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

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.

Kabuki SYNDROME FOUNDATION

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

Kabuki SYNDROME FOUNDATION

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

Kabuki SYNDROME FOUNDATION

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

Kabuki SYNDROME FOUNDATION

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 CHIDLREN'S HOSPITAL
NEURODEVELOPMENTAL ASSESSMENT IN KS FOR CLINICAL TRIAL READINESS

TARA DALY, PROGRAM MGR, ROYA KABUKI INITIATIVE, BOSTON CHIDLREN'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 CROWDSOUCING SURVEY RESULTS AND IDENTIFYING POTENTIAL AREAS OF RESEARCH

ANGELA JOCSON, 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

Kabuki SYNDROME FOUNDATION

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 INSTITUE, JOHNS HOPKINS UNIV.
DESIGNING AN EFFECTIVE CLINICAL TRIAL IN KABUKI SYNDROME

LIVE Q&A TO FOLLOW PRESENTATIONS

PROGRAM 10/23

Kabuki SYNDROME FOUNDATION

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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Thank You

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