Kabuki SYNDROME FOUNDATION

2024

A YEAR OF GROWTH

ANNUAL REPORT

January 20, 2025

Despite the well-documented burden and life-long impact of Kabuki syndrome, no approved treatments exist.

KSF is making treatments possible.

KSF has invested $2.4 million in a diverse portfolio of therapeutic strategies, maximizing the chances of success for the Kabuki syndrome community. In 2024, KSF awarded $700,000 in new research grants that can help unlock healthier futures for people impacted by Kabuki syndrome.

KSF Impact to Date

7	$2,400,000	464	1,196
potential treatments supported by KSF	dedicated to Kabuki syndrome research	participants recruited to 6 studies	patients in our contact registry

13 active grants across 14 institutions and 5 industry partners supporting the essential pillars of rare disease treatment development:

- lead candidates
- disease mechanisms
- outcome measures
- drug discovery
- models & tools
- clinical outcomes

Our roadmap to treatments for Kabuki syndrome prioritizes:

- accelerating the most promising treatments from lab to clinical trials
- discovering more about the cellular mechanisms of Kabuki syndrome, new treatment targets, and potential therapeutics utilizing community-managed animal and cell models
- preparing for successful clinical trials by validating outcome measures via the Kabuki Syndrome Outcome measures & biomarkers Consortium (KSOC) and building our global contact registry, Kabuki Count.

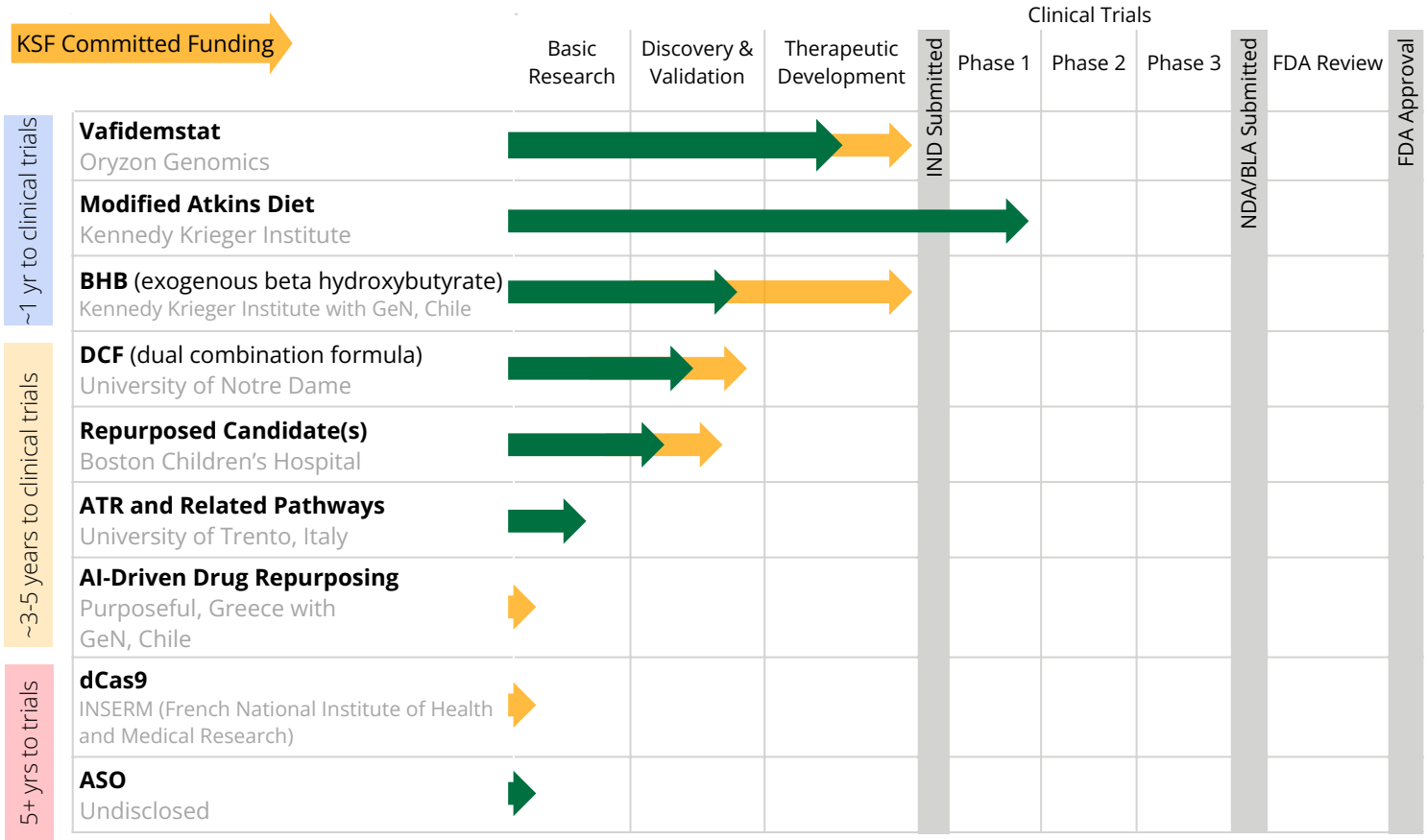
Thank you to those who have participated in research and to our generous donors! Together we are transforming the future of Kabuki syndrome research.

The Kabuki Syndrome Foundation is led by families affected by Kabuki syndrome, leveraging their professional expertise alongside collaborative input from our Industry Advisory Board, Medical & Scientific Advisory Board, and community panels of caregivers & adult patients.

Funding Research, **Delivering Treatments**

Nine potential treatments for Kabuki syndrome are in development. However, inadequate funding remains the greatest obstacle to advancing these treatments into clinical trials. Two trials could begin by the end of 2026, a milestone we can reach with your help. **Raising $5 million could fund a clinical trial for one treatment as early as next year.**

The Kabuki Syndrome Therapeutic Pipeline



To accelerate progress in the therapeutic pipeline, KSF is funding the following projects in 2024-2025:

1. **Advancing two lead candidates toward clinical trials**
   - Funding a study to evaluate the efficacy of Oryzon's compound, vafidemstat, in a mouse model of Kabuki syndrome to de-risk future investments in clinical trials *($300,000 committed)*
   - Determining the dose of BHB that causes symptom improvement in mice *($45,000 committed)*
2. **Identifying repurposed drug candidates**
   - Therapeutics already in use for other disease(s) could be "fast-tracked" for Kabuki syndrome; at least one promising candidate has been identified *($200,000 committed)*
3. **Exploring novel therapeutics**
   - Supporting the discovery of new treatment options and leverage the latest advances in biomedical technology such as AI-driven therapeutic discovery and dCas9.

Scan the QR code or click here to watch a walkthrough of the pipeline:

Celebrating Milestones, **Fueling Progress**

Key Events

Quest for A Cure Galas in Los Angeles and New York collectively raised over $1,300,000

First in-person meeting of KSOC leaders: Established in 2023 with funding of $1,350,000, the Kabuki Syndrome Outcome measures & biomarker Consortium (KSOC) includes experts from 6 institutions across North America

Our Annual Research Conference + 2 Community Webinars were attended by over 300 virtual guests

3 community panels enabled direct caregiver and patient input on grants and research priorities

For Researchers

Monthly lab meetings hosted by KSF are attended by 40+ investigators

Aggregated resources and tools for research available

Annual grant opportunities for translational research

Snapshots

60%
of funds raised originated from private events

$450,000
allocated to reimburse research participants' travel

212
researchers and clinicians registered with KSF

3,482
social media followers

72
videos in our resource library on YouTube

100%
of donations came from families impacted by Kabuki syndrome and their networks!

Through our global network of families, researchers, and clinicians, we are finding treatments faster. However, our work is not yet complete. **Your support will make treatments a reality.**

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MAKE A DONATION

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On behalf of our staff and the families we support worldwide, thank you.

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