

You're Invited to Participate

Thank you for your interest in the GestaltMatcher Research Study and Database (GMDB)! Your participation in this study is voluntary. Before providing data/photos/further information, please make sure you have read the following study information and have given the necessary consent.

Using Computer-Aided Image Analysis to Help Diagnose Kabuki Syndrome

The diagnostic process of rare genetic disorders can be lengthy. Often, affected individuals and their families report complicated paths of diagnosis until a possible diagnosis is found. **Our research study aims to shorten this diagnostic process.**

In many genetic disorders, disease-specific features on the face give a hint of the underlying disease. A specialized doctor can recognize many more and rarer diseases by specific facial features. However, the doctor must have sufficient experience with the individual clinical patterns. But it is precisely this experience that is often lacking in the case of rare diseases.

Modern computer-aided image analysis methods can also learn to recognize such features in the face and thus reliably calculate similarities to individuals with an already confirmed diagnosis. Like a doctor, however, the computer system must first learn the characteristics of the diseases in order to recognize them. **Photos of people with a diagnosis of Kabuki syndrome can help us teach the computer system and may help improve the care of other people with Kabuki syndrome.**

Study Process

Go to bit.ly/KSF_GMForm or scan the QR code to fill out interest form:

Receive a link to consent and participate.

Upload photos from your phone or computer.



Photos Requested

We encourage uploading 5 or more photos at different ages, including before the person was diagnosed if possible. Photos should be as clear as possible and focus on the face.

Questions? Please contact us: info@gestaltmatcher.org or clara@kabukisynromefoundation.org

The database is funded by the non-profit organization Arbeitsgemeinschaft Gen-Diagnostik (AGD) e.V. The Institute for Genomic Statistics and Bioinformatics (IGSB) at the University of Bonn is responsible for conducting and managing the study and for the scientific analysis of the data.

The Kabuki Syndrome Foundation is providing outreach and recruitment support for this research study.

