Kabuki SYNDROME FOUNDATION

2023

MIDYEAR REPORT

To Our Global Community

The Kabuki Syndrome Foundation (KSF) remains dedicated to advancing new areas of research that will lead to life-changing treatments for those affected by Kabuki syndrome.

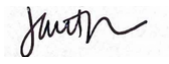
This year we've grown, identified new therapeutic opportunities, and started executing our new plan to accelerate treatments. We have designated over $500,000 to fund Kabuki syndrome research that will have the greatest impact. Without committed families like you, we would not have been able to achieve this incredible milestone.

We are now at the forefront of taking breakthrough research from dream to reality with LEAP, our strategic roadmap. **LEAP - Learn, Enable, Accelerate, Prepare - is the path to finding and accelerating treatments that target the most significant symptoms faced by the Kabuki syndrome community.**

LEAP is a 2 year, $3.5 million innovative research plan that prioritizes projects that will deepen our understanding of this rare disease, which can lead to the development of targeted treatments; enable collaboration between patients, researchers, and clinicians to fast-track progress; and accelerate therapies from the lab to patients through funding and support for promising therapeutics.

We have ambitious goals mapped out to unlock the brightest futures possible for our loved ones. It is essential for families to take initiative and engage in our efforts to support and fundraise for research. With your continued support, we can find treatments faster.

With hope,



Janet Lee, Mom to Emily & KSF Executive Director

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LEAP: The Roadmap to Treatments

LEAP FOR KABUKI SYNDROME TREATMENTS

LEAP is the key to unlocking life-changing treatments for Kabuki syndrome on an accelerated timeline.

We're committed to raising $3.5 million over the next two years to drive LEAP.

The LEAP roadmap was created based on the Kabuki syndrome community's greatest needs, input from our Medical & Scientific Advisory Board, and lessons from over 20 years in rare disease research.

L: LEARN

We are **learning more** about Kabuki syndrome by awarding collaborative Discovery Grants aimed at identifying new therapeutic opportunities, disease-causing targets, and pathways.

Funding needed: $600,000

- **6/2023** - 9 grant applications received
- **11/2023** - First grant of $150K to be awarded
- **4/2024** - New grant round opens

E: ENABLE

We are **enabling collaboration** between patients, researchers, and clinicians which is essential to fast-tracking treatments for rare diseases.

Funding needed: $1.8 million

- **10/2023** - Scientific Symposium for Researchers
- **Ongoing** - Development of a Kabuki Syndrome Consortium (contracts currently in review at 5 institutions)
- **11/2023** - Kabuki Syndrome Outcome measures & biomarker Consortium to be launched

A: ACCELERATE

We are **accelerating therapies** from the lab to patients by funding and supporting promising therapeutics, meaningful translational research, and patient enrollment in clinical trials.

Funding needed: $1 million

- **Ongoing - consulting with ORYZON** on HOPE (Vafidemstat) clinical trial design

P: PREPARE

We are **preparing for clinical trials**. KSF is driving the validation of several outcome measures, supporting patient registry efforts, improving diagnostic methods, and creating accessible resources for our community.

Funding needed: $100,000

- **Ongoing -** Optimizing **patient data** collection and recruitment

Dive into LEAP with us! Join our next live webinar and catch up with the recordings on our YouTube channel: @kabukisyndrome Find details and more resources on our site: www.KabukiSyndromeFoundation.org

Growing Our Network to Support You

We are providing our community with immediate access to the skills, scientific knowledge, and industry connections needed to push progress forward.

In April, we welcomed Dr. Bruce Bloom and Dr. Clara Tang, as KSF Chief Scientific Officer and Director of Research. Drs. Bloom and Tang share in our vision for changing lives and have over 20 years combined experience in rare disease treatment development. **In June, they unveiled LEAP at a live webinar. Watch the recording on our Youtube: @kabukisyndrome**

In a collaborative effort, Kabuki UK and KSF were selected to attend the Aspire Biosciences event - a day of networking focused on finding treatments for rare diseases!

In May, Jessica McAndrew joined our Board of Directors. Jessica is a Kabuki syndrome parent and digital marketing powerhouse who's helping expand our global reach and impact.

Kabuki syndrome was represented at **St. Jude Children's Research Hospital's first workshop** to accelerate treatment-focused research in neurological conditions by Dr. Bruce Bloom, KSF Chief Science Officer, and Janet Lee, KSF Executive Director.

KSF Research Ambassador Kristin Anzelc was selected to join the Global Genes RARE Compassion Program. Kristin is an adult with Kabuki syndrome and is partnering with medical students to share her experiences as a patient and research participant. Thank you, Kristin, for representing Kabuki syndrome!

Opportunities & Clinical Trials

The quest for treatments for rare diseases is powered by patient families who refuse to sit on the sidelines.

You're the champions who propel us forward. We're committed to keeping you connected and able to drive research that matters to you. On our website, research opportunities are organized by virtual and in-person. Below, find the latest in clinical trials and stay updated for what's next.

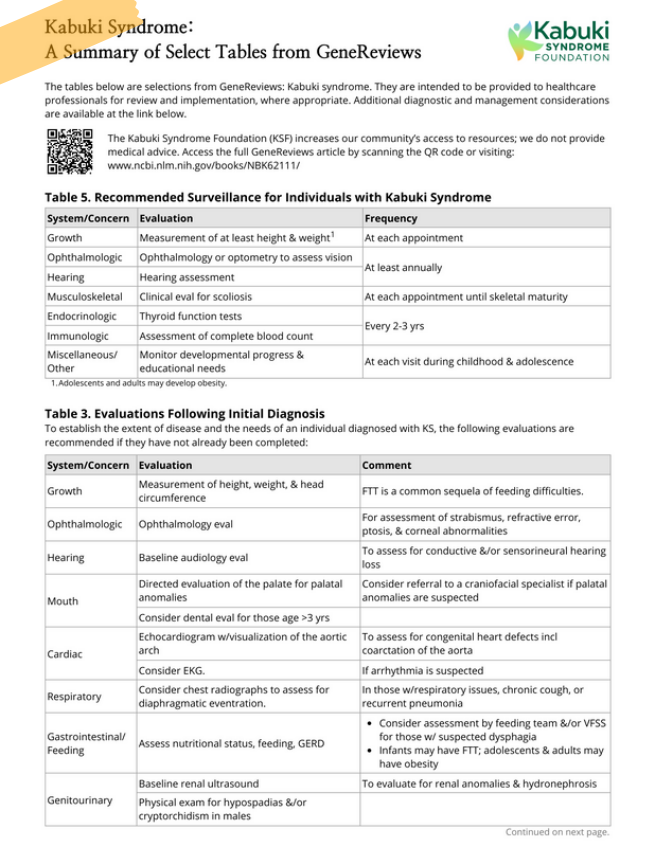
Clinical Trial	Led By	Updates
A Pilot of A Diet Therapy for Kabuki Syndrome (Modified Atkins)	Dr. Jacqueline Harris at Kennedy Krieger Institute	• Closing in October 2023. Results will be shared as soon as available.
HOPE (vafidemstat)	ORYZON; site locations and PIs to be announced	• KSF is now consulting with ORYZON on clinical trial educational materials

» Thanks to your input in our dietary survey, Dr. Harris is exploring expansion opportunities to include younger participants in a future version of the trial!

» Stay tuned for an update from ORYZON at our virtual family conference on Tuesday, November 7, 2023.

» **What's coming next - data and sample collection.**
In Q4, we'll be announcing the two most impactful ways you can participate in LEAP. **P is for Preparing for Clinical Trials.**

Tools For Families & Providers



Kabuki Syndrome:
A Summary of Select Tables from GeneReviews

Kabuki SYNDROME FOUNDATION

The tables below are selections from GeneReviews: Kabuki syndrome. They are intended to be provided to healthcare professionals for review and implementation, where appropriate. Additional diagnostic and management considerations are available at the link below.

The Kabuki Syndrome Foundation (KSF) increases our community's access to resources; we do not provide medical advice. Access the full GeneReviews article by scanning the QR code or visiting: www.ncbi.nlm.nih.gov/books/NBK62111/

Table 5. Recommended Surveillance for Individuals with Kabuki Syndrome

System/Concern	Evaluation	Frequency
Growth	Measurement of at least height & weight[1]	At each appointment
Ophthalmologic	Ophthalmology or optometry to assess vision	At least annually
Hearing	Hearing assessment	
Musculoskeletal	Clinical eval for scoliosis	At each appointment until skeletal maturity
Endocrinologic	Thyroid function tests	Every 2-3 yrs
Immunologic	Assessment of complete blood count	
Miscellaneous/ Other	Monitor developmental progress & educational needs	At each visit during childhood & adolescence

1. Adolescents and adults may develop obesity.

Table 3. Evaluations Following Initial Diagnosis

To establish the extent of disease and the needs of an individual diagnosed with KS, the following evaluations are recommended if they have not already been completed:

System/Concern	Evaluation	Comment
Growth	Measurement of height, weight, & head circumference	FTT is a common sequela of feeding difficulties.
Ophthalmologic	Ophthalmology eval	For assessment of strabismus, refractive error, ptosis, & corneal abnormalities
Hearing	Baseline audiology eval	To assess for conductive &/or sensorineural hearing loss
Mouth	Directed evaluation of the palate for palatal anomalies	Consider referral to a craniofacial specialist if palatal anomalies are suspected
	Consider dental eval for those age >3 yrs	
Cardiac	Echocardiogram w/visualization of the aortic arch	To assess for congenital heart defects incl coarctation of the aorta
	Consider EKG.	If arrhythmia is suspected
Respiratory	Consider chest radiographs to assess for diaphragmatic eventration.	In those w/respiratory issues, chronic cough, or recurrent pneumonia
Gastrointestinal/ Feeding	Assess nutritional status, feeding, GERD	• Consider assessment by feeding team &/or VFSS for those w/ suspected dysphagia • Infants may have FTT; adolescents & adults may have obesity
Genitourinary	Baseline renal ultrasound	To evaluate for renal anomalies & hydronephrosis
	Physical exam for hypospadias &/or cryptorchidism in males	

Continued on next page.

Just diagnosed or wondering what evaluations are recommended for people with Kabuki syndrome?

Check out the *For Families* tab of our website and download your copy.

We're on YouTube! @kabukisyndrome

Learn from adults with Kabuki syndrome, parents who have successfully navigated school options, and researchers who lead deep dives into symptoms and more.

Did you know that 12 organizations around the globe support people with Kabuki syndrome? A local network may be closer than you think. Find yours: www.KabukiSyndromeFoundation.org/for-families

Thank you, Kabuki UK for welcoming Clara, KSF Director of Research, to your Family Day in June!

We're committed to being the global hub of Kabuki syndrome research and resources for families. Find these tools and more on our website.

50 Recordings in our YouTube library

11 Research opportunities

10 Expert physicians around the globe

www.KabukiSyndromeFoundation.org

Save the Dates

Our webinars and conferences are your bridge to our global network of experts.

Events for Our Community

August 1

Live Webinar

Dive into LEAP: LEARN the Science Behind the Symptoms. We'll break down the science of the syndrome and how it relates to ongoing research and potential treatments.

November 7

Virtual Community Conference

Our biggest event of the year featuring panelists from the community, tools to hep you now, and research updates from Kabuki syndrome experts. Discover what actions you can take to find treatments faster with us.

Events for Researchers & Clinicians

October 26

Virtual Scientific Symposium

Our inaugural Scientific Symposium is designed to foster collaborations, encourage knowledge-sharing, and enable professionals to share their latest findings related to Kabuki syndrome.

Open Now

Symposium: Call for Abstracts

Prospective speakers are asked to submit an abstract containing a concise summary of original research, published or unpublished. Details and form are on our website at *For Researchers.*

For more details or to submit a question or abstract, visit:
www.KabukiSyndromeFoundation.org

Fueling the Research

We've committed to raising $3.5 million over the next two years to find treatments faster. Your contribution can change lives. It fuels the research, partnerships, and hope that propel us forward.

We thank all our donors and want to take a moment to especially honor those who support Kabuki syndrome research beyond their own lifetimes. We are humbled by your donations received in memory of a loved one. These legacy donations have a lasting impact on improving the quality of life for people with Kabuki syndrome.

How Ordinary Families Become Extraordinary Donors

"Steve had 5 grandchildren and they were one of the most important parts of his life. He loved them all dearly and made it a priority to try and talk with them every day. From the beginning, he had a very special bond with Madison. She was born in 2015 with congenital heart defects and spent 2.5 months in the hospital. Later in 2017, through genetic testing, Madison was diagnosed with Kabuki syndrome (KMT2D). Steve was her best friend, he always met her where she was and really knew how to communicate with her. He loved to spend time with her building towers (knocking them down), playing pretend kitchen, and could always ground her when needed. We lost him too soon.

We are grateful for the support we received from friends and family in honor of him to support Madison and further Kabuki syndrome research."

-Susan Paquette and Kara Willhoite on behalf of The Steve Paquette Memorial Fund

Steve Paquette and his granddaughter, Madison

Change lives with us:
www.KabukiSyndromeFoundation.org/donate/
Please contact us to learn more about planned giving, giving through a DAF, or how we can support your fundraising ideas:
info@kabukisyndromefoundation.org

Funding Summary

Thank you to our donors who have increased our ability to drive research.

$927,000

RAISED TO DATE TO FIND TREATMENTS FOR KABUKI SYNDROME

$531,500
committed to research

$45,000
included travel funds for over 12 families to participate in-person

We are unlocking the healthiest and brightest futures possible for our loved ones.
Donate today to help us deliver treatments to them.

Network Snapshot

2470
social media followers

1485
newsletter recipients

111
KSF-registered researchers & clinicians

The Kabuki Syndrome Foundation (KSF) is a 501(c)(3) nonprofit charity, exempt from federal taxation. Transparency is both a requirement and an obligation that is vitally important to our Directors and staff.

Thank you to our volunteers, partners, staff, Medical & Scientific Advisory Board, and our Board of Directors, who made this report possible.

Thank you for supporting our efforts to find life-changing Kabuki syndrome treatments.

Contact Us

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