



2021 KABUKI SYNDROME FOUNDATION RESEARCH CONFERENCE

OCT 22-23, 2021
10AM - 4PM EST

A VIRTUAL EVENT TO EDUCATE THE KABUKI SYNDROME
COMMUNITY ON GLOBAL RESEARCH EFFORTS

FROM RESEARCH TO REALITY



Multiple research projects focused on Kabuki syndrome are reaching critical junctures in their progress. There is consensus that within the next five years there could be a drug therapy that targets the effects of the Kabuki syndrome gene mutation. A breakthrough of this magnitude would be life changing for those affected by Kabuki syndrome.

Funding for rare disease research is an unfortunate barrier to accelerating a treatment. But, with your help, we can enable research that will produce therapeutic treatments.

Funds raised by KSF will help drive advances in new areas of research, support the creation of a collaborative research network, and provide important educational materials to the Kabuki syndrome community to support research recruitment.

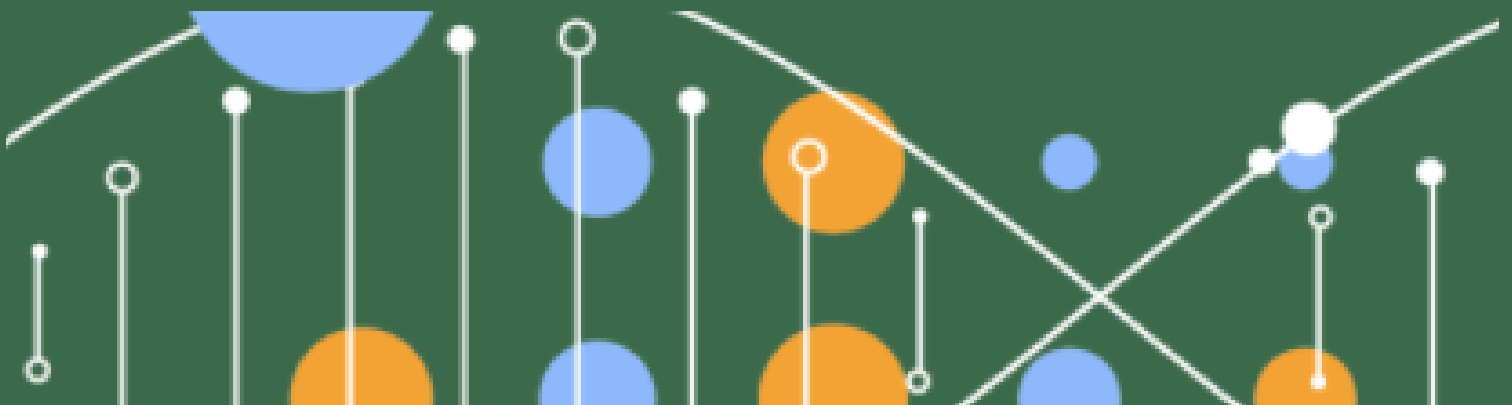


COLLABORATION AS A DRIVER OF CHANGE

A decorative graphic in the top right corner featuring four green plus signs arranged in a 2x2 grid, and a molecular structure with a central white sphere, a blue sphere, and a grey sphere, connected by lines.

Kabuki Syndrome Foundation is the only organization dedicated to accelerating research efforts that show promise to treat, prevent, or cure Kabuki Syndrome. We have built a coalition with major research centers globally that are influencing the direction of future research efforts. A strong collaboration with researchers and clinicians is critical in focusing on what matter most to patients, families, and advocates.

Patient participation in research is a powerful tool in accelerating Kabuki syndrome research. For any rare disease, the only way to make progress is to come together as a community and unite ourselves in the path to progress.



VIRTUAL RESEARCH CONFERENCE PROGRAM

10/22/2021 - 10/23/2021

10AM-4PM EST

REGISTRATION REQUIRED
REGISTER [HERE](#)



EMILY, AGE 12, PARTICIPATES IN A RESEARCH STUDY
AT KENNEDY KRIEGER INSTITUTE

PROGRAM 10/22



10:00AM

WELCOME & OPENING REMARKS FROM KSF

10:10 AM

**HOW RARE DISEASE RESEARCH HAS EVOLVED:
UNDERSTANDING THE CHANGING LANDSCAPE
OF KABUKI SYNDROME RESEARCH**



DR HANS BJORNSSON, JOHNS HOPKINS UNIV.
KABUKI SYNDROME: INSIGHTS FROM THE FIRST
10 YEARS SINCE DISCOVERY OF THE GENE.



DR. HADIA HIJAZI, JOHNS HOPKINS UNIV.
POSTNATAL EFFECTS OF KMT2D DISTURBANCE:
IMPLICATIONS FOR THERAPEUTIC
DEVELOPMENT



DR. LEANDROS BOUKAS, JOHNS HOPKINS UNIV.
INSIGHTS FROM COMPARING KABUKI
SYNDROME TO OTHER RELATED DISORDERS TO
UNCOVER THE MECHANISTIC BASIS.

LIVE Q&A TO FOLLOW PRESENTATIONS

11:20 AM

**DEFINING THE ROLE OF THE PATIENT IN
DISCOVERY AND BASIC RESEARCH**



**DR BRITTANY SIMPSON, CINCINNATI
CHILDREN'S HOSPITAL MEDICAL CENTER**
SHARING STORIES, IMPROVING SCIENCE: HOW
THE PATIENT EXPERIENCE SHAPES RESEARCH
ENDEAVORS



**DR. ANGIE SERRANO, CENTER FOR
REGENERATIVE MEDICINE, BOSTON
UNIVERSITY PATIENT-DRIVEN RESEARCH:
BRINGING BASIC SCIENCE, CLINICIANS AND
PATIENTS' FAMILIES CLOSER TOGETHER**

LIVE Q&A TO FOLLOW PRESENTATIONS

****PRESENTATION TIMES ARE SUBJECT TO CHANGE**

PROGRAM 10/22



12:00 PM

KSF CROWDSOURCING INITIATIVE TO GAUGE THE COMMUNITY'S MOST PRESSING NEEDS (LIVE)

12:10 PM

CROWDSOURCING SURVEY & BREAK

12:50 PM

ADDRESSING THE CURRENT CHALLENGES IN KABUKI SYNDROME: HOW RESEARCH EFFORTS AT KENNEDY KRIEGER INSTITUTE ARE FOCUSING ON MOVING THE FIELD CLOSER TO CLINICAL TRIALS DESIGNED TO TREAT THE NEURODEVELOPMENTAL ASPECTS OF KABUKI SYNDROME

**TIFFANY MATTHEWS LAY, MOM TO NASIR,
A PATIENT PERSPECTIVE ON PARTICIPATING IN RESEARCH**



**DR. JACQUELINE HARRIS, KENNEDY KRIEGER INSTITUTE, JOHNS HOPKINS UNIVERSITY
NEURODEVELOPMENT IN KABUKI SYNDROME: FROM CLINIC TO RESEARCH AND BACK AGAIN**



**ALLISON KALINOUSKY, PHD STUDENT, JOHNS HOPKINS UNIVERSITY
ONGOING CLINICAL AND TRANSLATIONAL RESEARCH PROJECTS IN KS AND WHY THEY MATTER SO MUCH**

LIVE Q&A TO FOLLOW PRESENTATIONS

1:55 PM

BEYOND THE MOUSE MODEL: AN INTRODUCTION TO DIFFERENT MODEL SYSTEMS IN KABUKI SYNDROME RESEARCH



**DR. MAX HEIMAN, HARVARD MEDICAL SCHOOL AND BOSTON CHILDREN'S HOSPITAL
FAST, CHEAP, AND POWERFUL: A WHOLE-ANIMAL MODEL OF KABUKI SYNDROME IN C. ELEGANS**



**DR. ALESSIO ZIPPO, UNIV. OF TRENTO, ITALY
FISHING OUT THE ROLE OF MECHANOBIOLOGY IN KABUKI SYNDROME: FROM IN VITRO TO IN VIVO MODEL SYSTEMS**



**DR. ANDREW KODANI, CENTER FOR PEDIATRIC NEUROLOGICAL DISEASE RESEARCH, ST. JUDE CHILDREN'S HOSPITAL
SYSTEMS APPROACH TO UNCOVERING THE PATHOGENESIS OF KABUKI SYNDROME**

LIVE Q&A TO FOLLOW PRESENTATIONS

PROGRAM 10/22



2:55 PM

CELEBRATING THE ROLE OF PATIENT ADVOCACY GROUPS IN RARE DISEASE

A PANEL OF GLOBAL PATIENT ADVOCATE LEADERS INCLUDING: (LISTED ALPHABETICALLY BY LAST NAME)

EMILY BENTON - TEXAS KABUKI GATHERING

AMANDA GAMBOA - PACIFIC NORTHWEST KABUKI

LISA INGRAM - KABUKI KARNIVAL

ALEXANDRA KENTER - KS NETWORK-NETHERLANDS

RENE KING - ALL THINGS KABUKI

PILAR LUCERO - KABUKI SPANISH ASSOCIATION

SALLY TREWARTHA - KABUKI UK

LIVE Q&A TO FOLLOW PRESENTATIONS

3:45 PM

CELEBRATING ACHIEVEMENTS OF INDIVIDUALS WITH KABUKI SYNDROME

FEATURING A VIDEO BY JASON REYNOLDS, DAD TO JONNY AND INVITATION TO JOIN THE KSF WALL OF ACCOMPLISHMENTS

3:55 PM

CLOSING REMARKS AND CALL TO ACTION

4:00 PM

END OF DAY 1

DAY 2 PROGRAM DETAILS ON THE NEXT PAGE

****PRESENTATION TIMES ARE SUBJECT TO CHANGE**

PROGRAM 10/23



10:00 AM

WELCOME AND OPENING REMARKS

10:10 AM

THE ROYA KABUKI INITIATIVE: A PATIENT CENTERED APPROACH TO INVESTIGATING THE UNDERLYING MECHANISMS OF KABUKI SYNDROME AND ADVANCING THERAPEUTIC DEVELOPMENT

LEILA AND EDOARDO ZEGNA, KSF & ROYA KABUKI CLINIC



DR. OLAF BODAMER, ROYA KABUKI INITIATIVE, BOSTON CHILDREN'S HOSPITAL
STRATEGIES OF DRUG DEVELOPMENT IN KS



DR. EMANUELA GUSSONI, ROYA KABUKI INITIATIVE, BOSTON CHILDREN'S HOSPITAL
MUSCLE STEM CELL STUDIES IN KABUKI SYNDROME



DR. BEN GOODLETT, ROYA KABUKI INITIATIVE, BOSTON CHILDREN'S HOSPITAL
NEURODEVELOPMENTAL ASSESSMENT IN KS FOR CLINICAL TRIAL READINESS



TARA DALY, PROGRAM MGR, ROYA KABUKI INITIATIVE, BOSTON CHILDREN'S HOSPITAL
OPPORTUNITIES FOR RESEARCH PARTICIPATION

LIVE Q&A TO FOLLOW PRESENTATIONS

11:20 AM

DRIVING THE RESEARCH AGENDA TO FIND A THERAPEUTIC TREATMENT

KSF CROWDSOURCING SURVEY RESULTS AND IDENTIFYING POTENTIAL AREAS OF RESEARCH



ANGELA JOCSO, ALLSTRIPES
HOW YOUR MEDICAL RECORDS CAN POWER RESEARCH!



CAITLIN NICHOLS, ALLSTRIPES
HOW YOUR MEDICAL RECORDS CAN POWER RESEARCH!

LIVE Q&A TO FOLLOW PRESENTATIONS

12:15 PM

BREAK

PROGRAM 10/23



12:55 PM

IMPROVING DIAGNOSIS IN KABUKI SYNDROME



DR SIDDHARTH BANKA UNIVERSITY OF MANCHESTER; MANCHESTER CENTRE FOR GENOMIC MEDICINE
RECENT AND ONGOING RESEARCH AROUND IMPROVING DIAGNOSIS AND MANAGEMENT IN KABUKI SYNDROME



LUKAS LANGE, PROBABLYGENETIC
INCREASING ACCESS TO EARLY DIAGNOSTICS USING MACHINE LEARNING, TELEMEDICINE, AND SOCIAL MEDIA.

LIVE Q&A TO FOLLOW PRESENTATIONS

1:45 PM

PRECLINICAL STUDIES ON LONG-TERM TREATMENTS FOR COGNITIVE FUNCTIONS IN A KABUKI SYNDROME MOUSE MODEL



JENNIFER MANTEY GOLDSMITH, JAMES AND SANDRA MANTEY FAMILY FOUNDATION

DR. KASTURI HALDAR, BOLER-PARSEGHIAN CENTER FOR RARE AND NEGLECTED DISEASES; UNIVERSITY OF NOTRE DAME

2:10 PM

RESEARCHER ROUNDTABLE AND LIVE Q&A

DR. HANS BJORNSSON, DR. JACQUELINE HARRIS, DR. OLAF BODAMER, DR. ALESSIO ZIPPO, DR. SID BANKA, DR. KASTURI HALDAR

2:40 PM

POTENTIAL OF A SMALL MOLECULE DRUG FOR USE IN KABUKI SYNDROME



DR. CARLOS BUESA, CEO ORYZON
VAFIDEMSTAT, A POSSIBLE THERAPEUTIC OPTION IN KABUKI SYNDROME



DR. MICHAEL T. ROPACKI, CNS CHIEF MEDICAL OFFICER
VAFIDEMSTAT, A POSSIBLE THERAPEUTIC OPTION IN KABUKI SYNDROME



DR. JACQUELINE HARRIS, KENNEDY KRIEGER INSTITUTE, JOHNS HOPKINS UNIV.
DESIGNING AN EFFECTIVE CLINICAL TRIAL IN KABUKI SYNDROME

LIVE Q&A TO FOLLOW PRESENTATIONS

PROGRAM 10/23



3:25 PM

CELEBRATING ACHIEVEMENTS OF ADULTS WITH KABUKI SYNDROME

FACILITATED BY MICHELE WESTMAAS, MOM TO AUBRIE

INSPIRATIONAL VIDEO MONTAGE, LIVE PANEL DISCUSSION, AND Q&A WITH THESE AMAZING ADULTS WHO WILL SHARE THEIR JOURNEY WITH THE COMMUNITY. PANELISTS INCLUDE:

JENNY ANDERSON - 29 YRS OLD, UK

KRISTIN ANZELC - 36 YRS OLD, USA

ELISE HARREN - 26 YRS OLD, USA

SHANNON LOFTUS - 28 YRS OLD, USA

AARON SENIOR - 31 YRS OLD, UK

AUBRIE WESTMAAS - 23 YRS OLD, USA

4:00 PM

CLOSING REMARKS AND CALL TO ACTION

THANK YOU TO ALL OF OUR PRESENTERS, VOLUNTEERS, SPONSORS, AND PARTNERS WHO HELPED MAKE THIS EVENT POSSIBLE!

SPECIAL THANK YOU TO OUR SPONSORS

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OF COLUMBUS**

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LISA INGRAM



Thank You

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