



Understanding Sleep, Behavior, And Quality of Life in Kabuki Syndrome

What: A research study looking at a variety of factors in Kabuki Syndrome (KS) such as sleep patterns and difficulties, anxiety, behaviors, and general quality of life. The purpose of this study is to better understand important day-to-day factors that have not yet been studied well in KS. This will help guide future trials and treatments.

First, the study involves a series of questionnaires that can be completed remotely on a computer. These will ask many questions that can be completed by individuals with KS and their parents or caregivers. There may also be separate questions about siblings. You may also be asked to provide genetic testing and neuropsychological testing.

Next, a member of the study team may reach out to follow up via telephone/video conference.

Who: Any individual genetically diagnosed with Kabuki Syndrome (type 1 or type 2).

Where: The study will take place remotely. All components can be completed on a computer without the need to visit our site in Baltimore.

Why: Other genetic disorders have an increased rate of issues concerning sleep, anxiety, and quality of life compared to the general population. Many of these factors have not been fully explored in individuals with KS. Studying day-to-day behaviors and factors that shape the life of someone with KS is critical in completely understanding the condition. Further developments in these areas will help guide future trials and treatments.

When: The study is currently ongoing.

How: If interested in participating or if there are further questions, please reach out to Dr. Jacqueline Harris and/or her research coordinator Jennifer Johnson at the following emails:

Dr. Harris: harrisjac@kennedykrieger.org

Jennifer Johnson: johnsonjen@kennedykrieger.org

Phone: 667-205-4295