SMITH-KINGSMORE SYNDROME FOUNDATION

Creating A Sunny World of Hope

ANNUAL REPORT 2019/2020
Letter from the President

It has been an exciting inaugural year at the Smith-Kingsmore Syndrome Foundation (SKSF)! Last August, a small group of families came together to build a vision where individuals with SKS and their families can live in a sunny world of hope through increased global awareness and cutting-edge medical and scientific breakthroughs. After completing multiple forms and assigning responsibilities, the foundation incorporated in Cincinnati, OH in August 2019.

There was no time to relax, however, because the foundation helped support the first-ever SKS family conference at Cincinnati Children’s Hospital Medical Center (CCHMC) in October. 16 families from across the US, the UK and Brazil attended this event, meeting with doctors and researchers from CCHMC and other medical research institutions. A dinner was held on the first evening of the event and it felt more like a family reunion than individuals meeting each other for the first time. At the close of this conference, our group committed to plan our next SKS family conference for fall, 2021. Stay tuned for more details!

In January, 2020, SKSF received its first official donation from the 2019 Blue Signal Gives Back Initiative. Blue Signal’s commitment towards our purpose was a great way to start the calendar year on a high note. We participated in Rare Disease Month in February by having families share their stories of their SKS diagnosis journeys on our website and social media.

Despite challenges associated with the global pandemic, our organization kept focused on our mission. In April, after filling out the long-form 1023 application, we received our official recognition as a 501c3 charity organization. Our fundraising began in earnest with a virtual event of the first-ever SKS Workout of the Day (WOD), raising over $11,500! With our digital store we sold SKS t-shirts, tanks and hats in preparation for this month’s first SKS Awareness Day on August 15.

This spring we assembled a stellar group of doctors and researchers on our medical and scientific advisory and held our first meeting in June. This has assisted in our program development to fund research. Our partnership with Coordination of Rare Diseases at Sanford (CoRDS) will allow us to develop a patient registry which will further serve to demystify this new and rare condition. We are creating a brochure for families to show their medical professionals who may be unfamiliar with SKS. We joined Global Genes and the National Organization of Rare Diseases (NORD) in order to leverage community support and were recognized as a platinum member. This fall, we plan to participate in a virtual newborn screening workshop with 3 patient advocacy organizations for other genetic conditions. Earlier diagnoses for families allows for more targeted treatments for their children.

As we move into 2021, we will continue to focus on research which impacts our SKS community. We will continue to seek out creative and diverse ways to develop funding for basic, translational and clinical research. Your support will allow us to create an environment conducive to accelerated medical breakthroughs. We thank all of our sponsors, clinicians, researchers, families and friends. Your generosity and support has helped make the Smith-Kingsmore Syndrome Foundation’s first year in existence a productive one. I look forward to seeing what 2021 holds in store for our community.

Thank you,

Kristen Bales
Who We Are

Our Vision:
Creating a sunny world of hope for children with Smith-Kingsmore Syndrome by improving their quality of life through medical advancements and community support.

Our Mission:
To improve the quality of life for children and families impacted by Smith-Kingsmore Syndrome by supporting cutting-edge research and collaboration among medical professionals and strengthening community through worldwide awareness.

Our Core Values:
1. Support: Leading with love, kindness, and support in every action we take and every decision we make.
2. Service: Supporting our community’s greater good in order to change the world.
3. Collaboration: Identifying and leveraging strengths within our community and the medical community we partner with in order to effectively execute our mission.
4. Integrity: Treating every individual with respect and honoring our commitments - including using our resources to achieve the greatest possible impact for our community.
5. Transparency: Providing transparency to our community explaining where we’ve been and where we’re going.

Our Goals:
1. To continue engaging top medical and research experts throughout the world to work collaboratively, advancing the science of this genetic condition.
2. To fund different avenues of research that impact the greatest percentage of our population.
3. To launch a SKS patient registry to make comprehensive data points available for all researchers.
4. To define and better understand the multiple medical complications arising from SKS through natural history studies and patient registry.
5. To host a conference in 2021 to increase the collaboration between families and researchers to accelerate understanding of the condition.

Who We Serve:
We serve individuals with Smith-Kingsmore syndrome and their families. SKS was recently diagnosed in 2013. It is a rare genetic condition caused by mutations in the MTOR gene (mechanistic target of rapamycin), found at chromosome location 1p36. The specific genetic changes may vary for individuals with SKS, so the symptoms vary too, and can cause a wide range of medical, intellectual, and behavioral challenges. Epilepsy is one common condition resulting from SKS. Currently there are less than 100 patients diagnosed with this condition globally, but that number will continue to increase due to improved access to advanced genetic testing. We serve not only the individuals currently diagnosed with SKS, but also those individuals yet to be born.
**Highlights**

**First Family Conference:**

Cincinnati Children’s Hospital Medical Center hosted the first-ever Smith-Kingsmore Syndrome Family Conference in October 2019. The formation of the Smith-Kingsmore Syndrome foundation was announced during the conference and provided the first opportunity for SKS families to meet in person and build friendships.

**Thank You Blue Signal!**

The Smith-Kingsmore Syndrome Foundation received the first major donation as the recipient of the 2nd Annual Blue Signal Gives Back Initiative and received $2,052.50.
SKS Athletes & Workout of the Day:

The foundation held its first interactive fundraiser by hosting a Workout of The Day event where participants completed a series of exercises over a four-hour period. The event brought out more than 40 athletes and raised over $10,000 in donations.
Facebook Live:

The Smith-Kingsmore Syndrome Foundation strives to bring information and awareness to the SKS community by hosting live interviews with medical experts covering relevant and useful topics. This year the foundation hosted three separate interviews covering Smith-Kingsmore Syndrome information and diagnosis, sleep, and epilepsy.

Discussed SKS History, Diagnosis, and Medical Resources
1.6K Views

Discussed Sleep Importance and Hygiene, Sleep Clock, Sleep Studies, and Sleep Resources
737 Views

Discussed Epilepsy and Seizure Triggers, Identifying Seizures, Monitoring for Nighttime Seizures, and Medical Resources
617 Views
Medical & Scientific Advisory

As part of our mission, we are committed to supporting research investigating the underlying causes of Smith-Kingsmore Syndrome and treatment for this rare disease. The purpose of our Medical & Scientific Advisory (MSA) is to offer expertise on scientific developments, to provide insights on the needs of Smith-Kingsmore syndrome’s population, and to ensure that Smith-Kingsmore Syndrome Foundation’s policies, research, grants, marketing, communications, and publications meet the highest standards of scientific rigor and accuracy. In the future, we hope to accomplish this by awarding research grants from our charitable fundraising efforts.

At our inaugural meeting in June, we were thrilled to connect doctors and researchers all over the world to discuss Smith-Kingsmore Syndrome. We expressed the critical clinician endpoints important to families that research should be focused on improving, current research that is ongoing, and ways to push research forward.

In addition to discussing current research, during the first meeting this team provided valuable feedback on the creation of a Patient Registry (see below). Future work includes the creation of a medical brochure for families to be able to provide their medical provider on symptoms and management of patients living with Smith-Kingsmore Syndrome.

Our MSA Team:

Dr. John Hogenesch
Professor of Pediatrics, Genetics Chair in Systems Biology, Interim Director of Human Genetics, Cincinnati Children’s Hospital Medical Center

Dr. Darcy Krueger
Director, Tuberous Sclerosis Clinic, Associate Professor, Clinical Pediatrics and Neurology, Associate Director, Research in Neurology, Cincinnati Children’s Hospital Medical Center

Dr. Andrew Liu
Associate Professor, Department of Physiology and Functional Genomics, University of Florida College of Medicine

Dr. Victor Martinez-Glez
Investigador CIBERER (U753) [www.ciberer.es], Responsable Sección de Malformaciones Vasculares, Sección de Genética Clínica, Instituto de Genética Médica y Molecular, INGEMM, IdiPaz-Hospital Universitario La Paz [www.hulp.es]

Dr. Ghayda Mirzaa
Assistant Professor, Genetic Medicine, Seattle Children’s Hospital, Co-Director, Northwest Clinical Genomics Laboratory, University of Washington

Dr. Carlos Prada
Associate Professor, Clinical Genetics, UC Department of Pediatrics, Co-Director, Rasopathy Program, Co-Director, Neurofibromatosis Program, Cincinnati Children’s Hospital Medical Center

Dr. David Smith
Assistant Professor, Co-Director, Center for Circadian Medicine, Divisions of Pediatric Otolaryngology, Pulmonary Medicine, and the Sleep Center, Cincinnati Children’s Hospital Medical Center, Department of Otolaryngology-Head and Neck Surgery, University of Cincinnati College of Medicine
Patient Registry:

We are pleased to announce that we have joined Sanford CoRDS to create our own comprehensive SKS Patient Registry, with the help of the SKSF Medical and Scientific Advisory Team. Based at Sanford Research, a nonprofit research institution, CoRDS is a centralized international patient registry for all rare disease. Sanford CoRDS coordinates the advancement of research for over 7,000 rare diseases, supporting rare disease communities to build robust registries, providing researchers with the information they need to drive research forward. This service is free to families and researchers. Live enrollment will commence in Fall 2020.

Dr. Laurie Smith


Dr. Kate Tatton-Brown

Professor, Clinical Genetics and Genomic Education, Consultant Clinical Geneticist, St George’s University NHS Foundation Trust, London
Treasury Report

Money We Raised

$16,863

72% WOD
12% Blue T-Shirts
7% Signal
9% Other

May'20

90% Savings
10% Expenses

$15,094
Saved for Research
Thank You To Our Donors!

Robert Agnello
Jennifer Alge
Ashley Alvarado
Terri Ashhurst
Sayyad Bacchus
Wadi Bacchus
Jonathan & Jenny Barr
Reid Beggs
Christina Bembeneck
Kimberly Bergman-Gandara
Susan Berres
Pamela Blizzard
Blue Signal
Benjamin Boivin
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Arin Gile
Charlotte Gill
Annette Goodson
Rich Gordon
Matthew Gouge
Ellen Groseclose
Heather Groseclose
Mike & Kristen Groseclose
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Laura Haberstroh
Sara Hancock
Joe Hanrahan
Darlyn Hargis
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Hunter Kirk
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Jill Lindsey
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Glenda Parker
Jennifer Paterson
Nicholas Peregoy
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Francisco Pulido
Kristina Ramos
Wesley Reid
April Reneau
Susan Risty
David Salazar
Chris Sampson
Kathy Sattele
Cornelia Schadler
Mitch Schott
Mary Kay Schrock
Dennis Shurin
Jeremy Simmons
Jen Smith
Allira Smith-Connor
Lauren Stadig
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Looking Ahead

• Funding SKS research directly.
• Encouraging publication of current and future studies in medical journals.
• Engaging with other rare genetic condition foundations.
• Planning 2021 family conference.
• Continuing awareness-building through family outreach, social media campaigns, and other media outlets.
• Future goals:
  ▪ To reduce the average age of diagnosis through increased testing, newborn screening and general awareness in the medical community.
  ▪ To reduce the frequency, intensity and commonality of seizures in the SKS condition through pharmaceutical or other medical breakthroughs and interventions.
  ▪ To improve average duration and sleep quality in persons with SKS through MTOR/circadian research and technological advances in sleep tracking.
  ▪ To improve motor, communication and other skills impacted by SKS through medical advances.