2025 Midyear Impact Report

6 Treatment Pathways Gaining Momentum in 2025

The Kabuki Syndrome Foundation (KSF) is the only nonprofit solely dedicated to advancing research toward life-changing treatments for Kabuki syndrome.

We're thrilled to share this incredible start to 2025! Read on about the potential treatments and more.

Thanks to the generosity of our donors, **we have awarded over $560,000 in 2025**, with an additional $1 million allocated to advance Kabuki syndrome research, delivering meaningful impact for families around the world. Here's what we've been working on:

$345,000
PRECLINICAL STUDIES

TESTING TWO POTENTIAL THERAPIES IN KABUKI SYNDROME MOUSE MODELS

Two preclinical studies using KSF-sponsored mouse models are evaluating **Oryzon's vafidemstat** and **exogenous beta-hydroxybutyrate** (BHB), with the goal of uncovering therapies that can improve brain function.

$380,000
DRUG REPURPOSING

FAST-TRACKING TREATMENTS THROUGH DRUG REPURPOSING

We are funding studies to evaluate existing FDA-approved drugs for potential use in treating Kabuki syndrome, shortening the path to clinical trials by building on medicines with known safety profiles.

$145,000
NOVEL THERAPIES

ADVANCING PROOF-OF-CONCEPT RESEARCH FOR GENE-TARGETED THERAPIES

We are supporting research into gene activators that aim to boost KMT2D expression, offering hope for a targeted therapy that could benefit many individuals living with Kabuki syndrome.

$50,000
CLINICAL MANAGEMENT

ESTABLISHING GLOBAL CLINICAL GUIDELINES FOR KABUKI SYNDROME

We brought together more than 45 clinicians and community leaders for a consensus meeting to review 120+ care recommendations, which will shape important international guidelines for managing Kabuki syndrome.

The Clinical Management Guidelines Consensus Meeting, Boston, May 2025

Two preclinical studies funded by KSF are laying the groundwork for potential clinical trials in 2026.

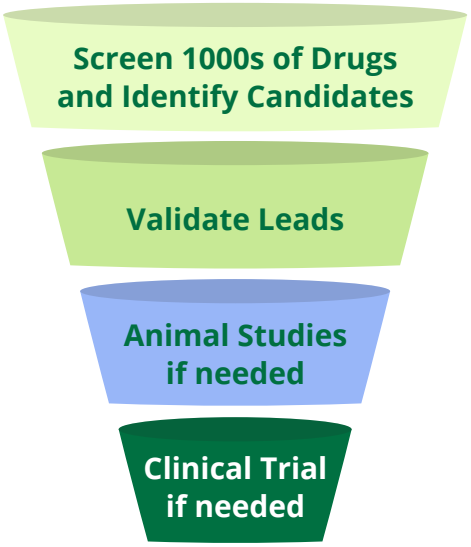
Vafidemstat, a drug that targets the LSD1 pathway, may help improve cognitive and behavioral symptoms in Kabuki syndrome. This study is evaluating its therapeutic potential and will help guide a decision on whether to pursue a $5 million fundraising effort for the first industry-led clinical trial for KS. Results are expected later this year.

Separately, researchers are testing **beta-hydroxybutyrate (BHB)**, a compound linked to the benefits of the ketogenic diet, to see if it can improve memory and brain function in KS mice. This study will determine the optimal dose, with results expected in late 2025.

These studies could lead directly to our first commercial clinical trials.

Repurposed Drugs Offer Fastest Path to Treatments

By identifying existing approved drugs that could also treat Kabuki syndrome, we can get treatments to people faster. Here are the 3 repurposed drug screening programs we're funding:



Boston Children's Hospital: The team at the Roya Kabuki Program has screened hundreds of FDA-approved drugs as potential therapies for both **KS type 1 and type 2**. They are now preparing to test the most promising candidate in animal models, with an update expected in Q4 2025.

Purposeful: Through a KSF-funded grant, researchers at Purposeful are using AI to identify repurposed drug candidates specifically for **KS type 1**. The next phase will involve testing lead candidates in mouse models.

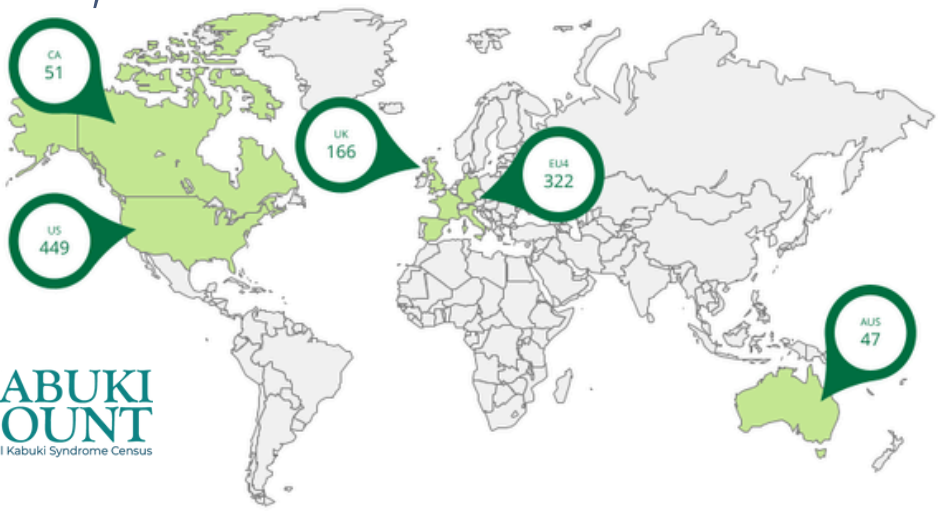
Unravel Biosciences: KSF engaged Unravel Biosciences to identify repurposed drug candidates for **KS type 2**. Once the list of top candidates is finalized, KSF will evaluate the findings and determine the best path forward.

The information provided in this report is for general informational purposes only and is not intended as medical advice, diagnosis, or treatment.

2025 Impact To Date

$1,560,000	4	6	83
allocated towards research in 2025 (to date)	new KSF grants awarded in 2025	therapies advanced with 2025 KSF grants	experts convened in KSF-led meetings (to date)

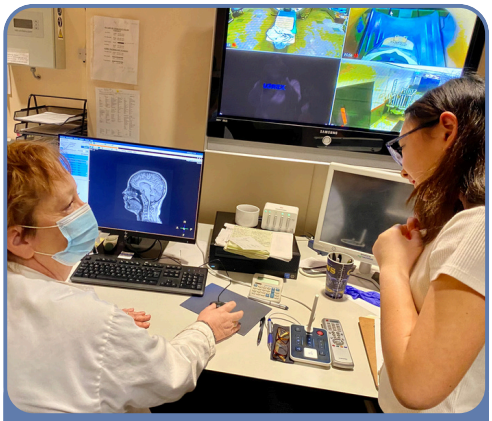
1,237 people in our global contact registry, Kabuki Count



KABUKI COUNT
Global Kabuki Syndrome Census

Building the Tools We Need to Test Treatments:

Investing $1.4 Million to Build The Kabuki Syndrome Outcome Consortium (KSOC)



KSF funded support staff at each KSOC site and covered travel costs for 150 families to meet with Kabuki syndrome experts across North America.

Before treatments can advance to clinical trials, tools are needed to reliably measure whether those treatments are working. With the support of our donors and over 150 participating families, KSF launched the Kabuki Syndrome Outcome Consortium (KSOC)—a collaboration with six leading hospitals across the U.S. and Canada. Together, we're working to validate three key biomarkers by 2026: a DNA methylation signature, metabolomic markers from blood or urine, and Event-Related Potentials (ERP) to measure brain activity.

KSOC researchers are collecting patient samples and clinical data to support this work while building the most comprehensive Kabuki syndrome dataset to date, laying the foundation for future clinical trials and treatment development.

Because of you, progress is not only possible—it's happening.
Thank you for your participation, donations, and support!

Donate today: www.kabukisyndromefoundation.org

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