

# KSF Scientific Symposium Summary

October 26, 2023



The 2023 KSF Scientific Symposium took place virtually on October 26, 2023, bringing together over 60 clinicians, researchers, and experts from around the globe. The Symposium presentations collectively highlighted the complex molecular and genetic mechanisms underlying Kabuki syndrome, its diverse clinical manifestations, and potential pathways for targeted treatments. The KSF Scientific Symposium not only provided a platform for sharing the latest research on Kabuki syndrome but also fostered collaboration among experts in the field. Feedback from Symposium participants has been overwhelmingly positive. They found the scientific content to be outstanding; researchers forged new connections and collaborations that will manifest in the sharing of protocols, reagents, and models relevant to their respective studies. By collaborating and building on these research findings, the community is poised to make significant strides in the diagnosis, treatment, and overall care of individuals with Kabuki syndrome.

To recap, the meeting featured opening and closing remarks by leading Kabuki syndrome researcher, **Dr Hans Bjornsson** from Johns Hopkins University / University of Iceland, as well as fifteen presentations on four themes.

The first two sessions were chaired by **Dr Siddharth Banka** from the University of Manchester and **Dr Jacqui Harris** from the Kennedy Krieger Institute / Johns Hopkins University. Italian and American investigators shared their latest insights and discoveries on various Kabuki syndrome phenotypes, including:

- mechanisms underlying immune dysfunction in Kabuki syndrome Type 1 (**Dr Sarah J. Potter**, Research Associate at Cincinnati Children's Hospital Medical Center and **Dr Veronica De Rosa**, Senior Researcher at Institute for Experimental Endocrinology and Oncology "G. Salvatore" - National Research Council of Italy (IEOS-CNR)),
- the roles of *KMT2D* and *KDM6A* in regulating brain development (**Dr Angie Serrano**, Assistant Professor at Boston University and **Dr Jamy C Peng**, Associate Member at St. Jude Children's Research Hospital),
- cardiovascular effects of different *KMT2D* pathogenic variants (**Dr H. Joseph Yost**, Vice Chair for Basic Science Research, Department of Pediatrics, University of Utah),
- novel palate and craniofacial defects observed in a new mouse model of Kabuki syndrome (**Dr Hyuk-Jae Edward Kwon**, Assistant Professor at State University of New York at Buffalo),

- sleep disturbance in individuals with Kabuki syndrome (**Dr Tyler Rapp**, Pediatric Resident Physician at Children's National Hospital).

The next session was chaired by **Dr Margaret Adam** from the University of Washington School of Medicine, Seattle Children's Hospital. It included presentations from researchers based in the US and Canada covering the genetics and epigenetics of Kabuki syndrome:

- *KMT2D*'s role in gene regulation and its effects on tissue development and tumor suppression (**Dr Kai Ge**, Senior Investigator at the National Institute of Health (NIH))
- the effects of *KMT2D* variants on gene activity and the identification of unique signatures with potential applications for monitoring therapeutic efficacy (**Dr Youngsook Lucy Jung**, Instructor at Boston Children's Hospital),
- changes in brain activity caused by Kabuki syndrome mutations (**Dr Christopher Patzke**, Assistant Professor at University of Notre Dame),
- using DNA methylation profiling to identify and stratify Kabuki syndrome cases for clinical trials (**Dr Sanaa Choufani**, Senior Research Associate at the Hospital for Sick Children).

The last two sessions, titled therapeutic opportunities and clinical trial readiness, were chaired by the KSF Chief Science Officer, **Dr Bruce Bloom**. These sessions showcased data from American and Italian researchers on:

- two potential treatment strategies for Kabuki syndrome: DCF (**Dr Kasturi Haldar**, Professor at University of Notre Dame) and ATR inhibitors (**Dr Alessio Zippo**, Associate Professor at University of Trento),
- the development of biomarkers and outcome measures for Kabuki syndrome clinical trials (**Allison Kalinousky**, PhD Candidate at Johns Hopkins University and **Dr Jacqui Harris**, Director of the Epigenetics Clinic at Kennedy Krieger Institute and Associate Professor at the Johns Hopkins University School of Medicine).

We look forward to the many new discoveries and advances that will be revealed in 2024 and beyond. For more details on each of the presentations, please refer to the [Symposium abstract booklet](#) or contact the KSF Director of Research, Dr Clara Tang at [clara@kabukisynndrome.foundation.org](mailto:clara@kabukisynndrome.foundation.org).