



Clinical Survey Results

Kabuki Syndrome

Please note all information presented here is the result of a small research study meant to contribute to the existing body of information about Kabuki Syndrome.

Overview

Between **November 2017 and March 2018**, an **online survey** was conducted in order to better understand the **characteristics of Kabuki Syndrome and their impact on daily life**. Participants were recruited with the help of physicians and **All Things Kabuki**. A total of **71 participants** completed the survey.



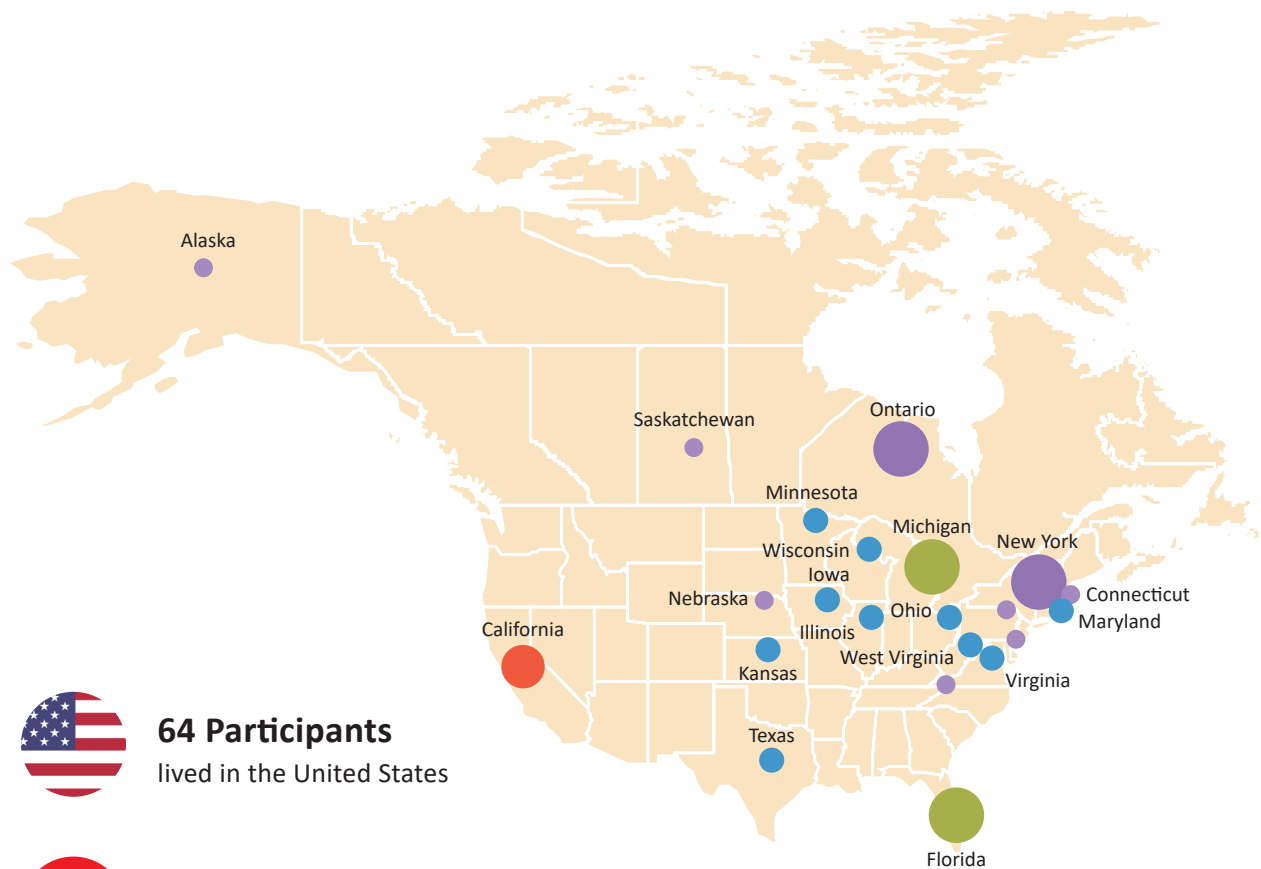
57 caregivers
of someone with Kabuki Syndrome completed a survey



11 adolescents
with Kabuki Syndrome (all associated with caregiver participants) completed a survey



3 adults
with Kabuki Syndrome completed a survey

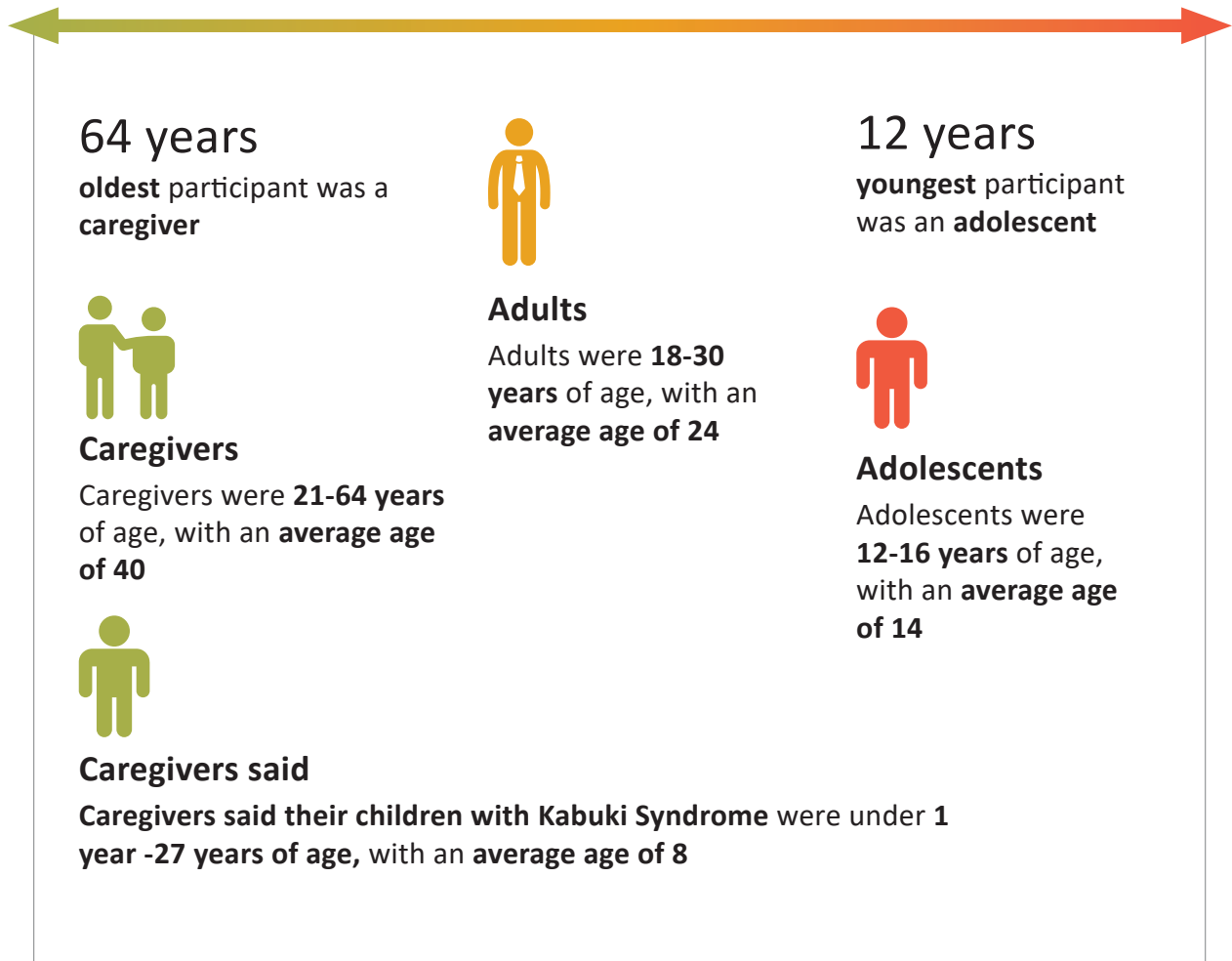


64 Participants
lived in the United States



7 Participants
lived in Canada

Snapshot of survey participant age and gender

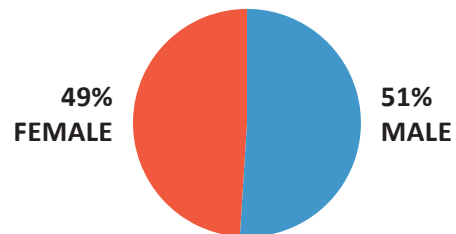


93%
of caregiver and adult participants were **female**



7%
of caregiver and adult participants were **male**

Kabuki Syndrome patients



Caregivers had **both male and female children** with Kabuki Syndrome

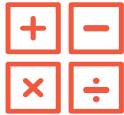
Education and Employment

Adults and caregivers reported on themselves or their child with Kabuki Syndrome:



75

average IQ score



65%

Reported difficulty with math problems



65%

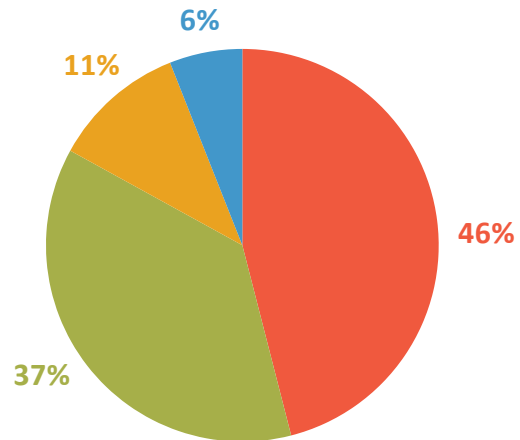
Reported difficulty reading a map



63%

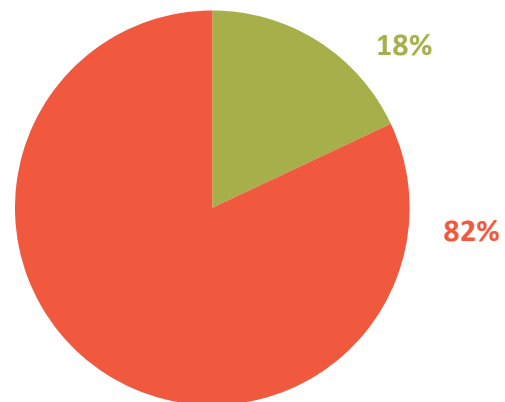
Reported difficulty completing a maze

Type of school attended by 35 individuals with Kabuki Syndrome



- Special education public school
- Traditional public school
- Special education private school
- Traditional private school

Employment status of 11 individuals 16 years and up with Kabuki Syndrome



- Has a job*
- Does not have a job

*Survey results indicated that Kabuki Syndrome patients worked at retail stores, for example, and volunteered as teacher's assistants

Characteristics of Kabuki Syndrome



3 months - 30 years
Age range of diagnosis

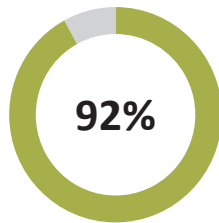


Geneticist
Most common diagnosing provider

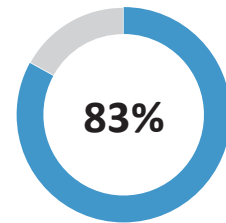
Adults and caregivers were asked to report on **characteristics of their or their child's Kabuki Syndrome** as part of the **screening process**. The most common characteristic reported was **"unique facial features."**



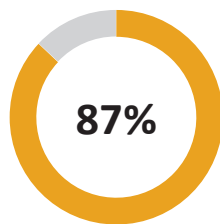
UNIQUE FACIAL
FEATURES



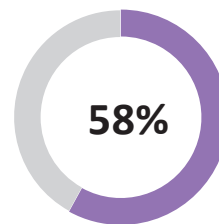
ABNORMAL
FINGERPRINTS



GROWTH
DEFICIENCIES



MILD OR MODERATE
INTELLECTUAL DISABILITY



ISSUES WITH
BODY SKELETON

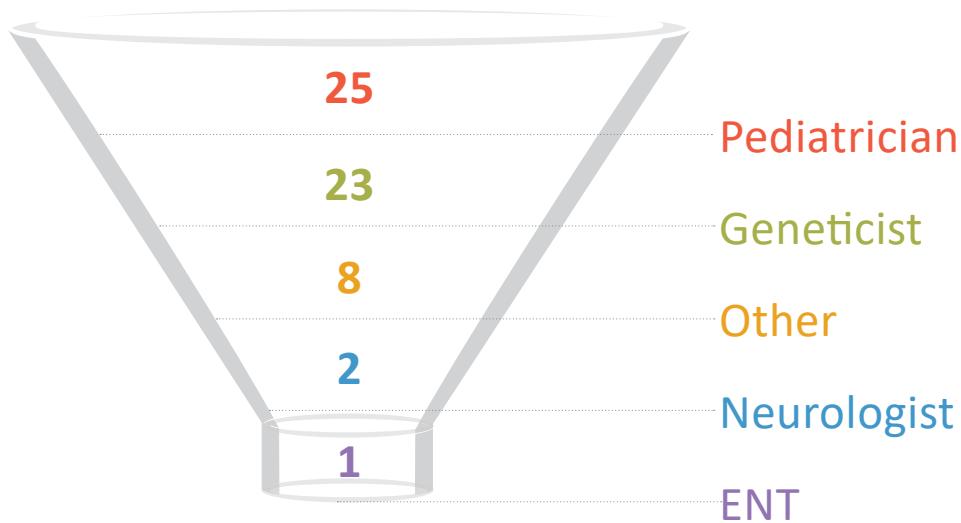


56 Adults and caregivers reported they (or their child in the case of caregivers) received **genetic testing**



KMT2D was the most commonly reported mutation

Primary treatment provider reported by caregivers and adults



Caregivers and adults most commonly said they visit their primary treatment provider on an as needed basis

Other treatment providers reported



Caregivers rated the level of difficulty with **daily tasks** for their children with Kabuki Syndrome (top). **Adolescents** also rated their level of difficulty (bars below).

Caregiver reports on Kabuki patients



42% of caregivers rated **washing hair** as extremely difficult



28% of caregivers rated **brushing hair** as extremely difficult



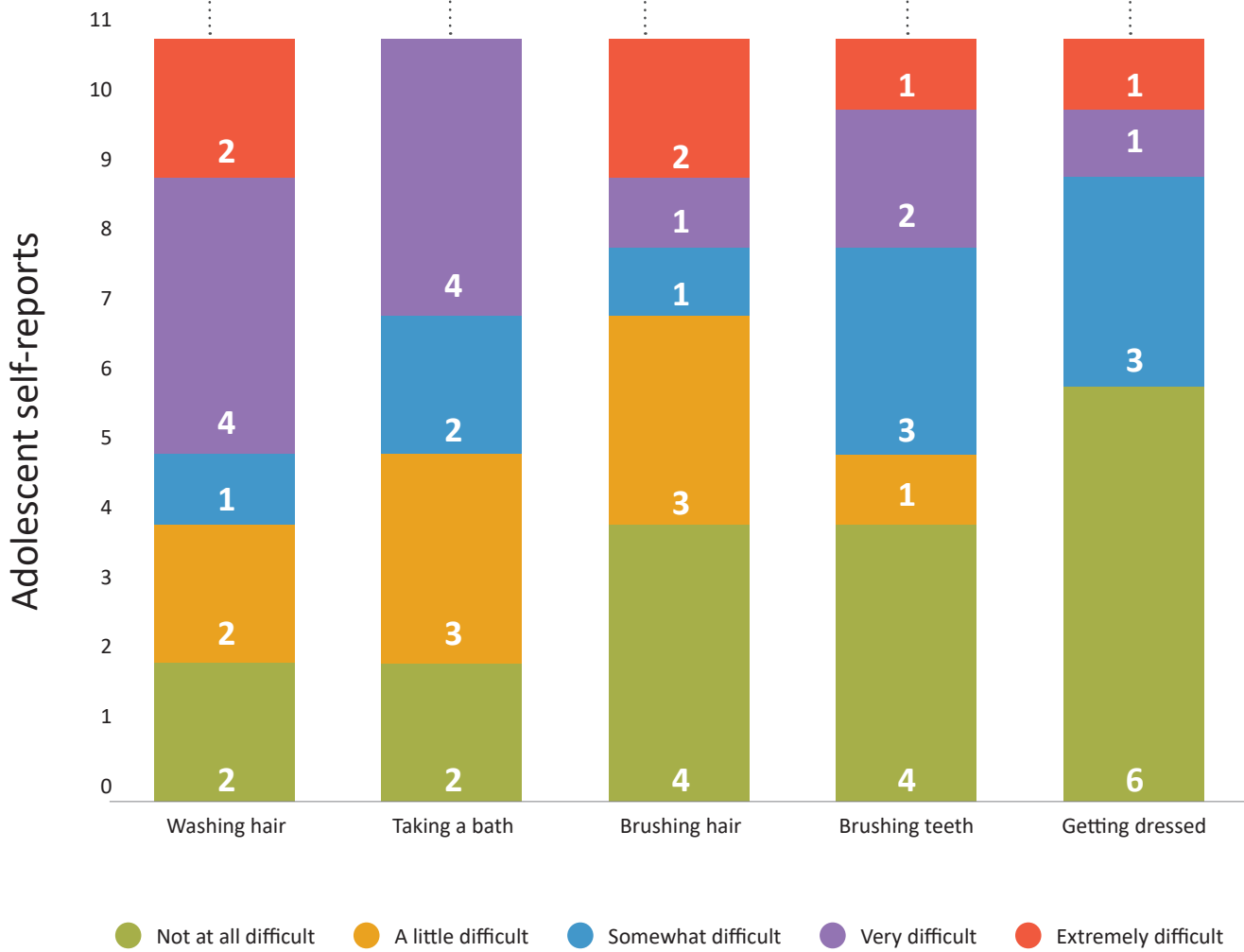
21% of caregivers rated **getting dressed** as extremely difficult



32% of caregivers rated **bathing** as extremely difficult



25% of caregivers rated **brushing teeth** as extremely difficult



Adults and caregivers reported on various issues:



75% had eye issues

Common eye issues included: glasses/contacts, eyes do not always look the same direction, whites of the eyes appear blue, and upper eyelid droops over the eye



70% had dental issues

Common dental issues included: missing teeth, unusual tooth shape, and excessive cavities



62% used a feeding tube

Almost half of participants reported difficulty eating food



47% had hearing loss

Most of those who reported hearing loss wear a hearing aid



22% reported having an **immune disorder**

10% reported having an **autoimmune disorder**

Infection history



65% experienced ear infections

48% experienced lung/respiratory infections

27% experienced urinary tract infections

25% experienced some other type of infection

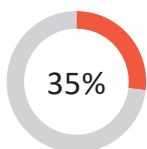


77% had a history of joint issues

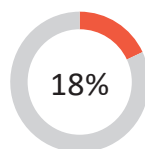
Most joint issues did not require surgery



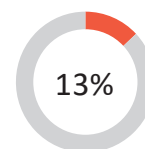
Mouth issues



Split in the roof of the mouth



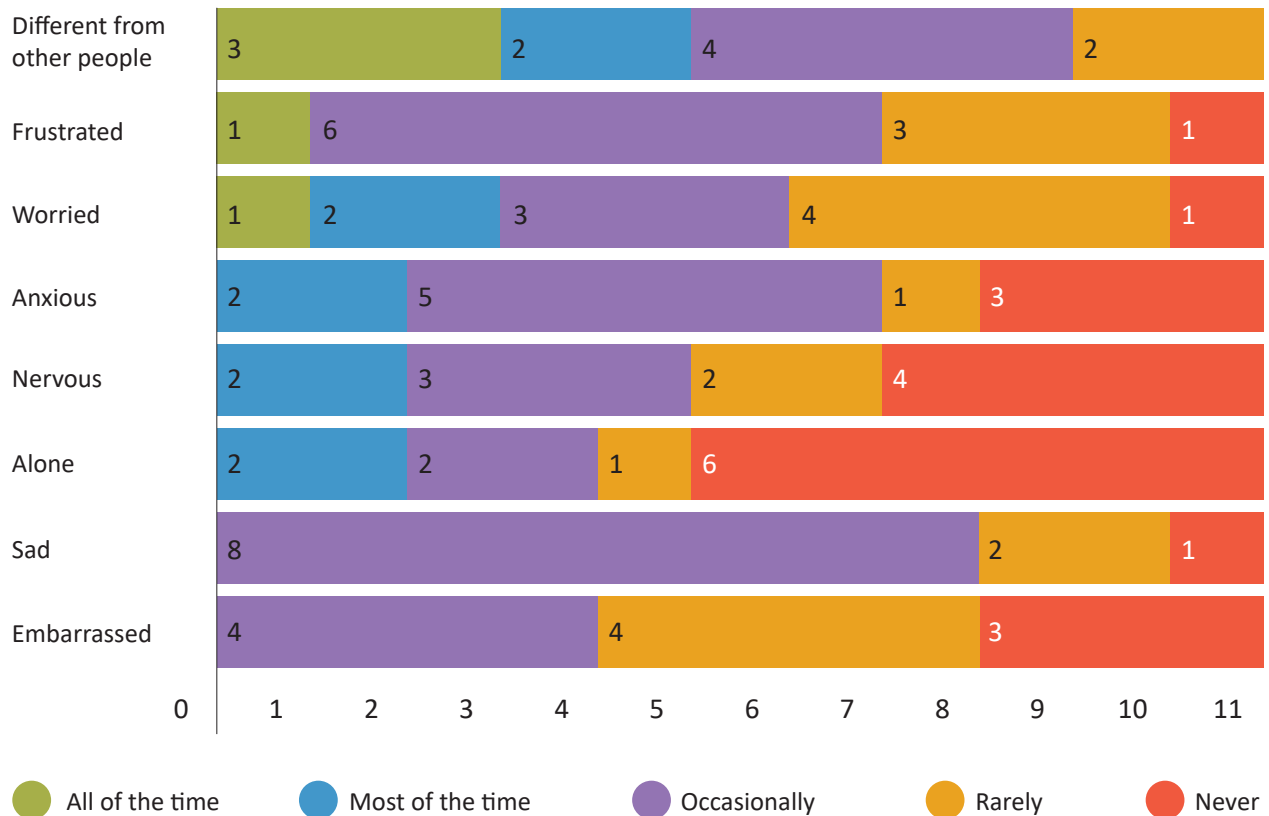
Cleft or fork in the skin that hangs down from the back part of the roof of the mouth



Not enough tissue in the roof of the mouth

11 adolescents responded to the question

“Because of my Kabuki Syndrome, I feel ... “



57 caregivers responded to a similar question about their child:



58% of caregivers said their child with Kabuki Syndrome **feels frustrated occasionally**

19% of caregivers said their child with Kabuki Syndrome **feels anxious/worried most of the time or all of the time**

28% of caregivers said their child with Kabuki Syndrome **feels isolated occasionally, most of the time, or all of time**

Adults and caregivers were asked whether they or their child with Kabuki Syndrome experiences anxiety or depression:



27% experience anxiety

Anxiety treatments included therapy, medication, and home calming strategies

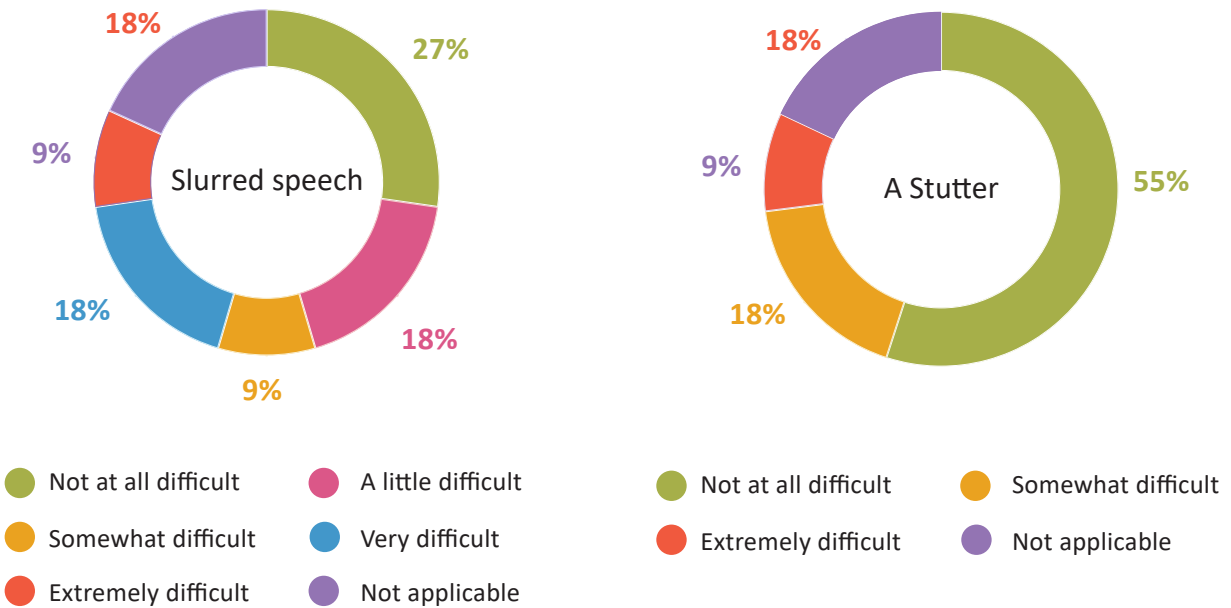


8% experience depression

Depression treatments included therapy and medication

11 adolescents responded to the question

"I have trouble talking or having conversations because of ..."



Adolescents said they experienced the following social impacts

"occasionally," "most of the time," or "all of the time."



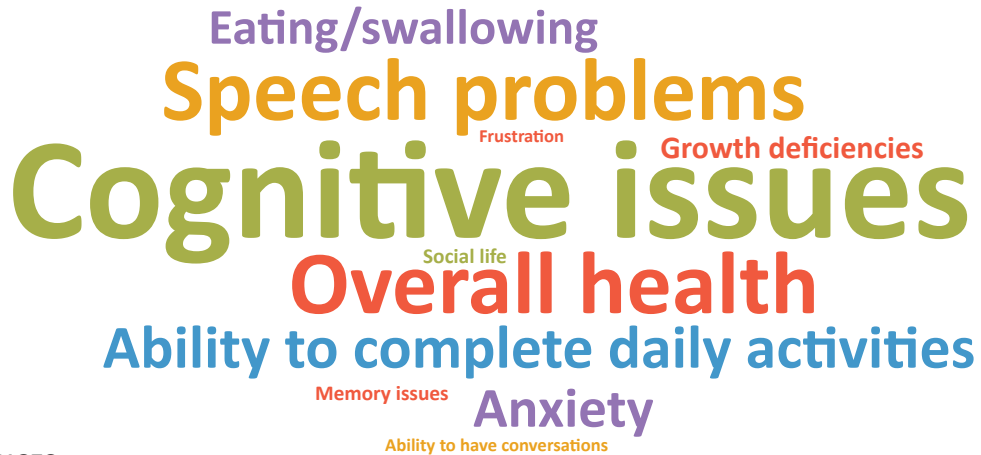
Overall, the three adults with Kabuki Syndrome said they did not experience difficulty with communication. They either *"somewhat disagreed"* or *"somewhat agreed"* to the statement: *"I find it difficult to have conversations with others."*

Adults agreed with the impact of *"having difficulty making friends"* but did not strongly agree with any of the other social impacts they were asked to rate.

Treatment goals & preferences

Adults and caregivers said they would like a **potential treatment** to improve

GOALS



PREFERENCES



68% of caregivers would prefer a liquid treatment



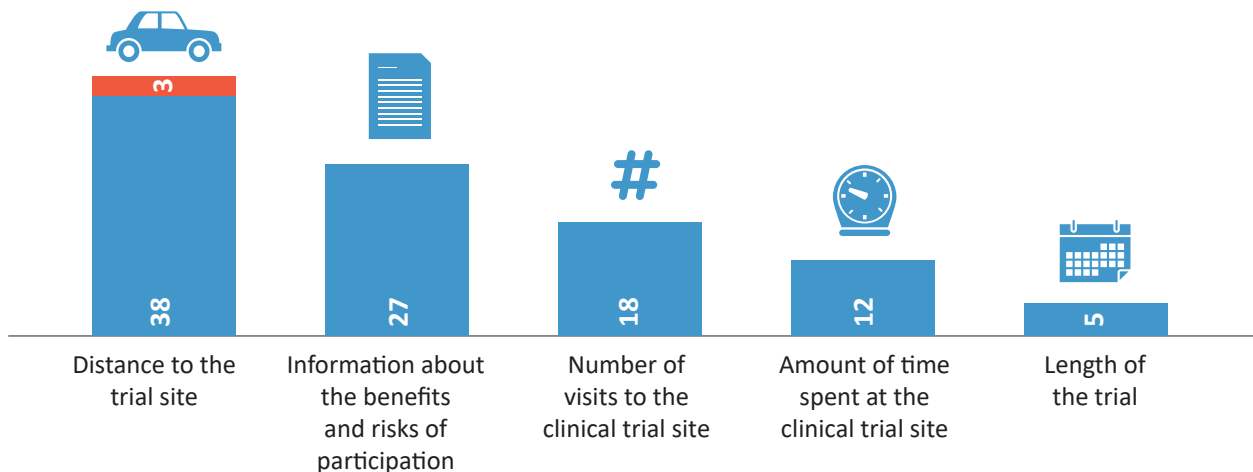
All 3 adults would prefer a pill treatment

What **adolescents** said they would want a **Kabuki Syndrome treatment** to do

Make me normal

help me with schoolwork

Factors that would affect clinical trial participation



Kabuki Syndrome Survey Research

Study Overview



The primary objective of this study was to obtain information about Kabuki Syndrome from both a patient and a caregiver perspective. This information is the result of a research survey among patients and caregivers conducted to learn more about Kabuki Syndrome.



Participants

A total of 71 individuals participated in the survey research study. 57 caregivers participated, along with 11 adolescents who completed separate adolescent surveys with or without their caregiver's assistance. Three adults with Kabuki Syndrome also completed the adult version of the survey.



Survey Topics

Caregiver Survey Topics:

Characteristics of Kabuki Syndrome, Impacts of Kabuki Syndrome and Future Clinical Trial Participation

Adolescent Survey Topics:

Impacts of Kabuki Syndrome

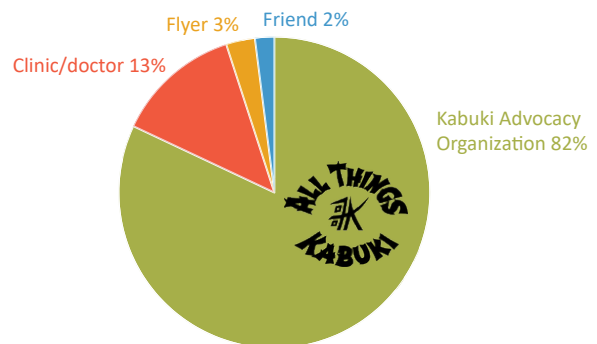
Adult Survey Topics:

Characteristics of Kabuki Syndrome, Impacts of Kabuki Syndrome and Future Clinical Trial Participation



How did participants hear about the survey?

- Friend
- Flyer
- Clinic/doctor
- Kabuki Advocacy Organization



Thank you!

Takeda Pharmaceuticals would like to thank all of the patients, parents, and caregivers who so generously completed these surveys, and extend a special thank you to **All Things Kabuki** (www.allthingskabuki.org) for their assistance in spreading the word about this study.



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