UNIVERSITY OF ICELAND

LANDSPÍTALI

Do you or your child have Turner syndrome and features of Kabuki syndrome?

We are conducting a research study to better understand why some individuals with Turner syndrome also have clinical features associated with Kabuki syndrome.

Who are we looking for?

We are interested in hearing from individuals who:

- Have Turner syndrome, particularly involving a ring X chromosome or another structural X-chromosome difference
- Have been diagnosed with Kabuki syndrome or have features suggestive of Kabuki syndrome, such as:
  - Immune deficiency
  - Intellectual disability
  - Distinctive facial features
- Have not had a disease-causing variant identified in *KMT2D* or *KDM6A*

Interested in learning more?

Participant information sheet: Link

For questions or to express interest in participating in the study, please contact:

Arnhildur Tómasdóttir, CGC
University of Iceland
art17@hi.is

Hans Tómas Björnsson, MD, PhD
University of Iceland
htb@hi.is