and concerns about the resulting reduced life expectancy, were further corroborated in the meeting's live poll as the participants' top worries about the future. Because of this high risk of leukemia and MDS, specialists recommend that patients with SDS undergo regular bone marrow biopsies and blood tests for surveillance. Cresta, who drives seven hours each way to see her specialist, described the financial burden and time involved, and highlighted the frequent and invasive surveillance as “painful.” She shared the impact frankly:



It causes me anxiety to wait weeks for the results and wonder if it will be positive or negative, and will I be facing death soon.

Digestive System Challenges and Exocrine Pancreatic Insufficiency

Patients consistently reported challenges with digestion, usually attributed to exocrine pancreatic insufficiency (EPI). Many described poor weight gain and feeding difficulties, particularly in young children. Several reported that despite taking pancreatic enzyme replacement therapy (PERT) such as CREON or Zenpep with every meal, they still struggled to gain weight or maintain adequate nutrition, and discomfort may still persist. As one adult with SDS described: “I have to take six to nine enzymes every day [...]. **Without the CREON, I could not digest my food or even survive SDS.**”

² SDS causes clonal hematopoiesis at a much younger age than in the general population. In SDS patients, biallelic somatic TP53 mutations are associated with a high risk of progression to AML. Patients are recommended to undergo regular surveillance to monitor for such mutations, and their presence is generally considered an indication for HSCT.

With young children, parents described significant anxiety about their child's nutrition and growth. One parent highlighted just how persistent these challenges can be, describing how even intensive, multi-pronged interventions did not lead to significant growth for her son:



He's been on the pancreatic enzyme since 8 months old, an appetite inducer since 12 months old, and we have not seen the jump in weight that everyone thought that we would see. As of now, he's in feeding therapy, but he still lacks the desire to eat, the ability to eat. And at two years old, he still predominantly gets his nutrition from toddler formula. [...] We weigh him, and he's below the first percentile every single time. There's not even a dot on the chart for him. And it's not for lack of trying.

Feeding challenges and interventions can significantly impact children's school participation and social activities. One parent shared how feeding tube management prevented her child from participating in normal childhood experiences like sleepovers — hard to manage away from home, and complex enough that even extended family has been reluctant to help.

Skeletal and Orthopedic Issues

Families consistently described skeletal issues — such as hip dysplasia, metaphyseal changes, scoliosis, and knee issues — as a major factor limiting daily function and quality of life.

Rebecca, mother of six-year-old Declan, described first noticing something wrong when she looked at his legs: “When he was two years old, I looked at his legs, and they just looked off — you could see the skin and the bones just thinner or something.” He had a distinctive waddle, and “he would want to sit a lot. He would want me to carry him. And he was older at a point where you would be walking and running.” After bilateral valgus femoral osteotomies, the change was striking: “He loves to bike, swim. Play on the playground. Very active.” But Rebecca's worry didn't end there:



"Nobody can guarantee me: is this going to come back? Are his hips going to become deformed over time, and are we going to go backwards?"

Orthopedic complications often require multiple surgeries over years. A young adult patient described a long sequence of interventions:



I've had 25 skeletal operations, including those hip surgeries, knee surgeries, and even the spinal fusion. [...] Hip pinning on both sides, back bracing, magnetic rod lengthening, spinal fusion, hip reconstruction, and then surgery on my knees to correct alignment. [...] am slowly developing arthritis of my hips. I do have a slight gait and that waddle.

Other families spoke of how orthopedic issues have a profound impact on quality of life and limit participation in typical childhood activities: “My son had such high pain tolerance that we didn’t realize how much he was struggling. He couldn’t run, couldn’t do the school mileage club, couldn’t keep up with his classmates. It broke my heart.”

Fatigue and Stamina

Patients and caregivers consistently reported fatigue and poor endurance as significant challenges, describing difficulty keeping up with peers and needing frequent rest. One parent shared: “She fatigues quickly. [Although] she looks like a completely normal child and it’s an invisible disease to try and explain to people.” One young adult with SDS reflected,



So, I distinctly remember when I was really young in PE [physical education] classes: whenever we would start PE, we would do warm-up laps, and the instructor would say, like, ‘hey, do five, six laps around the gym’. Everyone would be done with their laps, and I would be struggling to even do two or three. So, there’d be most days when, like, I want to try to do all five, but the instructor was like, ‘I’m sorry, we need to move on. We’ve got other exercises we need to do’.

The emotional impact of exclusion from group physical activities is evident in one mother’s account of her son with SDS: “He could do none of those things. He could barely walk... He was not able to participate [in sports], which broke his heart, and mine, and his older sister’s, who gave him her water bottle because he was so devastated that he couldn’t participate.”

Participants talked about invisible symptoms, such as fatigue, heat intolerance, and reduced physical endurance, which often require accommodations. Jess, whose eight-year-old daughter underwent transplant as a young child, described how temperature dysregulation shapes every outing: “She doesn’t sweat, so she doesn’t control her body temperature, which has quite an impact on her daily life and ours.” Activities her daughter loves — like riding horses — became impossible in warm weather: “One of the activities that we can do is swimming. Anything that involves water gives us the ability to be outside.” For longer outings: “We do use a large stroller...she’s gotten bigger, and it’s hard for me to carry her. If we don’t bring it, at some point she’s wanting to be carried.” Although her daughter “looks like a completely normal child,” Jess noted:



It’s an invisible disease to try and explain to people.

Cognitive and Neurodevelopmental Impacts

Families underscored the impact of neurodevelopmental and psychosocial challenges caused by SDS, noting that schools and healthcare often overlooked or dismissed these issues. A mother in the audience, whose son was diagnosed with both SDS and autism two weeks apart, described the neurodevelopmental dimension that often goes unaddressed:



He is incredibly smart, so it's more of the social side of things — the sensory overload. There's a lot of structure in their day, so sometimes he just needs a break to take a walk for 10 minutes and then come back to it. 90% of the time, he's cognitively like any other kid in the class; there's that 10% of the time where he just needs a minute.

Another mother described the struggle to get her 11-year-old son's neurodevelopmental needs — such as speech delay and sensory issues — supported by the school system: “Even though there is documented history in the literature of the difficulties in SDS with these things, it's not [...] a diagnostic tool. He doesn't fit inside the boxes that do exist.”

Quality of Life and Mental Health

Across discussions, patients and families emphasized the ongoing impact of SDS on school, work, and daily routines. Missed school days, medical appointments, fatigue, mobility limitations, and hospitalizations shape everyday family life. One adult with SDS, who had been getting pneumonia every three months for years, summarized the cumulative impact: “Just losing time — losing time, which is so precious.”

Many expressed frustration that the seriousness of the condition is often dismissed: “There's the, ‘well, you don't look sick’. That's an everyday thing.” This invisibility leads to misunderstanding, stigma, and the need for constant advocacy in educational and workplace settings.

Nicole, mother of Roman (age 10), shared how this mental and social burden affected their whole family:



Everything we do revolves around [our son with SDS], whether it means we travel only where there are knowledgeable children's hospitals, or are extremely selective on extracurricular activities that can keep him. SDS doesn't give you an exemption from other health issues or life events—it just makes those issues so much harder to manage. SDS is a house of cards that will fall not just for the patient, but those around them physically, financially, and emotionally.”

She also shared the unpredictability of SDS progression and the fear of new or worsening symptoms – a recurring theme throughout the meeting. The persistent worry about future health, independence, and life expectancy weighs heavily on families. “I’m not sure if I should be hopeful, be cautious. It feels like we are driving blind. Will he make it to college? Will he develop more SDS-related health issues? Will he be able to maintain a job, or will his health needs take a toll?”

She described how SDS shapes her son’s sense of himself and his future: “He is aware that the life he is living is radically different than his peers in too many ways.” She added, “I don’t think about Roman’s future the same as I do my healthy children. There is an asterisk there.”

Joyce, an adolescent with SDS, described the hidden weight of an invisible disease: “When you first meet me, you would never know the hidden struggles that I face due to my disease being invisible. The truth is, a disease is not something that could be put aside. It is something that is constantly present.” She added:



A kid like me shouldn’t have to determine their dream university based on whether there is a medical professional within 100 miles who is even aware of their condition.

One parent in the afternoon session raised another important topic – the importance of giving patients, including young children, some agency and control over their own care: “Kids pick up that they have no control in their life. Letting them make their own decisions helps. I don’t care if it’s where you stick that needle – ask them where they want their Neupogen shot. Give them some control.”

A longtime community member in the audience, whose daughter lived with SDS for 52 years before passing from liver and kidney failure, offered a stark warning about how SDS can cause progressive organ dysfunction later in life:



SDS affects every organ in the human body – every one. So while your kids are young, you are focused on the digestive issues and certainly on the neutropenia and the infections, but as the human body ages, other organs are going to become involved.

She described how her daughter’s pancreatic disease eventually spread to her liver, progressing rapidly to cirrhosis and kidney failure: “We were just taken completely by surprise.”

What patients and families made clear is that SDS is not a childhood disease. It is lifelong and progressive. Symptoms may change over time, but SDS doesn’t go away – and neither should the support, monitoring, and therapies available. Patients need transition-of-care support, lifelong surveillance/monitoring, and therapies that address the full spectrum of SDS.

Topic 2: Patient Perspectives - Current & Emerging Treatments



Overview and Live Polling

Patients and caregivers described treatments for SDS they have tried, with insights into what worked, what didn't, and the side effects and complications they experienced. Their stories spanned life without transplant and life post-transplant. Despite different experiences, several themes were consistent – SDS treatments can be lifesaving, but they are often traumatic, burdensome, and show mixed results in addressing symptoms. They all aim to manage symptoms as they arise, not to prevent them, and none addresses the underlying cause of SDS.

These themes also came through in the live polling of meeting participants. Patients and caregivers reported that current treatment regimens are only somewhat helpful in controlling SDS symptoms – and that they don't address their ever-present fear of leukemia and anxiety about the future at all, which were among the most prominent themes of the meeting.

When asked about what **medications and treatments the patient has used** to treat SDS symptoms, participants reported the following five the most:

1. Pancreatic Enzyme Replacement Therapy
2. Fat soluble vitamin replacement (A,D,E)
3. Laxatives/stool softeners
4. Granulocyte stimulating factors (GCSF)
5. Medication for anxiety, depression, ADHD

To further support their health, patients also used other **(non-drug) therapies:**

1. Diet modifications
2. PT/OT
3. Psychological counseling
4. Isolation/masking

When asked about the **biggest drawbacks of current treatments**, participants emphasized several themes.

1. Only treats some, but not all symptoms
2. High cost/insurance doesn't cover
3. Route of administration
4. Risks and side effects

Perspectives on Current Treatments

Hematopoietic Stem Cell Transplant (HSCT)

For some families, HSCT³ brought stability they had never before experienced. Families shared the heavy burden of emergency room visits with any fever. When patients with SDS are neutropenic and develop a fever, they need to seek emergency care to assess for potentially life-threatening infections and initiate aggressive treatment if needed. Brittanya, whose five-year-old son Jayden is about two years post-HSCT, described the elimination of emergency healthcare visits for fever protocols after her child's transplant, and how this helped lift some of the heavy emotional and logistical burden of caring for a child with SDS:



Anytime a child...gets a fever, you have to rush to the ER.

After transplant?

We have not had to go to the ER one time due to an illness, so that's huge—huge.

Will, who underwent HSCT at age 7 — and is a beneficiary of one of the earliest successful transplants in an SDS patient — is now an adult, nineteen years post-procedure. He shared: “Blessfully, my transplant was a success, and I’ve had no complications for 19 years.”

Patients and caregivers described tremendous relief after successfully undergoing HSCT, even though it does not address non-hematologic symptoms. Chilton's mother described her son's reaction shortly after transplant — and was careful to clarify what he meant:



He said, ‘I don’t have SDS anymore.’ Yes, he has the skeletal issues, and he knows he has the pancreatic issue — but what was the number one scare everybody had? Leukemia. After transplant, you’re not worried about the number one concern. And that’s what he meant. The pancreas, we take the pills. The skeletal, those are things we think we can handle. But when he says ‘I don’t have SDS,’ he means in his bone marrow. And to him, that’s number one — which was number one for most of you. So hallelujah.

On the other hand, despite these life-altering successes, participants described a range of serious risks and lasting consequences of HSCT that extended well beyond the transplant itself. Even with modern low-intensity conditioning regimens, patients with SDS are at heightened risk for severe toxicity. In the weeks and months after transplant, patients are extremely vulnerable to infections and require strict isolation and monitoring. There is a real possibility of death, serious long-term side effects from toxicity, immune suppression, or Graft-versus-Host Disease (GvHD), and

³ HSCT, also known as bone marrow transplant, is a high-stakes, but often transformative, procedure. When successful, HSCT resolves patients' bone marrow failure-related symptoms, such as low blood counts (e.g., anemia or pancytopenia), and immune deficiency (neutropenia and risk of serious infections and sepsis), and the risk of MDS and AML. It comes with serious risks and side effects, and does not improve symptoms outside the bone marrow.

transplant can fail when donor cells don't engraft successfully. Families also described the weight of decisions that had to be made before transplant even began.

One mother shared what it meant to be asked about fertility preservation for her three-year-old son: "I would love to see a treatment where you don't have to have those conversations about fertility preservation and what you think your then three-year-old may or may not want. I'm sure a lot of families have had those similar conversations." For many, these compounding issues — physical, emotional, and logistical — began long before the transplant itself and continued long after.

Will, whose transplant was ultimately successful, also described how the precautions to avoid infections required for weeks to months following HSCT layered on top of a longer-term social isolation experienced as an SDS patient: "For much of my life, I felt like most of us did during COVID—cut off from the world." During transplant, he recalled: "I reached my hand out to my brother through the [glass window in the hospital] door. The door divided us." Later surgeries and recoveries left him homebound for months: "I felt isolated. I felt cut off from the rest of the world."

Some HSCT recipients experience life-threatening and long-lasting complications. Brittanya described her son's devastating post-transplant course as he developed lung and other complications. She was told by the medical team, "Your son has a 5–10% chance of survival. This is a Hail Mary." Over the course of his treatment, he ultimately required 12 days on extracorporeal membrane oxygenation (ECMO), 20+ days intubated, multiple months on dialysis, and 222 days away from home. Brittanya recalled: "By all accounts, he was an infant in a toddler's body. He had to learn to walk, talk, eat, and potty train all over again."

Even when transplant succeeds, its side effects can be permanent. Brittanya further shared:



My now five-year-old has permanent lung and kidney damage that can't be reversed...He cries when he can't keep up with the other kids.

Chilton, a 22-year-old who had recently undergone transplant, further highlighted the potential cascade of medical complications — cytomegalovirus (CMV), BK virus, kidney damage, severe fatigue—as well as the emotional toll of isolation during treatment:



I told my parents one night, 'I can't do this anymore.'

Antibiotics

Patients and families talked a lot about frequent or severe infections, often requiring hospitalizations. While prophylactic antibiotic use is not common practice in the US, many patients need prolonged or strong antibiotic treatment to help fight infections when they occur or to respond aggressively to early signs of sepsis. Many young patients have a fever protocol in place,

meaning they have to go to their nearest ER for any fever to undergo a workup and to rule out or treat sepsis on an emergent basis. Some emphasized that, though imperfect, supportive care has kept them alive today. One adult patient said:



**People are living longer.
Without antibiotics, I would have been long gone.**

Granulocyte-Colony Stimulating Factor (G-CSF)

Families widely recognized G-CSF (such as Filgrastim and Neupogen) as one of the most effective existing therapies, particularly for preventing severe bacterial infections. Avoiding hospitalizations for infection reduced disruptions to patients' day-to-day lives, as one parent described: "Before Neupogen, our son constantly had life-threatening infections. After Neupogen, he can usually clear an infection and get back to being somewhat normal."

Maria, mother of eleven-year-old Felicity, noted that after starting daily filgrastim,



Her neutrophil count is up, she's no longer getting severe infections...we're no longer terrified of school, public spaces, swimming, or her favorite foods.

However, these injections also carried significant challenges, including, for some, intense anxiety, needle phobia, and medical trauma. Maria described the trauma her daughter faces with daily G-CSF injections: "It is not a daily ritual—it is a daily ordeal. [...] On a bad day, everything falls apart, and she dissolves into angry tears, screaming, biting, hitting, and kicking while we hold her down."

Pancreatic Enzyme Replacement Therapy (PERT)

Patients and families shared in polling and discussions that PERT (such as CREON and Zenpep) is one of the most commonly used treatments among patients living with SDS. Most children experience pancreatic exocrine insufficiency at least intermittently and benefit from PERT, though about half outgrow the need by age 6. In young adults and adults, the need for PERT often returns. As one mother pointed out in her introduction of the meeting, CREON saved her child's life, allowing her to thrive, sleep through the night, and smile for the first time at age 7 months. CREON, even if a weight gain is not immediately apparent, also dramatically improves the patient's quality of life.

However, PERT did not work for all patients for weight gain, with one parent noting:



**We expected weight gain with Creon.
We still haven't seen any difference.**

As parents often share, PERT is hard to administer to infants who can't take solids yet and toddlers who are picky eaters or have oral aversions. It can also be hard to access, as some providers don't understand its critical importance, or insurance may not cover the required amount. For some, there are also challenges with social pressure (such as teens not wanting to take the capsules in public) or schools not facilitating access to PERT during school meals.

Other Gastrointestinal and Feeding Support

For many patients, feeding support beyond PERT is essential from the earliest months of life. Several families described feeding tubes (such as G-tubes) as necessary interventions that, while scary and burdensome, provided critical nutritional support when oral feeding alone was insufficient.

Simple over-the-counter interventions could also make a profound, even life-saving difference. In her opening remarks, meeting host Eszter Hars described MiraLAX (polyethylene glycol 3350) as one of three interventions that saved her daughter's life in her first three years — alongside a blood transfusion and pancreatic enzyme replacement therapy.

Kavan, a 22-year-old who has lived with a G-tube since third grade, described its ongoing realities: "I mean, obviously, change the G-tube once every three months, but there's still a hole in my stomach. Living with a G-tube can have its own set of challenges. It has to be replaced every once in a while, and it's prone to infections. Just last week, I was hospitalized with an infected abscess around my tube." The social impact was also significant — getting his G-tube coincided with becoming more withdrawn: "I stopped hanging out with some school friends and became more of an introvert."

Patients also sometimes experienced serious, unexpected side effects from certain medications. One mother recalled trying an appetite stimulant: "We started Periactin when he was about four years old, and shortly after that, we also had to start risperidone because he was so behaviorally affected by it. There was no correlation or any papers anywhere that said that was a reaction, because it should not have been."



The only way we found out was from being in a room like this, and another parent said, 'Hey, wait. That happened to us, too.'

Burden of Daily Treatment Demands

The variety of treatments and supportive care required to manage SDS symptoms took a cumulative toll on patients and families. One recurring theme was the relentlessness of daily demands. As Kavan described, managing daily medications required a MedMinder — a smart pill dispenser that locks and alerts caregivers if a dose is missed: "Every Tuesday I refill my MedMinder... I have alarms set for when to take pills, start my tube feeds, and testing to prevent bladder infections."

Several families described the deep social and psychological toll of living around treatment needs. Leah captured what this meant for her son's childhood: "He had an NG tube from about five months old, and now he's got a gastrostomy... It just makes it really hard. He's at the age now where he'd love to have sleepovers and that sort of thing, but hard to turn up with your child and be like, oh, here's all this. Like, people just don't really do that. Even my family have been scared of using it, so it's been us doing it basically the whole time."

Several participants echoed this. Joyce, a high school student living with SDS, described the complexity of growing up managing pills: "Before every meal and snack, I must take multiple pills. And when I was younger, I was terrified to swallow pills, and my family had to come up with creative ways to try to feed me the pills." She also described developing a needle phobia from years of blood draws:



My veins are shot due to the thousands of blood tests I have endured throughout my life. The complexity of my anatomy, combined with my developing phobia of needles due to those thousands of blood tests, led to many panic attacks.

Caregivers also described how their children missed out on normal developmental experiences. Maria introduced a paper plate award — misspelled by a swim coach to read "most *relisient*" — given to her daughter Felicity, not for athletic achievement, but because she attended the final swim meet after spending most of the summer hospitalized:



**This symbolizes everything my daughter is—
and everything I wish she did not have to be.
Give my daughter the chance to be resilient in the normal ways.**

Priorities for Future Treatments

Despite the diversity of their experiences — from infants to adults, transplanted or not — patients and caregivers showed strong alignment on what they wish for from future therapies. Polling confirmed that the top priorities, by a wide margin, were reducing leukemia risk and avoiding or improving the transplant experience. The discussions that followed shed light on what it would actually mean, in everyday life, for a treatment to succeed.

Reduce the risk of leukemia

The fear of leukemia was the single most dominant theme of the afternoon session — and it was the top-ranked treatment goal in live polling, with avoiding the need for transplant and improving

transplant outcomes close behind. For many families, managing daily symptoms is possible; it is the fear of leukemia that keeps them up at night.

One audience member shared why she selected reducing leukemia risk, avoiding transplant, and improving transplant outcomes as her top three priorities in the poll:



Those are the scariest parts of Shwachman...everything else, I feel, can be managed with the right care. And those three are just the scariest aspects to me, and they're the biggest and the toughest to overcome.

Another parent, who has not yet had to face a transplant, shared: “Yes, what we’re doing is helping. But selfishly, I want that fear removed—of it developing into something worse. Why is transplant our only hope?”

As one adult with SDS put it:



[A cure would mean] not having bone marrow biopsies anymore and not living every day with the fear of my SDS turning into leukemia or MDS.

Provide alternatives to transplant, or make it safer

For families whose children have already undergone HSCT, the ask was direct: no other family should have to go through what they did. Brittanya, whose son Jayden nearly died of post-transplant complications requiring ECMO, dialysis, and months of rehabilitation, articulated a vision for what better would look like:



The effects of transplant are forever. My dream is to find a therapy that can stop the mutations that cause leukemia, so other children like Jayden don't have to undergo transplant. What if we could find a therapy that has the same effect as transplant but without all the complications and side effects? What if we could find a therapy where annual bone marrow biopsies aren't necessary to determine if mutations are occurring?

Chilton, a 22-year-old who underwent transplant the previous year and spent months away from home managing complications including CMV, BK virus, and kidney damage, shared the toll it took on him emotionally and physically. He donated his stem cells to a gene therapy research program before his transplant, and was clear about his motivation: “I don’t want other SDS kids to go through a bone marrow transplant, like I did.”

For families who have not yet reached a transplant decision, the calculation is different but no less urgent. One parent whose son currently receives G-CSF and Creon noted that gaps remain: “Yes, what we’re doing is helping, but it doesn’t treat every symptom.” Another shared what it meant to have a child with a TP53 clone: “All we can do is monitor, we pray, but that’s all we can do for now.”

Participants made clear they would want future treatments to reduce not only the need for transplant but also its risks when it is necessary — shorter and less toxic conditioning regimens, and better management of post-transplant complications. Most of all, they want treatments that work before leukemia develops, and prevent it altogether.

Chilton's father, reflecting on his son's transplant experience, emphasized the urgency: "What you want to make sure is that your kid is alive to take advantage of the medical improvements—and we're trying to speed that up."

Develop treatments that children can tolerate

Maria, whose daughter Felicity (age 11) had experienced a life-changing improvement in infection control after starting filgrastim, raised a critical issue with treatment delivery:



Please remember that while it is, of course, critical that a treatment be effective, please also consider the impact of how the treatment is administered. These kids have experienced so much, and anything that makes receiving the treatment easier may be worth some of the effectiveness.

Felicity has severe medical anxiety and needle phobia, and her daily G-CSF injection has become a daily ordeal. Maria described the spectrum: on a good day, an hour of preparation, deep breaths, and careful de-escalation. On a bad day, "my sweet, happy child dissolves into angry tears, screaming, biting, hitting, and kicking while we hold her down to give her her shot." When asked whether an oral version of the drug—at half the efficacy—would be worthwhile, Maria's answer was unequivocal:



Half the benefit without the fear and trauma we face every day? Sign us up.

Patients and families repeatedly raised the notion that considerations for tolerability, administration route, and feasibility need to be front and center when developing therapies. Kavan, a 22-year-old living with SDS, captured the cumulative burden from the patient's perspective: "I would really appreciate less needles. I wish I didn't have to take so many pills and injections." He added simply: "At this point, anything would help."

Families also emphasized the importance of developing treatments specifically for children. As Maria noted, drug development often starts with adults, "but when it comes to SDS, many of the patients are children, and they need these treatments sooner rather than later."

Address growth, stature, and bone complications

Improved physical growth and reduced bone-related limitations emerged as deeply meaningful in the patient and family discussions — not just for practical and physical reasons, but also because of their profound impact on patients' mental health, social development, and sense of belonging.

Leah, speaking on behalf of her eight-year-old son, described what this priority means in practice:



He's the smallest kid in his class. It makes it a lot harder for him to participate in sports. I feel like he's already at a disadvantage before he even starts. And I see his heart sink when we meet new people, and he's eight, and they're like, 'Oh, is he four?' He has a four-year-old sister who is almost heavier than him—and for him, that's a really big deal.

Leah noted that even a modest improvement in height and stature would make a meaningful difference. She also flagged the importance of avoiding treatments with serious risks and side effects: her son had tried growth hormone but discontinued it due to concerns about bone marrow complications. This is consistent with other families' priority of not worsening leukemia risk.

Reduce the burden of treatments and surveillance

Patients and families consistently described ongoing surveillance and monitoring as physically exhausting and mentally taxing. Annual bone marrow biopsies, frequent blood draws, and regular injections take a toll that compounds over years.

Cresta, an adult with SDS, emphasized the logistical and emotional toll of ongoing surveillance to assess leukemia risk: a 7-hour drive each way for an annual bone marrow biopsy at a specialist center. She described waiting weeks for results, “wondering if it will be positive or negative, and will I be facing death soon.”

Joyce captured another aspect of biopsy experience: “While others were planning their summer vacations, I was planning my annual bone marrow biopsy — a painful and invasive procedure that leaves me sore for days. I remember one night having to cry myself to sleep because every inch I would turn would send a jolt of pain through my body.”

Kavan shared feeling like “a pincushion” after a hospitalization that involved multiple blood draws per day.

Patients and families understand why surveillance is necessary. But they expressed a need to reduce its physical and emotional burden. Their ultimate vision is treatments that would make surveillance obsolete by reducing the leukemia risk sufficiently.

Enable participation in school, work, and everyday life

A common thread across all the patient priorities was the goal of enabling children and adults with SDS to live full lives, fully integrated into their communities. What they are seeking is better immune function, fewer infections, better growth, and fewer hospitalizations.

A mother – speaking on behalf of her 11-year-old son, who has severe stomach pain that makes eating at school difficult – described what his treatment goal actually was: “He just wants to eat lunch at school.” She explained: “He’s afraid to eat lunch at school because when he does eat, it hurts. And then imagine a table of kids asking, ‘Why did you only take one bite?’ He doesn’t want to explain it.” His stated goal – eating lunch with his friends – comes easily to all his classmates.

As described above, Maria has one ask for her daughter with SDS:



Let's give her a chance to be resilient for working hard at the things that she loves.

Another parent shared her son's words – a ten-year-old navigating school with executive functioning challenges and learning disabilities: “I’m tired of getting in trouble. I’m tired of trying so hard to get through school.” His frustration is not with school. It’s having to work twice as hard to get through what comes easily to others.

Kavan’s mother described what it meant for him to compete at the Special Olympics New Hampshire state summer games – even after a week in the hospital, even knowing his times would be slower: “He’s not really thinking he’s going to get the gold this time. He was saying, ‘I got a gold medal in this event last year. Probably not going to happen this year because I haven’t been able to train – but still working.’” That determination – to keep showing up, keep competing, keep working toward his goals – is what patients and families are asking researchers and drug developers to help make possible.

Perspectives on Unmet Needs

Across both the discussion and the prepared statements, the message was clear: current treatments keep many patients alive – but at tremendous physical and emotional costs. Families are grateful for existing options, yet desperate for therapies that address the root cause of the disease rather than merely managing some of the many symptoms.

A longtime active member of the community, whose family has lived with SDS for decades, captured both the exhaustion and the determination that defined the day:



**I hate transplants. I hate Neupogen. I hate it all.
But there is progress. Keep pushing.**

Risk-Benefit Considerations

Patients and caregivers shared how the seriousness of SDS shapes their view of treatment risks, supported by examples of decisions they had already made and the trade-offs they had faced.

Chilton's father described the family's decision to decline transplant when Chilton was three years old as a calculated bet on the future:



We felt like we were able to buy 18 years' worth of time for improvements to where they're at today.

By waiting, they benefited from safer protocols that were developed in the meantime. That decision — declining a high-risk intervention in the early years to benefit from scientific and medical advances later — demonstrates one aspect of how SDS families assess risk-benefit calculations at different timepoints and stages of disease progression. In Chilton's family's recount, those years deferring transplant were marked by the worries that come with living with SDS: the constant vigilance around infections and ongoing monitoring for leukemia risk, which ultimately became urgent when a TP53 mutation was detected.

Another parent described a similar calculation around transplant safety: “When my son was first diagnosed, my hematologist said, ‘I bet you’re wondering why we just don’t transplant everyone from the get-go.’ And then he explained the risks, which I did not know about. So, now, making it safer, more effective, shorter — that would be a game changer.”

Maria's account of Felicity's daily G-CSF ordeal approached the question from another angle: treatment burden. Given the choice between an injectable or an oral alternative (that's half as effective), she didn't hesitate. The benefit of reduced trauma outweighed the trade-off in efficacy.

Leah articulated another aspect of the decision-making process — risks families are not willing to take. When asked about growth hormone for her son, she noted they had tried it but discontinued due to concerns about possible bone marrow complications: “It would have to be something that didn't have those big risks associated with it.” Even if a treatment offers a meaningful benefit, it could be declined if the risks in other areas (especially leukemia risk) were too high.

Drug Formulation Considerations

Patients and families highlighted the impact of formulation and route of administration on whether a therapy can realistically be used. Maria shared her family's experience with Felicity's daily G-CSF ordeal, and how they would happily trade reduced efficacy if an oral formulation could replace the current injectable.

Kavan's mother added:



If there was a non-injection form of G-CSF, that would be absolutely amazing.

Meeting participants reinforced these insights through live polling: the route of administration was among the top three drawbacks of current treatments reported by participants, alongside efficacy gaps and cost. While this topic was covered less in the discussions during the meeting, it represents a significant concern in the daily lives of SDS patients and families. Patients have unique experiences with medications and procedures that shape what works for them⁴.

⁴ While pill swallowing is often assumed to be challenging in pediatric populations, this is not the case for many in the SDS community. Many children with SDS learn to swallow multiple large capsules at a time from an early age — because they have to take PERT with every meal, every day. Therefore, oral pill or capsule formulations may be feasible or even preferred.

On the other hand, some children with SDS have oral aversions, sensory issues, or developmental challenges that make pill swallowing or oral feeding difficult. Some have a feeding tube to supplement or replace oral feeding. PERT and other oral medications are difficult to administer to infants who cannot take solid food yet. For these patients, liquid or tube-compatible formulations are necessary.

Summary and Future Direction

The voices shared at this meeting tell a clear and consistent story about living with Shwachman-Diamond Syndrome. SDS takes a profound toll — not only on the person diagnosed, but on their entire family. As Nicole, mother of ten-year-old Roman, put it: “SDS doesn’t just steal health — it robs childhoods, careers, and stability.” Joyce, a high school student living with SDS, captured what that looks like from the inside:



When you first meet me, you would never know the hidden struggles that I face due to my disease being invisible. The truth is, this disease is not something that can be put aside. It is something that is constantly present.

The fear of leukemia runs through nearly every story and experience shared. It shapes where families travel, what kind of school a child can attend, and how every treatment decision is made. The risk of leukemia is what keeps patients and caregivers up at night — and it is what they most urgently need the research and regulatory community to help address.

SDS is not just a childhood disease. Patients and families shared their experiences with symptoms evolving over time and new ones emerging, including organ complications later in life, and the growing risk of leukemia. Patients face lifelong surveillance. And they need treatments that address the full spectrum of the disease from infancy through adulthood.

Current treatments — bone marrow transplant, G-CSF, pancreatic enzyme replacement, and supportive care — have extended and improved lives, and patients and families are genuinely grateful for them. But they treat some symptoms and not others, none addresses the underlying cause of SDS, and for many families, they carry a high financial, physical, and emotional cost. The community is asking, clearly and urgently, for treatments that address the underlying cause, so that complications such as progression to MDS/AML can be prevented.

Meeting participants consistently identified the same priorities for future therapies: reducing the risk of leukemia; providing alternatives to transplant or making it safer; developing treatments that children can actually tolerate; improving growth, digestive health, and skeletal function; reducing the burden of treatment and surveillance; and enabling participation in school, work, and everyday life. When patients and families say they want treatments that are safe, they mean above all else treatments that don’t increase the risk of leukemia.

This report documents what patients and families have shared — and what they are asking researchers and regulators to understand and act on. As one longtime community member put it:



I hate transplants. I hate Neupogen. I hate it all. But there is progress. Keep pushing.

Appendix

Agenda.....	36
Speakers.....	39
PFDD Discussion Questions.....	42
Meeting recording and photography.....	43
Live Polling.....	44
SDS Documentary Film “Until There’s A Cure”	46
Patient Posters.....	47

Agenda

Externally-Led Patient Focused
Drug Development Meeting for
Shwachman-Diamond Syndrome

EL-PFDD MEETING FOR SDS

June 4th, 2025
Hybrid (Zoom/Cincinnati, OH)
www.SDSAlliance.org/pfdd



EL-PFDD for Shwachman-Diamond Syndrome (SDS)

8:00 AM In-person attendees: Breakfast and Registration

10:00 AM Welcome and Introduction by the Meeting Host

Eszter Hars, Ph.D.
Shwachman-Diamond Syndrome Alliance
Parent of a child with SDS

10:10 AM Opening Remarks by an FDA Representative

Dr. Martha Donoghue
FDA, Pediatric and Rare Cancers Oncology Center of Excellence

10:20 AM Clinical overview of Shwachman-Diamond Syndrome

Dr. Kasiani Myers
Cincinnati Children's Hospital, North American SDS Registry

10:30 AM Meeting Overview and Logistics by the Meeting Host

10:35 AM Audience and Demographic Polling

James Valentine
Hyman Phelps McNamara

10:40 AM Topic 1: Health Effects and Daily Impacts of SDS

■ **10:40-11:10 Panel of 5 Patients/Caregivers Sharing their Stories**

Nicole - mother of Roman (10), who lives SDS

Cresta - adult living with SDS
Joyce - high school student living with SDS
Joe - adult living with SDS
Kaitlyn - mother to Ezekiel (2), who lives with SDS

- **11:10-12:10 Audience Polling & Moderated Audience Discussion**
Audience in the room and remotely via interactive webinar

12:10 PM Lunch Break

- **1:00 PM: Documentary Film Premiere: Until There's a Cure.**
The documentary includes discussions of serious illness and the loss of loved ones. Viewer discretion is advised.
Declan and his family
Elijah's family
Addie and her family
Whitner and her family

1:30 PM Welcome to the Afternoon Session by the Meeting Host

1:35 PM Introduction to the Treatment Landscape by Clinical SDS Expert
Dr. Alyssa Kennedy
St. Jude Children's Research Hospital

1:45 PM Topic 2: Current Approaches to Treatment of SDS

- **1:45-2:15 Panel of 5 Patients/Caregivers Sharing their Stories**

Brittanya - mother of Jayden (5), who lives with SDS
Kavan - adult living with SDS
Will - adult living with SDS
Chilton - adult living SDS
Maria - mother to Felicity (11), who lives with SDS

- **2:15-3:15 Audience Polling & Moderated Audience Discussion**

3:15 PM Summary Remarks by a Healthcare Research Expert
Dr. Vanessa Merker
Massachusetts General Hospital and Harvard Medical School

3:25 PM Closing Remarks and Next Steps by the Meeting Host

3:30 PM Adjourn - Thank you all! Refreshments for in-person attendees.

Bonus for Patients and Caregivers only

3:30–5:00 **Debrief and Support Group for Patients and Caregivers.** Facilitated by a qualified professional. Advanced registration is required. Patients and caregivers will be notified in advance.

Project PACER Kickoff Meeting and Dinner

www.SDSAlliance.org/pacer

6:00 PM **Working Dinner Starts**

6:30 PM **Project Overview by the Project Lead**
Eszter Hars, Ph.D.
Shwachman-Diamond Syndrome Alliance
Parent of a child with SDS

6:45 PM **What is Patient-Centered Comparative Effectiveness Research?
And How Can All Stakeholders Play a Vital Role?**
Dr. Vanessa Merker
Massachusetts General Hospital and Harvard Medical School

7:15 PM **Patient Poster Session**
Exploring the impact of patient stories and the power of connections

7:45 PM **Discussion about Posters and Other Methods as Tools for Sharing the Lived
Experience and Patient Voice**



This project is funded through a Patient-Centered Outcomes Research Institute (PCORI) Eugene Washington PCORI Engagement Award (EASO-42419). Early funding support was provided by the Chan Zuckerberg Initiative (Rare As One, Cycle II) and by the SDS Alliance patient and family community.

Speakers

Host

Eszter Hars, Ph.D.

Host of the EL-PFDD meeting on Shwachman-Diamond Syndrome

Mother to a child who lives with SDS

Founder and CEO, Shwachman-Diamond Syndrome Alliance

Panelists (Patients and Caregivers)

Panel I

- | | |
|------------------|--|
| Nicole S | Mother to Roman (age 10), who lives SDS. Roman was diagnosed at age one, and receives G-CSF therapy due to a history of serious infections |
| Cresta M | Adult living with SDS, diagnosed with SDS as a baby after the loss of her sister in infancy |
| Joyce F | High school student living with SDS and a passionate patient advocate |
| Joe O | Adult living with SDS who was diagnosed as a child and was one of the first SDS patients to successfully receive a stem cell transplant |
| Kaitlyn R | Mother to Ezekiel (age 2), who lives with SDS |

Panel II

- | | |
|--------------------|--|
| Brittanya B | Mother to Jayden (age 5), who lives with SDS and received a stem cell transplant at age 3. |
| Kavan B | Adult living with SDS |
| Will L | Adult living with SDS, who received a stem cell transplant as a child, and many orthopedic surgeries |
| Chilton P | Adult living with SDS, who received a stem cell transplant last year |
| Maria M | Mother to Felicity (age 11), who lives with SDS |

Families Featured in the Documentary Film

Amanda J	and Melvin J , parents to Addie (age 10), who lives SDS, and received a stem cell transplant last year.
Rebecca H	James H , and Emma , family of Declan (age 6), who lives with SDS
Whitner W	Adult who lived with SDS, and her mother Susanne W . Whitner passed away at age 32 from bone marrow failure after a failed transplant during editing of the film.
Erika LC	Mother, and Kassi S , widow of Elijah Thompson , who lived with undiagnosed SDS until he developed MDS/AML at age 23. He passed away from complications of AML at age 24.

Discussion Moderator

James Valentine

Moderator of the interactive audience discussions

Hyman, Phelps, & McNamara.

For the past 18 years, James has been a champion of the patient voice in the drug development process. He previously worked at the FDA as a patient liaison, helping to incorporate the patient voice into medical product review across the FDA's various medical product centers and review divisions. He also helped to develop and launch the Patient-Focused Drug Development initiative at the FDA. In private practice, James has moderated over 70 externally-led PFDD meetings. In addition to serving as the moderator at today's meeting, he and his team have provided expert guidance on its planning.

Guest Expert Speakers

Dr. Martha Donoghue

Introduction and welcome from the FDA

Dr. Martha Donoghue is a board-certified pediatric oncologist who serves as the Associate Director for Pediatric Oncology and Rare Cancers in the FDA's Oncology Center of Excellence, Office of the Commissioner, and the Acting Associate Director for Pediatric Oncology in the Office of Oncologic Diseases, Center of Drug Evaluation and Research (CDER). In these roles, she oversees the implementation of regulations designed to facilitate the timely investigation of drugs and biological products for pediatric patients with cancer, supports and promotes consistency of regulatory work relating to pediatric oncology and rare cancer drug development across CDER and the Center for Biologics Evaluation and Research (CBER), and works with stakeholders to address challenges and foster development of drugs to treat pediatric and other rare cancers. During her 15-year tenure at

the FDA, she has overseen the regulatory aspects and approval of numerous drug and biologic development programs for rare cancers. Areas of special interest include the use of innovative clinical trial designs and real-world data to optimize drug development for rare cancers.

Dr. Kasiani Myers

Clinical overview of SDS

Dr. Myers is a Professor of Pediatrics in the Division of Bone Marrow Transplantation and Immune Deficiency at Cincinnati Children's Hospital Medical Center and Co-Director of the North American SDS Registry. She specializes in hematopoietic stem cell transplantation for bone marrow failure syndromes, with a particular focus on Shwachman-Diamond syndrome — one of the very few clinicians in the country who has dedicated her career to this disease. Her research spans leukemogenesis, transplant outcomes, and the development of rational surveillance strategies for SDS patients at risk of malignant transformation. She has led or co-authored many of the field's most important clinical studies on SDS, and her work has directly shaped current transplant protocols and surveillance guidelines. For the SDS community, Dr. Myers represents both clinical expertise and genuine commitment — a physician who knows this disease, knows these families, and has spent her career working to improve outcomes for both.

Dr. Alyssa Kennedy

Overview of the research landscape and therapies for SDS

Dr. Kennedy is a physician-scientist and assistant member at St. Jude in the departments of Hematology and Pathology. She takes care of patients with bone marrow failure and leukemia. She has previously worked in Boston with Dr. Shimamura's team and published a groundbreaking article on clonal hematopoiesis with Dr. Coleman Lindsley.

Vanessa Merker

Summary of the learnings and highlights of the EL-PFDD meeting

Dr. Merker is a health services researcher at Massachusetts General Hospital who works to improve clinical trials and clinical care for people with rare, genetic diseases. She specializes in collecting information directly from patients and caregivers about their healthcare experiences and needs, using tools such as patient-reported outcome measures and qualitative interviews. She is also an Assistant Professor in Neurology at Harvard Medical School and serves on the SDSA Expert Advisory Board.

PFDD Discussion Questions

Topic 1: Health Effects and Daily Impacts

Panel (30 min)

A panel of five patients and caregivers provides comments to start the discussion on health effects and daily impacts of the condition.

Large-Group Facilitated Discussion on Topic 1 (65 min)

Patients and patient representatives in the audience are invited to add to the dialogue.

Questions:

1. Of all the symptoms that you/your child experiences because of Shwachman-Diamond Syndrome or SDS, **which 1-3 symptoms have the most significant impact on your life?** Examples may include frequent infections, fatigue, feeding challenges, learning difficulties, etc.
2. Are there **specific activities that are important to the patient, but they cannot do** at all or as fully as they would like because of SDS? Examples of activities may include participation in sports or social activities, driving a car, maintaining a job, or keeping up in school, etc.
3. As it relates to SDS, **what does a good day look like?**
What does a bad day look like?
4. How have SDS and its symptoms **changed over time?**
 - a. Would you define your condition (SDS) today as being **well-managed?**
5. **What worries you most** about having SDS?

Topic 2: Current Approaches to Treatment

Panel (30 min)

A panel of five patients and caregivers will provide comments to start the discussion on current approaches to treatment for the condition.

Large-Group Facilitated Discussion on Topic 2 (60 min)

Patients or patient representatives in the audience are invited to add to the dialogue.

Questions:

1. What are you **currently doing to help treat SDS or its symptoms**? Examples may include prescription medicines, hematopoietic stem cell transplant (aka bone marrow transplant), over-the-counter products, and other therapies, including non-drug therapies such as diet modification.
 - a. What are you **currently doing to help monitor/detect risks or complications** posed by SDS (i.e., issues that could happen in the future due to SDS)? Examples may include regular blood draws and/or bone marrow biopsies, and liver function tests.
 - b. How has your/your child's **treatment regimen changed over time**, and why?
2. **How well does your/your child's current treatment regimen treat the most significant symptoms of SDS**? For example, how well do your treatments improve your ability to do specific activities?
 - a. **How well** have these treatments worked for you/your child as your SDS symptoms have **changed over time**?
3. What are the **most significant downsides** to your/your child's current treatments, and how do they affect your daily life? Examples of downsides may include risks and side effects, hospital visits for treatment, and time devoted to treatment.
4. Short of a complete cure, **what specific things would you look for in an ideal treatment for SDS**?
 - a. What would you consider to be a **meaningful improvement** (for example, symptom improvements or functional improvements) in your condition that a treatment could provide?
 - b. **Are there types of risks or adverse events** that you would not be willing to tolerate?
 - c. Are there **types of administration** of treatments that would pose a bigger or smaller burden (e.g., oral pill vs injection)?

Meeting Recording and Photography

The full recording of the meeting, as well as event photography, are available on our website at www.SDSAlliance.org/pfdd

Live Polling

We conducted interactive live polling during the meeting, as well as a comprehensive survey covering the same topics before and after the meeting. The results of both will be available in a separate document and posted on our website at www.SDSAlliance.org/pfdd. For inquiries, please email us at patientvoice@sdsalliance.org. Questions covered include:

#	Question	Answer options
Intro/Demographic questions		
1	Are you a patient (person with SDS) or a caregiver?	<ul style="list-style-type: none"> • Patient (self) • Caregiver (parent, spouse, etc.)
2	Where do you currently reside?	<ul style="list-style-type: none"> • US - Eastern time zone • US - Central time zone • US - Mountain time zone • US - Pacific time zone • US - Alaska or Hawaii time zone • Canada • Latin America • Europe and UK • Australia • Asia • Middle East • Other
3	What sex was assigned to the patient at birth?	<ul style="list-style-type: none"> • Female • Male • Other
4	How old is the SDS patient?	<ul style="list-style-type: none"> • 0-4 years of age • 5-9 years of age • 10-17 years of age • 18-35 years of age • 36-55 years of age • 56+ years of age or older
5	How long has it been since the SDS symptoms first started?	<ul style="list-style-type: none"> • Less than 1 year ago • 1-2 years • 3-5 years • 6-10 years • 11-20 years • Greater than 20 years
5B	At what age was the patient officially diagnosed with SDS?	<ul style="list-style-type: none"> • In utero (before birth) • Under 1 year of age • 1-2 years • 3-5 years • 6-10 years • 11-20 years • 21-30 • 31-40 • 41-50 • 51 or older
5C	How was the SDS diagnosis established?	<ul style="list-style-type: none"> • Genetic testing confirmed the SDS diagnosis (SBDS gene) • Genetic testing confirmed the SDS diagnosis (EFL1 gene) • Genetic testing confirmed an SDS-like diagnosis (SRP54 or DNAJC21) • Clinical observation - no genetic testing performed • Clinical observation - genetic testing was inconclusive or negative • Unsure/Other
Topic 1: LIVING WITH SDS / SYMPTOMS AND DAILY IMPACT		
6	Which of the following SDS-related health concerns has the patient ever experienced? Select ALL that apply.	<ul style="list-style-type: none"> • Neutropenia (low counts of neutrophils) • Pancreatic exocrine insufficiency • Bone marrow failure (low platelets and/or low red blood cells) • Small stature or slow growth • Knee or hip issues requiring surgery or orthopedic services • Immune deficiency - frequent or severe infections • Digestive tract issues (other than the pancreas) • Developmental delay or cognitive impacts • Eczema or other skin issues • Mood and mental health issues • Issues with blood sugar regulation or diabetes • MDS/leukemia • Other

7	Select the TOP 3 most troublesome SDS-related health concerns that the patient has experienced. Select up to 3	<ul style="list-style-type: none"> • Neutropenia (low counts of neutrophils) • Pancreatic exocrine insufficiency • Bone marrow failure (low platelets and/or low red blood cells) • Small stature or slow growth • Knee or hip issues requiring surgery or orthopedic services • Immune deficiency (other than neutropenia) • Digestive tract issues (other than the pancreas) • Developmental delay or cognitive impacts • Eczema or other skin issues • Mood and mental health issues • Issues with blood sugar regulation or diabetes • MDS/leukemia • Other
8	What specific activities of daily life does the patient struggle with due to SDS that are most important to them? Select TOP 3.	<ul style="list-style-type: none"> • Daily chores in and outside of the house • Participating in sports and other recreational activities • Other hobbies • Driving a car • Socializing • Family relationships • Travel/vacationing • Maintaining personal hygiene • Attending to medical appointments and managing medical treatments • Attending school or having a job • None • Other
9	What worries you most about SDS's impact in the future? Select TOP 3	<ul style="list-style-type: none"> • Lack of independence • Loss of social support • Becoming a burden to my family/friends • Stress of not knowing if/when leukemia may develop • Symptoms (other than leukemia) may get worse, or new symptoms may come up • Reduced life expectancy/shortened life • Other

Topic 2: PERSPECTIVE ON CURRENT AND FUTURE APPROACHES TO TREATMENT

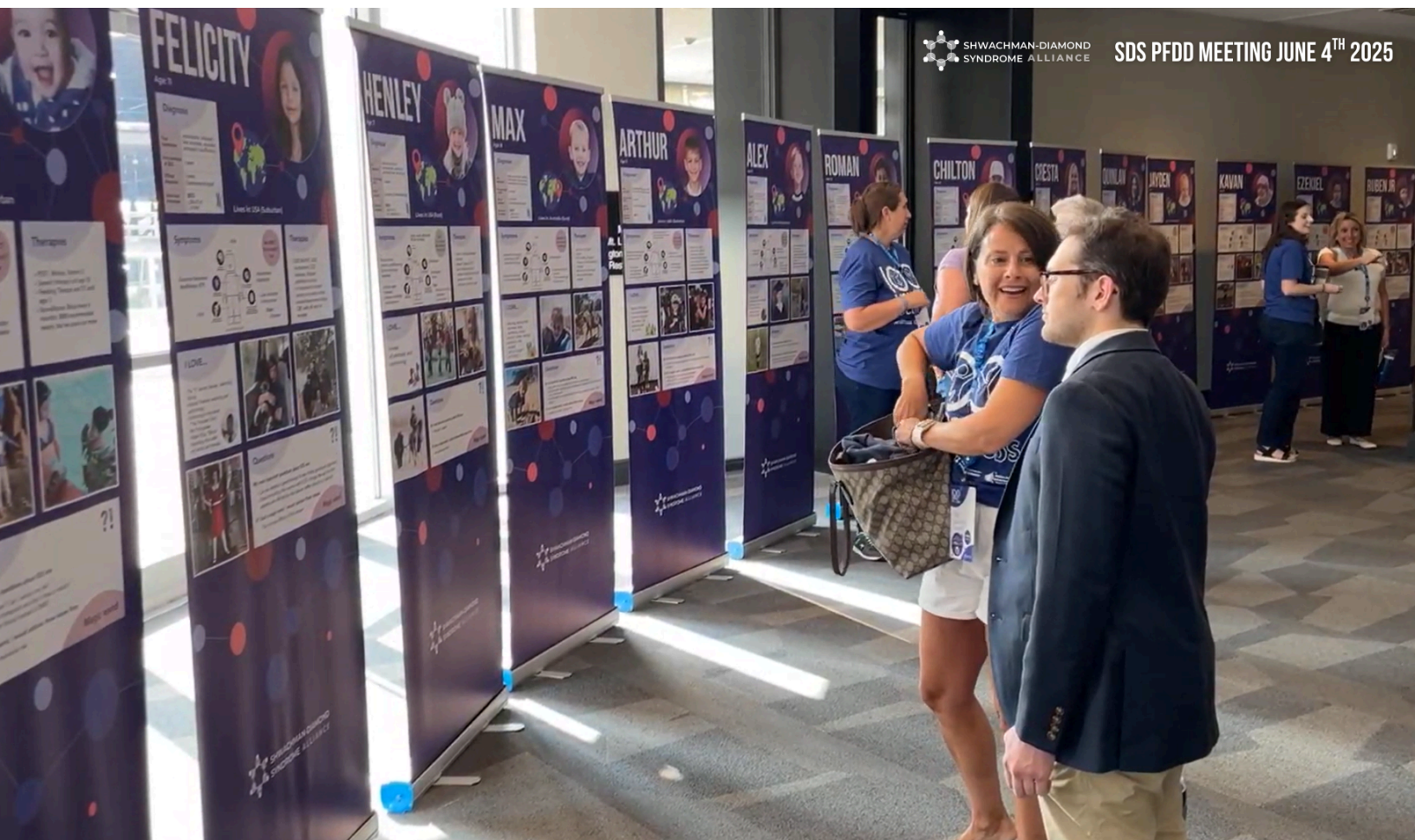
10	What medications or medical treatments has the patient used to treat symptoms associated with SDS? Select ALL that apply	<ul style="list-style-type: none"> • Pancreatic enzyme replacement therapy PERT (e.g., Creon, Zenpep, Viokace, etc.) • Fat-soluble vitamins (A, D, E, or K) • Laxatives/stool softeners (such as Mirlax) • Feeding tube • Blood/platelet transfusions • Granulocyte-colony stimulating factor (G-CSF) (e.g., Neupogen, Zarzio, Nivestym, etc.) • Immunoglobulin replacement therapy, like IVIG or SubQ IG • Prophylactic long-term use of antibiotics • Bone marrow transplant (stem cell transplant) • Growth hormone treatment • Orthopedic or other surgery • Medication for anxiety, depression, or ADHD • Participated in a clinical trial for an investigational drug • No medications or medical treatments • Other
11	Besides medications and treatments, what is the patient currently doing to help manage the symptoms of SDS? Select ALL that apply	<ul style="list-style-type: none"> • Diet modification • Physical therapy/occupational therapy • Isolation and/or masking • Psychologist/counseling • None • Other
12	How well does the current treatment regimen control the patient's symptoms overall?	<ul style="list-style-type: none"> • Not at all • Very little • Somewhat • To a great extent • Not applicable (not using anything)
13	What are the biggest drawbacks of your current approaches? Select up to three	<ul style="list-style-type: none"> • Not very effective at treating target symptoms • Only treats some, not all, symptoms • High cost or co-pay, not covered by insurance • Limited availability or accessibility • Risks and side effects • Route of administration (how it's taken) • Requires too much effort and/or time commitment • Other

Patient Posters

As a conversation starter and to give patients and families an opportunity to share their experiences in a new, impactful way, we created a series of large posters (pop-up banners) that were displayed at the Project PACER kick-off meeting, the EL-PFDD meeting, and the International Scientific Congress in Cincinnati, OH, on June 4th, 2025. The banners are portable, and we are planning to display them at additional meetings as opportunities arise. The objective was to highlight their experiences, perspectives, and humanity. We invited feedback about the posters from both the patient community and other stakeholders. The feedback was overwhelmingly positive. Inquire at patientvoice@sdsalliance.org.

To view digital copies and to zoom in, visit our website here:

<https://sdsalliance.org/pacer#patient-posters>



This Shwachman-Diamond Syndrome Voice of the Patient report, plus supplemental materials, related publications, background information, and the full meeting recording, are available on our website at www.SDSAlliance.org/pfdd

