

# Global Collaboration Aims for Consensus on Kabuki Syndrome Clinical Management Guidelines

**Kabuki Syndrome Foundation Leads Global Consensus Effort with International Meeting in Boston**



**Boston, Massachusetts Apr 29, 2025 ([Issuewire.com](https://www.issuewire.com))** - The [Kabuki Syndrome Foundation](https://www.kabukisyndrome.org) (KSF) is proud to support the advancement of new Clinical Management Guidelines for Kabuki syndrome. These guidelines will provide standardized, evidence-based recommendations to improve the diagnosis, treatment, and ongoing care of individuals living with this rare genetic disorder.

This May, KSF will host a Clinical Management Guidelines Consensus Meeting in Boston, Massachusetts, bringing together more than 46 international clinicians, researchers, caregivers, and adults with Kabuki syndrome. The goal of the meeting is to finalize guidelines across 14 clinical themes—ranging from cardiology and immunology to neurology and endocrinology. The resulting guidance will be grounded in the latest published research and complimented with real-world insights from the Kabuki syndrome community.

Kabuki syndrome is a complex, multisystem disorder that affects an estimated 1 in 32,000 births. It can cause a broad range of health issues, including intellectual disability, heart defects, immune system dysfunction, hearing loss, and growth challenges. While clinical experience with Kabuki syndrome has grown over the years, comprehensive, internationally recognized clinical management guidelines are

still lacking—leaving many physicians and families without clear direction when navigating care.

This global effort is led by Dr. Siddharth Banka, clinical lead for the guidelines, Professor of Genomic Medicine at the University of Manchester, and a member of the KSF [Medical and Scientific Advisory Board](#). Since 2022, KSF has partnered with Dr. Banka, who is also a Consultant Clinical Geneticist at [Manchester University NHS Foundation Trust](#) (MFT), and other international experts to develop updated clinical guidelines for Kabuki syndrome.

This work is also supported by the MFT-based Manchester Rare Conditions Centre (MRCC) and the Medical Research Council and National Institute for Health and Care Research (NIHR) funded UKRD EpiGenRare node, whose involvement further strengthens the collaborative nature of the project. KSF staff and volunteers have played a key role in supporting this work—collecting and reviewing current Kabuki syndrome publications, creating digital platforms for expert submissions, and organising the upcoming consensus meeting in Boston, MA, where the recommendations will be finalized.

By bringing together a diverse, international group of experts and community advocates, the project ensures the most well-rounded and inclusive approach to care recommendations.

*“Clinical management guidelines provide clarity, consistency, and confidence when making important medical decisions in a complex condition like Kabuki syndrome,”* said Dr. Banka, Clinical Director of the MRCC and Rare Conditions Co-Theme Lead at the NIHR Manchester Biomedical Research Centre. *“This project is a crucial step toward improving patient care, enabling healthcare providers to make informed decisions based on expert consensus and the latest research.”*

*“For families, having an official clinical reference will be transformational,”* said [Janet Lee](#), Executive Director of the Kabuki Syndrome Foundation. *“These guidelines will empower parents to advocate for appropriate care and help clinicians deliver more consistent, informed support—no matter where a child is diagnosed.”*

*“As a parent, it’s incredibly moving to see experts and families from around the world uniting with a shared goal: to improve care for people with Kabuki syndrome,”* said one mother who will be attending the meeting. *“The inclusion of family advocates ensures our voices are heard and our experiences are reflected in the guidelines. This kind of collaboration gives me hope—for my child’s future and for every family facing this rare diagnosis.”*

The [Kabuki Syndrome Foundation](#) is grateful for the support of its community, partners, and medical collaborators around the world. This initiative represents a critical step toward improving care, raising awareness, and accelerating treatment development for individuals with Kabuki syndrome.

To learn more about Kabuki syndrome or the work of the Kabuki Syndrome Foundation, visit [www.kabukisynfound.org](http://www.kabukisynfound.org).



## Media Contact

Kabuki Syndrome Foundation

\*\*\*\*\*@kabukisynndrome.foundation.org

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