



**2022 KABUKI SYNDROME RESEARCH CONFERENCE - FRIDAY, NOVEMBER 4  
PROGRAM DETAILS**

<b>10 AM EST</b>	<p><b>Welcome &amp; Opening Remarks</b></p> <p><b>Welcome - KSF Strategy and Event Details</b>  <i>Learn about KSF efforts and how we can work as a community to drive a therapeutic roadmap and support clinical trials</i>  <b>Janet Lee</b> Executive Director, Kabuki Syndrome Foundation</p>
<b>10:10 AM EST</b>	<p><b>Understanding the Epigenetic Mechanisms of Kabuki syndrome &amp; How this shapes the Research Landscape</b>  <i>Learn why the chromatin imbalance of Kabuki syndrome is key to understanding its symptoms and treatment.  How can Kabuki syndrome be a treatable cause of intellectual disability and what are some of the pathways being explored?  How does a State of the Science publication enable new research and treatment pipelines for all types of Kabuki syndrome?</i>  <b>Moderated by: Amanda Gamboa, Outreach Coordinator - Kabuki Syndrome Foundation</b></p> <p><b>Therapeutic Development for Kabuki Syndrome Using Mouse Models</b>  <i>What is the epigenetic mechanism of Kabuki syndrome? What are the affected systems and symptoms?  Why is it important for the patient community have a foundational understanding of Kabuki syndrome?</i>  <b>Dr. Hans Bjornsson</b> Associate Professor, Genetics and Pediatrics, Johns Hopkins University; Clinical Director, Landspítali University Hospital</p> <p><b>State of the Science: A Major Stepping Stone to Building Bridges in Kabuki Syndrome Research</b>  <i>Why do we need to understand the research landscape?  What is a rare disease State of the Science and how does it help the rare disease groups drive research?  How is KSF and the KS community driving this effort?</i>  <b>Dr. Brittany Simpson</b> Assistant Professor at Cincinnati Children’s Hospital Medical Center (CCHMC); Director of Epigenetic Syndromes Clinic  <b>Dr. Corinne Stobaugh</b> KSF volunteer, parent to a KS child, Pathologist - Veteran's Hospital Tampa</p> <p><b>Q &amp; A with Panel</b></p>
<b>10:55 AM ET</b>	<p><b>Addressing the Current Challenges in Kabuki Syndrome: An Overview of Research Initiatives at Key Institutions</b></p> <p><b>Introduction To A Series of 5-minute "Flash Talks" By Leading Experts At Key Institutions</b>  <b>Moderated by: Corinne Stobaugh, Research Advisor - Kabuki Syndrome Foundation</b></p> <p><b>An Overview of Research Initiatives at Boston Children's Hospital</b>  <b>Dr. Olaf Bodamer</b> Associate Professor Boston Children's Hospital Division of Genetics and Genomics</p> <p><b>An Overview of Research Initiatives at Kennedy Krieger Institute</b>  <b>Dr. Jacqueline Harris</b> Assistant Professor, Neurology, Pediatrics, and Genetics, Kennedy Krieger Institute, Johns Hopkins Medical Institution</p> <p><b>An Overview of Research Initiatives at Louma G Foundation Lab -John's Hopkins University School of Medicine; University of Iceland</b>  <b>Dr. Hans Bjornsson</b> Associate Professor, Johns Hopkins University; Professor Translational Medicine and Pediatrics, University of Iceland - Háskóli Íslands; Clinical Director, Landspítali University Hospital</p> <p><b>An Overview of Research Initiatives at Manchester Centre for Genomic Medicine</b>  <b>Prof Siddharth Banka</b> Professor of Genomic Medicine and Rare Diseases, University of Manchester; Consultant Clinical Geneticist, Manchester Centre for Genomic Medicine</p> <p><b>Fireside Chat with Research Panel</b>  <b>Moderated by: Dr. Brittany Simpson</b> Assistant Professor at Cincinnati Children’s Hospital Medical Center (CCHMC); Director of Epigenetic Syndromes Clinic</p>
<b>11:35 AM EST</b>	<p><b>Introduction to Deep Dive Breakout Sessions</b>  <i>We will offer 3 different breakout sessions that provide an in-depth look at research efforts that are important to patient families</i>  <b>Moderated by: Annie Dean, Founder &amp; Co-President - Kabuki Syndrome Foundation</b></p>
<b>11:40 AM EST</b>	<p><b>Deeper Dives - Breakout Sessions with Researchers to Further Share Initiatives and Offer Q&amp;A</b></p>

### Track 1: Understanding Muscle Hypotonia in Kabuki Syndrome

Low muscle tone is a key feature of Kabuki syndrome, but what causes it? Learn how changes in muscle cells cause symptoms and may be treatable with Dr. Gussoni from The Roya Kabuki Clinic.

**Dr. Emanuela Gussoni** Associate Professor Boston Children's Hospital Division of Genetics and Genomics  
**Rachel Gottlieb** Program Coordinator, Roya Kabuki Program

### Track 2: Developing Evidence-based Clinical Management Guidelines for Kabuki Syndrome

Is there a long-term plan of care for people with Kabuki syndrome? Beyond recommended screenings, what are the recommended treatments for symptoms? Dr. Banka of Manchester University will share an update about the development of these very specific guidelines.

**Dr. Siddharth Banka** Professor of Genomic Medicine and Rare Diseases, University of Manchester; Consultant Clinical Geneticist, Manchester Centre for Genomic Medicine

### Track 3: Profile of Immune Cells Within Individuals with Kabuki Syndrome Type 1

Why is the immune system impacted in Kabuki syndrome? What are researchers uncovering as the true cause and how does that create a path to future treatments? Join researchers from the Cincinnati Children's Hospital to learn more.

**Dr. Sarah Potter** PhD, Research Associate, Division of Allergy and Immunology; Cincinnati Children's Hospital Medical Center  
**Dr. Brittany Simpson** Assistant Professor at Cincinnati Children's Hospital Medical Center (CCHMC); Director of Epigenetic Syndromes Clinic

12:05 PM EST Mid-day Break

12:30 PM EST Preparing for Clinical Trials - Community Participation and Buy-in

#### The Power of Patient Data in Driving Research

Community participation is key to finding treatments, and your actions are already driving more research. Now, what do we need to know about the clinical trial process? How do clinical trials benefit the entire community, and how can we continue to open doors to treat the variety of symptoms of Kabuki syndrome?

**Moderated by: Jill Orum Dunbar, Founder & Co-President - Kabuki Syndrome Foundation**

#### Why Patient-Centered Research is Important for Clinical Trial Design

How patient communities can support ongoing efforts as we look at future treatment paths.

**Dr. Adam Hartman** Program Director, Division of Clinical Research, National Institute of Neurological Disorders & Stroke

#### Q & A

#### Biobanking in Kabuki Syndrome Research

Patients as valuable partners in sample collection and research

**Dr. Giuseppe Merla** Director of Fondazione Telethon-Genomic and Genetics Disorders Biobank (GGNB); Professor of Molecular Biology, University of Naples Federico II  
**Matteo Redenti** Patient Advocate: Italy

#### Participating in Kabuki Syndrome Research- A Patient's Perspective

Shared lessons from two Kabuki syndrome advocates and ambassadors of research

**Kristin Anzenc** Adult diagnosed with Kabuki syndrome  
**Mathew Horner** Adult diagnosed with Kabuki syndrome

1:25 PM EST Q & A with Panel

1:35 PM EST Building The Clinical Trial Pipeline to Treat The Neurological Symptoms of Kabuki Syndrome

#### Session Intro - Neurobehavioral Challenges in Kabuki Syndrome

How can we best identify and measure neurobehavioral symptoms? How can a drug already treating other diseases potentially improve symptoms of Kabuki syndrome, and how do we know if it's working in a clinical trial?

**Moderated by: Leila Zegna, Vice President - Kabuki Syndrome Foundation**

#### Oryzon Genomics - An Overview of Epigenetic Therapies

**Dr. Douglas Fallor** Global CMO, Oryzon Genomics SA

#### Targeting Molecular Mechanisms of Kabuki Syndrome to Treat Symptoms

**Dr. Jacqueline Harris** Assistant Professor, Neurology, Pediatrics, and Genetics, Kennedy Krieger Institute, Johns Hopkins Medical Institution

**Update on HOPE - A Double-blind, Randomized, Placebo-controlled, Multicenter, Phase I/II Trial to Evaluate the Efficacy and Safety of Vafidemstat in Subjects With Kabuki syndrome**

**Michael Ropacki** CMO for CNS, Oryzon Genomics SA

**Identifying Biomarkers and Designing Outcome Measures for Clinical Trials in Kabuki Syndrome**

**Allison Kalinousky** Human Genetics PhD student, Johns Hopkins University

**Challenges and opportunities for speech and language testing in clinical trials for neurodevelopmental conditions**

**Adam Vogel** Director of Pediatric / Neurodevelopment Communication. Redenlab

**2:30 PM EST Q & A with Panel**

**2:55PM EST Break**

**3:00 PM EST Therapeutic Approaches and Tools That Families Can Explore NOW**

*Moderator by: **Janet Lee, Executive Director - Kabuki Syndrome Foundation***

**Neuropsychological Evaluations: Clinical and Research Applications for Kabuki Syndrome**

*Learn how neuropsychological evaluations can drive research and provide supports for school aged children*

**Dr Rowena Ng** Neuropsychologist, Department of Neuropsychology

**Q&A**

**Exploring Resources That Help Families Advocate for Children with Disabilities in a School Setting**

**Daniel Levinson** PACER board member & Father

**Self Awareness and Self Advocacy to Enable Successful School Experiences**

**VIDEO by Alex Babarovic, 17 years old**

**Explore Educational Setting and Supports - A Parent's Perspective**

*Featuring parents of children with Kabuki syndrome who will share their perspectives on providing support in a variety of different school settings*

**Moderated by:** Dana Levinson Parent - school setting: public w/ support  
**Daniel Levinson**

Tina Babarovic Parent - school setting: public w/ support

Tiffany Lay Parent - school setting: private SN school

Jodie Esponda Parent - school setting: homeschool

Bonnie Bogdanoff Parent - school setting: college

**4:15 PM EST Celebrating Achievements of Individuals with Kabuki Syndrome**

*Introduced by: **Annie Dean, Founder & Co-President - Kabuki Syndrome Foundation***

**Panel Discussion with Adults Diagnosed With Kabuki Syndrome - How They've Found Success Through Self Advocacy and Determination**

**Moderator: Michelle Westmaas** **Panel Discussion with Adults Diagnosed With Kabuki Syndrome - How They've Found Success Through Self Advocacy and Determination**

Anna Smedley - USA

Karen Denton - Canada

Kevin Brassil - USA

Rothany Julien - USA

Allie Brown - USA

James Hamilton - Australia

**5:00 PM EST Closing Remarks**

**5:05 PM EST Happy Hour & Interactive Break-out Rooms for Meet & Greet**

Room 1: Meet & Greet for those wishing to connect with adults diagnosed with Kabuki syndrome

Room 2: Meet the Principal Investigator for the upcoming Oryzon clinical trial and ask questions!

**5:30 PM EST EVENT ENDS**