Smith-Kingsmore
SYNDROME FOUNDATION

Smith-Kingsmore Syndrome Research Strategy

AUGUST 2026

Accelerating progress towards better treatment and a cure

4th International SKS Family & Research Conference, Chicago, 2026

OUR RESEARCH STRATEGY

Smith-Kingsmore syndrome (SKS) is a rare genetic condition caused by variants in the MTOR gene. While our understanding of SKS has grown significantly, fundamental questions remain about how the condition affects each individual, how symptoms can best be treated, and which therapies could make a meaningful difference.

Our research strategy is focused on improving treatment options for people living with SKS today, while building the scientific foundation for more precise treatments and, ultimately, a cure.

As a rare disease foundation, we must be strategic about where we invest our resources. We will focus our investments where they can have the greatest impact: filling critical gaps, building the tools and data needed to advance research, bringing the right people together, and moving promising ideas closer to patients.

Throughout this work, the experiences and priorities of people with SKS and their families will be our north star, guiding where we focus and invest.

First International SKS Family & Research Conference, Cincinnati, 2019

Our Vision

Our vision is a world cured of Smith-Kingsmore syndrome.

Our Mission

Our mission is to improve the lives of all people impacted by Smith-Kingsmore syndrome by accelerating research and connecting our global community.

Who We Are

The Smith-Kingsmore Syndrome Foundation is a registered non-profit organization, founded in 2019. It is the only global non-profit funding SKS research and providing education and support to the Smith-Kingsmore syndrome community.

Dr Carlos Prada, Lurie Children's; Dr Matt Hall, NCATS; and Dr Darcy Krueger and Lindsey Aschbacher-Smith, MS, CCRP, Cincinnati Children's, at the 2026 SKS Family & Research Conference

WHERE WE ARE HEADED

Our Research Priorities

1. Improve treatment options now

Accelerate research and clinical understanding that can lead to better treatment decisions for people with SKS.

2. Build the science for precision treatments

Develop the knowledge, models, data, and tools needed to understand SKS biology and enable targeted therapies.

3. Grow a connected SKS global community

Bring together families, clinicians and researchers to accelerate discovery and sustain progress over the long term.

Enjoying the 2026 SKS Family & Research Conference

"We want transformative treatments for SKS. But we also know that progress doesn't have to be transformative to matter.

An improvement in something that affects daily life, every day, can be incredibly meaningful."

SUSAN DANDO

Executive Director
Smith-Kingsmore Syndrome Foundation

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Improve Treatment Options Now

People with SKS need better treatment options today. We will pursue opportunities to learn from existing medicines, strengthen our understanding of how SKS affects people, and turn emerging evidence into useful clinical information.

Our focus will be on research that has a realistic path toward improving care and treatment in the near term.

We will:

Accelerate drug repurposing research
Use SKS models to systematically test existing medicines and identify promising candidates for further study.

Build a clearer picture of SKS
Collect and strengthen data on genetic variants, inheritance, and the presence and severity of key symptoms across the SKS population.

Create pathways from research to treatment
Develop approaches for evaluating promising repurposed or off-label medicines in people with SKS, including clinical protocols and trials where appropriate.

Improve access to trusted treatment information
Develop practical resources on the clinical questions that matter most to families and clinicians.

Support earlier diagnosis
Advocate for inclusion of MTOR/SKS on appropriate diagnostic panels so that more people can receive an accurate diagnosis sooner.

2

Build the Science for Precision Treatments

Dr Christine Tabuloc answers families' questions about SKS research at the 2026 SKS Family & Research Conference

"Working with the SKS community for the past 3 years has further fueled my passion for science and discovery.

I am grateful to meet and work with the wonderful people who my research directly impacts."

CHRISTINE TABULOC, PhD

Researcher at the University of California, Davis, whose SKS research is supported by the Smith-Kingsmore Syndrome Foundation

SKS is caused by changes in MTOR, a gene at the heart of a complex biological pathway. Understanding exactly how different SKS-associated variants alter this biology, and how those changes relate to symptoms, is essential to developing more targeted treatments.

We will invest in the science, models, samples and clinical data that researchers need to answer these questions and move promising discoveries toward patients.

We will:

Deepen our understanding of SKS biology
Support research into how SKS-associated MTOR variants alter cellular signaling and lead to the features of SKS.

Build the tools researchers need
Continue to develop and support research models, including flies and mice, and expand access to cell lines, patient-derived samples and biobanking.

Understand differences across SKS
Improve how we characterize individuals with SKS, helping researchers understand why symptoms vary and identify groups that may respond differently to treatment.

Develop meaningful ways to measure change
Support development of clinical severity measures and identify outcomes that can be used to determine whether a treatment is working.

Learn from real-world treatment experience
Use registry and clinical data to better understand treatment outcomes, including experiences with off-label and compassionate-use therapies.

Together, these investments will create the foundation needed to move from broadly treating symptoms toward more precise treatments based on SKS biology.

Kristen and Mike Groseclose, founders of the Smith-Kingsmore Syndrome Foundation, and their son Jack

"For 15 years of Jack's life, we only had an autism diagnosis. We didn't know anyone who had similar daily challenges and we felt alone in our struggles.

Getting an SKS diagnosis inspired us to partner with other SKS families, doctors and researchers to create meaning from our pain. We believed that with all of us united, working together, we could improve the quality of life for families like ours."

KRISTEN GROSECLOSE

Co-Founder
Smith-Kingsmore Syndrome Foundation

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Grow a Connected Global SKS Community

Rare disease research moves faster when people work together.

The Smith-Kingsmore Syndrome Foundation has an important role to play as a connector: bringing together researchers studying SKS and related biology, clinicians caring for people with SKS, and families whose experience helps identify the questions that matter most.

We will continue building a collaborative global community around SKS research so that knowledge, resources and ideas can travel further and faster.

We will:

Bring researchers together
Actively build collaborations across laboratories, academic institutions and related rare disease communities to share knowledge and accelerate discovery.

Connect research and clinical expertise
Strengthen research representation within the Foundation's scientific advisory structure and continue building relationships with clinicians who care for people with SKS.

Bring families, clinicians and researchers into the same conversation
Continue supporting the SKS Family & Research Conference and other opportunities for collaboration around shared questions and priorities.

Advocate for SKS
Build resources that help families and clinicians raise awareness of SKS and encourage broader engagement from the medical and scientific communities.

Draw on the strengths of our community
Use expertise from across the SKS community, including science, medicine, data, communications, legal and other fields, to strengthen the Foundation's ability to advance research over the long term.

Sarah Lepore discussing the work of the Smith-Kingsmore Syndrome Foundation with families, researchers and clinicians

"Research is most powerful when it is connected to the people it is meant to serve. By bringing families, clinicians, and researchers together, we can ensure that our community helps shape the science.

When we bring those voices together, we create the opportunity to turn research into meaningful change for people living with SKS."

SARAH LEPORE
President
Smith-Kingsmore Syndrome Foundation

How We Will Work

The SKS Foundation cannot do this work alone.

Our role is to identify the barriers slowing progress and determine where a patient-led foundation can have the greatest impact. Sometimes that means funding research directly. Sometimes it means creating a model, dataset or resource that makes new research possible. And sometimes our greatest contribution is connecting the right researchers, clinicians and families around a shared problem.

Across all three priorities, we will:

Follow the science
We will adapt our investments as evidence develops and new opportunities emerge.

Focus on patient impact
We will prioritize questions and approaches with the potential to meaningfully improve the lives of people with SKS.

Build resources others can use
We will invest in data, models, samples and tools that can accelerate research beyond any single project.

Collaborate broadly
We will seek expertise and partnerships across SKS, mTOR biology, related rare diseases, academia, medicine and industry.

Use our resources strategically
We will focus Foundation funding where it can fill critical gaps, unlock additional investment or accelerate promising work.

KEY RESOURCES

Smith-Kingsmore Syndrome Foundation - www.smithkingsmore.org

Smith-Kingsmore Syndrome Family & Medical Provider Guide, available in multiple languages - smithkingsmore.org/family-medical-provider-guide/

Prada CE, Raski CR, Mirzaa G, et al. **Smith-Kingsmore Syndrome**. 2025 Nov 20. In: Adam MP, Bick S, Mirzaa GM, et al., editors. GeneReviews®, University of Washington, Seattle; 1993-2026. www.ncbi.nlm.nih.gov/books/NBK619247/

Odylia Therapeutics, **Smith-Kingsmore Syndrome Landscape Analysis.** odylia.org/resources-2/library/

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