Information leaflet (to be sent to interested members):

Study: Automatic Phenotype Identification of Kabuki Syndrome Patients

Dear Member of the Kabuki Syndrome Network,

As you undoubtedly know, due to its rarity, not all physicians know how to diagnose Kabuki Syndrome or are even aware of the existence of it. This has a material effect on Kabuki patients, who are often misdiagnosed or remain undiagnosed for long periods of time. This "diagnostic odyssey" can be very burdensome on patients and their families both financially and emotionally, and can lead to serious harm if the patient is deprived of appropriate medical care.

FDNA is a pioneer in the field of facial analysis, developing advanced proprietary tools that will help identify Kabuki Syndrome patients early in life by analyzing facial photos of patients and picking up subtle facial clues associated with Kabuki Syndrome.

We are contacting you through the Kabuki Syndrome Network however your participation is completely voluntary. If you choose not to take part in the research, it will not be reflected in your relation with the support organization or the researchers.

Study Goal: The goal of this study is to train and validate FDNA's system to identify Kabuki Syndrome more accurately. Once our system reaches acceptable accuracy levels, we will make this non-invasive technology available to all types of physicians. The system will be accessible through any handheld device (such as iPhones, tablets, etc.) and distributed for free exclusively to professional healthcare providers. We hope to increase awareness of rare diseases for all types of physicians and help them reach an early diagnosis of Kabuki Syndrome, as well as many other syndromes.

Participant Eligibility: If you, or a member of your family, have a confirmed diagnosis of Kabuki Syndrome, you are eligible to participate in this study. Our main focus is diagnosing children and newborns, but we accept images from all age groups.

Study Requirements: In order to participate in this study, you would be asked to submit the following information on behalf of the participant:

- Full frontal and lateral photos of participant's face (see photo instructions at: http://www.fdna.com/pictures/sample_report/FDNA_photo_instructions.pdf)
- Gender
- Age of participant (at the time the facial images were taken)
- Ethnicity (picked from a list)
- Results of confirmed diagnosis (molecular or clinical)

Participant Privacy: All participants, or their guardians, will be required to sign an informed consent, authorizing the use of the images and information for this study. Patient privacy is of the utmost importance to us and we keep all participants' information

submitted strictly confidential. This information will not be published, nor will it be used for any other purpose, without the explicit written consent of participants, or in any way that would jeopardize, publicize or reveal the participants' identity.

For further information or to apply to participate in this study, please visit www.fdna.com or contact Ms. Leslie Abuaf (leslie@fdna.com) or Mr. Dekel Gelbman (dekel@fdna.com).

Form of Patient Consent for Medical Photography

I, _____, hereby agree to submit identifiable photographs of _____(Patient's Name) to be evaluated by FDNA's image analysis technology for clinical, research, development and educational purposes.

None of the participant's personal data, such as photograph, name or date of birth will be displayed in any subsequent publication, without obtaining my explicit written consent.

I have the right to request no further usage of the photographs by contacting the following individuals:

_____

Prof. _____

Signature / Guardian's Signature	Date

Relationship (if signed by guardian)