Goals

- better understand the patient and family experience
- better define the range of symptoms
- identify gaps in treatment and patient care
- identify gaps in knowledge and understanding of this disease
- better understand the burdens of this disease on the patient and the family
- identify the most important components of a future natural history study
- develop and prioritize future service programs
- create and prioritize a patient-led strategic research plan
- develop better and more effective clinical trials for potential future treatments

**method**

anonymous survey, Qualtrics, Castle IRB
Demographics

183 complete responses

31 countries

United States 44%

28 states

UK 8%
Italy 6%
GER 5%
CAN 4%
ARG 3.3%
NL, ES, RO, PL 3%

all others 2% or less

260 responses
Demographics

female 57%  male 43%

caregiver or parent 95%

age and distribution

youngest infant
average 22 years
oldest 53 years

birth to 6 yr 23%
7-12 years 37%
13-17 years 17%
18-53 years 23%

average 22 years

urban 40.4%
suburban 34.3%
rural 25.3%

identity groups

Black 1%
Asian/PI 3%
Multi 4%
Other 2%
Hispanic 10%
White NH 79%

n=242
n=250
n=238
n=242
n=250

COLLECTIVE VOICES PROJECT
Diagnosis

average # of physicians seen to get diagnosis 8

who made the diagnosis?

- Nurse Practitioner
- Family Doctor
- Pediatrician
- Movement Disorder Specialist
- Epileptologist
- Metabolic Specialist
- Geneticist
- Neurologist
- Other

who made the diagnosis?

n=230

Symptoms

n=260

diagnostic tools used

- Genetic Testing
- Lumbar Puncture
- Fasting EEG
- Red Blood Cell Uptake Assay
- PET Scan
- METAGlut1
- Other

COLLECTIVE VOICES PROJECT
Diagnosis

additional family members 10% n=230

first symptom to diagnosis
shortest 1.5 weeks
longest 34 years
average 2.8 years

<table>
<thead>
<tr>
<th>Age Group</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 to 11 months</td>
<td>18%</td>
</tr>
<tr>
<td>1-4 years</td>
<td>44%</td>
</tr>
<tr>
<td>5-9 years</td>
<td>22%</td>
</tr>
<tr>
<td>10-14 years</td>
<td>9%</td>
</tr>
<tr>
<td>15-35 years</td>
<td>7%</td>
</tr>
</tbody>
</table>

n=203

age at diagnosis
youngest 12 days
oldest 39 years
average 6 years

n=227

year of diagnosis

n=230

COLLECTIVE VOICES PROJECT
Diagnosis

biggest obstacles to diagnosis

- lack of knowledge by healthcare team
- treated symptoms rather than looking for cause
- misdiagnosed
- didn't listen to family concerns
- misinterpreted or missed test results
- other
- lack of access to specialty care/testing
- no obstacles

n=260
Genetics

had genetic testing
- yes 92%
- no 6%
- unsure 2%

n=230

variant found
- yes 82%
- no 8%
- unsure 10%

n=209

additional genes
- yes 16%
- no 53%
- unsure 31%

n=192

type of testing

<table>
<thead>
<tr>
<th>Variant Type</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>unsure</td>
<td>30</td>
</tr>
<tr>
<td>single gene</td>
<td>20</td>
</tr>
<tr>
<td>epilepsy panel</td>
<td>16</td>
</tr>
<tr>
<td>WES</td>
<td>12</td>
</tr>
<tr>
<td>WGS</td>
<td>10</td>
</tr>
<tr>
<td>movement panel</td>
<td>6</td>
</tr>
<tr>
<td>other</td>
<td>6</td>
</tr>
</tbody>
</table>

Type of variant

<table>
<thead>
<tr>
<th>Variant Type</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>unsure</td>
<td>49</td>
</tr>
<tr>
<td>other</td>
<td>13</td>
</tr>
<tr>
<td>missense</td>
<td>13</td>
</tr>
<tr>
<td>whole deletion</td>
<td>9</td>
</tr>
<tr>
<td>small deletion</td>
<td>8</td>
</tr>
<tr>
<td>splice site</td>
<td>4</td>
</tr>
<tr>
<td>nonsense</td>
<td>4</td>
</tr>
<tr>
<td>duplicatiion</td>
<td>1</td>
</tr>
</tbody>
</table>

NF1, MBDS, TUBA1A, HCN4, SCN4A, SHH, CHD2 -
CHRNA2, GPR98, TANGO2, USP7, IFIH1, PIGG, EPM2A,
MTOR, SCN1A, BRAT1, BRCA1, CENPJ

COLLECTIVE VOICES PROJECT
Medical Care

Setting for highest level of care for Glut1 Deficiency (top 3):
- Neurologist within hospital clinic: 40%
- Epilepsy clinic within hospital: 23%
- Metabolic clinic within hospital: 14%

Quality of care rating average (0-10): 7

Worked with a dietitian: 90%

Quality of care rating average (0-10): 7

Roughly 1/2 eligible to transition to adult care have done so.

Roughly 1/3 have traveled out of state or country for medical care.

*30% have experienced transition difficulties, mostly around specialty care.

n=209
are your medical professionals proactive in learning about Glut1 Deficiency

68% report their healthcare providers have other Glut1 patients

71% of those feel it results in better care

unsure 28%

yes 55%

no 17%

n=209

COLLECTIVE VOICES PROJECT
Symptoms

**First symptom**

- Unusual eye/head movements: 34%
- Seizures: 27%
- Missing milestones: 12%
- Unusual body movements: 10%
- Floppiness/tone issues: 8%
- Other: 9%

**"Classical" symptoms**

- Seizures
- Cognitive
- Developmental delays
- Speech/language issues
- Floppiness/tone
- Eye/head movements
- Unusual body movements

n=203
Symptoms

<table>
<thead>
<tr>
<th>Symptoms</th>
<th>Percentages</th>
</tr>
</thead>
<tbody>
<tr>
<td>stamina/endurance</td>
<td>45%</td>
</tr>
<tr>
<td>low energy</td>
<td>44%</td>
</tr>
<tr>
<td>memory problems</td>
<td>34%</td>
</tr>
<tr>
<td>ADD/focus</td>
<td>31%</td>
</tr>
<tr>
<td>sleep disturbances</td>
<td>32%</td>
</tr>
<tr>
<td>temp regulation</td>
<td>26%</td>
</tr>
<tr>
<td>mood disturbances</td>
<td>24%</td>
</tr>
<tr>
<td>anxiety</td>
<td>23%</td>
</tr>
<tr>
<td>migraines</td>
<td>22%</td>
</tr>
<tr>
<td>swallowing/chewing issues</td>
<td>21%</td>
</tr>
<tr>
<td>retina/vision issues</td>
<td>21%</td>
</tr>
<tr>
<td>behavior disturbances</td>
<td>19%</td>
</tr>
<tr>
<td>OCD</td>
<td>19%</td>
</tr>
<tr>
<td>microcephaly</td>
<td>19%</td>
</tr>
<tr>
<td>episodic confusion</td>
<td>19%</td>
</tr>
<tr>
<td>excessive drooling</td>
<td>18%</td>
</tr>
<tr>
<td>gastro issues</td>
<td>19%</td>
</tr>
<tr>
<td>autism spectrum</td>
<td>14%</td>
</tr>
<tr>
<td>cyclic vomiting</td>
<td>12%</td>
</tr>
<tr>
<td>teeth/nail issues</td>
<td>12%</td>
</tr>
<tr>
<td>chronic pain</td>
<td>13%</td>
</tr>
<tr>
<td>MRI issues</td>
<td>8%</td>
</tr>
<tr>
<td>kidney issues</td>
<td>9%</td>
</tr>
<tr>
<td>heart issues</td>
<td>6%</td>
</tr>
<tr>
<td>chronic pain</td>
<td>2%</td>
</tr>
<tr>
<td>other</td>
<td>10%</td>
</tr>
</tbody>
</table>

n=260

Other symptoms:
- leg pain
- other
Seizures

36% report difficulty distinguishing between seizure & movement episode

84% experienced seizures

2/3 no longer having seizures - diet?

percentages

absence 47%
atypical absence 29%
tonic-clonic 27%
myoclonic 26%
atonic 20%
tonic 17%
automatisms 9%
sensations/emotions 6%

top frequency

n=199
Movements

Muscle or movement issues: 77% (n=202)

2/3 report certain body parts affected most and both sides of body affected equally:
- Legs, eyes, arms, fingers, mouth

Percentages:
- Ataxia: 71%
- Dysarthria: 48%
- Dyspraxia: 45%
- Hypotonia: 44%
- Dystonia: 43%
- Spasticity: 34%
- PED: 28%
- Tremor: 26%
- Athetosis: 25%
- Ballismus: 19%
- Hemiplegia: 16%
- Chorea: 15%
- Other: 14%

- 5%
Symptom Triggers

- **Fatigue**: 37%
- **Heat**: 31%
- **Hunger**: 28%
- **Prolonged Exercise**: 28%
- **Illness**: 25%
- **Dehydration**: 21%
- **Excitement**: 21%
- **Strong Emotions**: 20%
- **Anxiety**: 17%
- **Hormones**: 16%
- **Weather/Barometric Changes**: 10%
- **Other**: 6%
- **Altitude Changes**: 5%

Symptom triggers were reported by 61% of participants, with 25% unsure and 14% reporting no. The survey included 203 participants. Further analysis of symptom triggers included:

- **Fatigue**
- **Heat**
- **Hunger**
- **Prolonged Exercise**
- **Illness**
- **Dehydration**
- **Excitement**
- **Strong Emotions**
- **Anxiety**
- **Hormones**
- **Weather/Barometric Changes**
- **Other**
- **Altitude Changes**

The COLLECTIVE VOICES PROJECT logo is visible at the bottom of the image.
Other Symptoms & Behaviors

n=203

- **High pain threshold**: 44%
- **Sensory seeking**: 27%
- **Sensory avoidance**: 19%
- **Self-stimulatory**: 25%
  - Occasionally 66%
  - Frequently 33%

**Vascular issues**
- Port wine stain birthmark
- Unusual brain vascular findings (MRI)
- Hemangioma/strawberry mark
- Stork bite birthmark
- None

**Self-reported disease severity rating (0-10)**
- 6 avg.

n=211
Symptoms & Quality of Life

top 3 symptoms negatively impacting quality of life

- cognitive or intellectual difficulties: 44%
- speech/communication issues: 35%
- lack of independence: 24%
- movement issues present all the time: 23%
- seizures: 18%
- fatigue: 15%
- movement episodes: 15%
- ADD/focus: 9%
- other: 9%
- anxiety: 8%
- mood disturbances: 6%
- behavior disorders: 5%

n=260
41% have experienced or are currently in puberty

Did symptoms change in puberty?

- Yes: 56.2% (n=80)
- No: 17.5% (n=20)
- Unsure: 26.3%

How did symptoms change?

- Improved: none
- Stayed same: cognitive, speech/language
- Worsened: movements, stamina/energy, seizures, anxiety

Did treatments need to change?

- 45% yes

How did treatment need to change?

- Diet changes: 49%
- New meds: 36%
- New therapies: 8%
- Other: 8%
Puberty

Did you experience new symptoms for the first time in puberty?

- Yes: 37.7%
- No: 44.2%
- Unsure: 18.2%

n=77

What new symptoms did you experience for the first time in puberty?

- Seizures first time: 0%
- New seizure types
- Movements first time: 25%
- New movement types: 30%
- Anxiety: 10%

COLLECTIVE VOICES PROJECT
Adulthood

Did symptoms change in adulthood?
- Yes: 73.7% (n=38)
- No: 23.7% (n=38)
- Unsure: 2.6%

How did symptoms change?
- *Improved*
  - Seizures
  - Cognitive
  - Speech/language
  - Memory
- *Stayed same*
  - Speech/language
  - Memory
- *Worsened*
  - Movements
  - Stamina/energy

Quality of life in adulthood
- Improved: 45%
- Same: 34%
- Worsened: 21%

On KDT
- Able to manage diet independently: 20%
- 57%

10% have children of own
- 75% of children also have Glut1 Deficiency

How do symptoms compare?
- Similar: 67%
- More severe: 33%
- Less severe: 0%
Development

meet developmental milestones on time?

- Physical/motor: 75
- Speech/language: 50
- Cognitive: 25
- Social/emotional: 0

experience challenges in any of these developmental domains?

- Fine motor: 21.8%
- Speech/language: 21.1%
- Gross motor: 21%
- Cognitive: 20.5%
- Social/emotional: 15.6%

- Most major childhood milestones met but delayed
- Most major adult milestones not met

n=190
Development & Mobility

Do you walk independently?

- yes | without support: 71%
- yes | but use support only for longer distances: 14%
- yes | but use support: 8%
- no: 7%

average age for walking: 2 years
- oldest: 9 years
- youngest: 8 months

n=190

COLLECTIVE VOICES PROJECT
Development

early intervention services 73%

- adults able to manage KDT independently
  - yes 20%
  - no 80%
  - n=26

- adults able to manage medications independently
  - yes 45%
  - no 55%
  - n=38

toileting

- reached on time 30%
- reached but delayed 51%
  - n=190

n=190

COLLECTIVE VOICES PROJECT
Speech & Language

able to communicate using own voice: 84% (n=190)

- No issues: 32%
- Fluency: 31%
- Minor articulation: 18%
- Severe articulation: 9%

average age to speak well enough to communicate: 3.5 years (n=190)

receptive language better than expressive?
- Yes: 66%
- No: 3%
- Both equal: 22%
- Unsure: 9%

half feel speech issues make them appear less capable/intelligent than they are

2/3 experience frustrations around communication issues
Cognitive

83% report certain subjects harder
- math: 39%
- writing/composition: 33%
- reading: 15%
- spelling: 7%
- science: 3%
- social studies: 3%

87% reported deficits
- attention/focus
- visuospatial, visual attention
- planning and organization
- reasoning and problem solving
- memory
- language functions

Only 56% reported neuropsychological testing

Only 25% knew IQ score
- Majority reported 60-70 range
- Average of all scores reported was 79
### Cognitive & School

<table>
<thead>
<tr>
<th><strong>Performing at Grade Level</strong></th>
<th>33%</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Beneficial Supports</strong></td>
<td></td>
</tr>
<tr>
<td>IEP</td>
<td>66%</td>
</tr>
<tr>
<td>1:1 aide</td>
<td>60%</td>
</tr>
<tr>
<td>504 plan</td>
<td>18%</td>
</tr>
<tr>
<td>special transportation</td>
<td>31%</td>
</tr>
<tr>
<td>shortened school day</td>
<td>20%</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>College/Vocational</strong></td>
<td>27%</td>
</tr>
<tr>
<td><strong>Trouble Getting Good Curriculum Fit?</strong></td>
<td></td>
</tr>
<tr>
<td>unsure</td>
<td>16%</td>
</tr>
<tr>
<td>yes</td>
<td>42%</td>
</tr>
<tr>
<td>no</td>
<td>42%</td>
</tr>
</tbody>
</table>

All forms of school based therapies were reported as highly beneficial.

*COLLECTIVE VOICES PROJECT n=145*
Social & Emotional

Impacts on social life

- speech/articulation: 33%
- cognitive or intellectual: 31%
- ketogenic diet challenges: 25%
- lack of independence: 21%
- immaturity: 20%
- movement issues: 17%
- unpredictability of symptoms: 9%
- behavior disturbances: 8%
- seizures: 7%
- other: 7%
- autism spectrum symptoms: 4%

Self-reported happiness rating (0-10)
7.6 average

77% describe self as social
2/3 report having close friendships
27% report mood disturbances
20% report behavior disturbances

COLLECTIVE VOICES PROJECT
Social & Emotional

Sibling Impacts

- Close friendship with Glut1 Deficiency sibling: 77%
- Missed out on traditions and food celebrations due to KDT: 21%
- Taken on caregiving responsibilities: 21%
- Missed out on social opportunities: 21%
- Significant time at medical appointments, therapies, etc.: 15%
- Extreme jealousy or resentment: 7%
- Taken on advocacy role: 4%
- Other: 7%

n=260
Ketogenic Diets

91% have tried ketogenic dietary therapy

shortest: 1 month       longest: 21 years
average: 5 years

has KDT lost effectiveness over time?

no 64%
unsure 20%
yes 16%

7.5% have stopped KDT due to lack of effectiveness

n=260

has KDT lost effectiveness over time?

no 64%
unsure 20%
yes 16%

7.5% have stopped KDT due to lack of effectiveness

n=260

KDT types used

65% classical 3:1 or 4:1
41% modified ketogenic diet
32% modified Atkins diet
16% MCT oil version
5% low glycemic index

91% have tried ketogenic dietary therapy

shortest: 1 month       longest: 21 years
average: 5 years

KDT types used

65% classical 3:1 or 4:1
41% modified ketogenic diet
32% modified Atkins diet
16% MCT oil version
5% low glycemic index

7.5% have stopped KDT due to lack of effectiveness

n=260
Ketogenic Diet Benefits

- Seizures: very effective
- Movement issues: somewhat effective
- ADD/focus: not at all effective
- Energy/stamina: not at all effective
- Headaches: somewhat effective
- Behavior: somewhat effective
- Speech/language: very effective

Overall: n=167
Ketogenic Diet Benefits

- attention/focus
- behavior
- cognition
- energy/stamina
- headaches/migraines
- movement issues
- seizures
- speech/communication

**by age**

Age Group:
- 0-12
- 13 and Over

n=167
Ketogenic Diets

83% measure ketones

**method**
- Urine: 21%
- Blood: 78%
- Breath: 1%

**correlation to symptoms?**
- Yes: 65%
- No: 11%
- Unsure: 24%

53% report using MCT oils
65% of those feel MCT oil makes the diet more effective

Ideal ratio: 3:1
44%

"Just right" ketone level: 3-5

8% report using feeding tube

n=167
Ketogenic Diets

reasons for not trying KDT

- family wasn’t supportive: 0%
- doctor wasn’t supportive
- foods too expensive
- thought it was too difficult
- thought I was too old
- thought I wouldn’t be able to give up foods I like
- no KDT services available to me
- thought it wouldn’t work for my symptoms
- other

reasons:

- Doctor advised that Trileptal was working for me as well or better than I could expect with a ketogenic diet.
- I just got diagnosed and plan to meet with a dietician soon to start the diet.
- I just got diagnosed and plan to meet with a dietician soon to start the diet.
- GONG TO BE IN A STUDY THAT REQUIRES NO KETO DIET
- She does not eat enough with a diet will be worst, seizures are under control with medicine kapra
- Thought it might be too challenging since I am 14.
- Didn’t try because it would exclude her from the trial
- I do modified Atkins

COLLECTIVE VOICES PROJECT
### Ketogenic Diet Side Effects

#### Percentages

- **Constipation**: 59%
- **Weight Gain**: 26%
- **High Cholesterol**: 25%
- **Gastro Issues**: 23%
- **Slowed Physical Growth**: 23%
- **Kidney Issues**: 21%
- **Acidosis**: 17%
- **Weight Loss**: 16%
- **Bone Density Issues**: 16%
- **Other**: 10%
- **Pancreatitis**: 1%

#### Experienced Side Effects

- **Percentages**: 41%

#### Stopped KDT Due to Side Effects

- **Temporarily**: 4%
- **Permanently**: 3%

Additional Side Effects:
- Psychological stress, dehydration, hair loss, bad breath, headaches, carnitine & calcium deficiencies, leg cramps

---

*COLLECTIVE VOICES PROJECT*
Ketogenic Diet Challenges

choose top 3

- social: feeling different 54%
- social: impact on family celebrations/holidays 52%
- lack of opportunities for spontaneity 43%
- time required 40%
- costs of food and supplies 28%
- cooperation/compliance 18%
- managing complexities of diet (calculations, etc.) 17%
- lack of effectiveness for all symptoms 11%
- other 9%
- lack of medical support 7%
- managing side effects 6%

n=167

level of difficulty (0-10)

patient 6.4
family 6.6

do benefits outweigh challenges?

yes 83%
unsure 13%
no 4%

COLLECTIVE VOICES PROJECT
Ketogenic Diet Challenges

have you had issues getting KDT supplies and services covered by insurance?

- yes: 44%
- no: 56%

n=167

Chart showing the percentage of respondents who have had issues with different aspects of KDT supplies and services:

- Prescribed medical foods: 75%
- Prescribed supplements: 50%
- Ketone testing supplies: 25%
- Medications: 0%
- Dietitian services: 0%

COLLECTIVE VOICES PROJECT
Therapies & Assistive Devices

most common therapies

speech  occupational  physical

85% have tried and found beneficial

1/3 report that moderate physical exercise is beneficial for symptom management

assistive devices used most

orthotics, eyeglasses, wheel and stroller chairs, walkers, adaptive clothing
### Other Treatments

#### Have you tried other treatments that help?

<table>
<thead>
<tr>
<th>Condition</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>seizures</strong></td>
<td>35%</td>
</tr>
<tr>
<td>keppra/levetiracetam</td>
<td>22% or 14 total</td>
</tr>
<tr>
<td>sodium valproate/depakote</td>
<td>11% or 7 total</td>
</tr>
<tr>
<td>acetazolamide/diamox</td>
<td>8% or 5 total</td>
</tr>
<tr>
<td>ethosuximide</td>
<td>8% or 5 total</td>
</tr>
<tr>
<td>lamotrigine/lamictal</td>
<td>6% or 4 total</td>
</tr>
<tr>
<td><strong>movements</strong></td>
<td>17%</td>
</tr>
<tr>
<td>baclofen</td>
<td></td>
</tr>
<tr>
<td>acetazolamide/diamox</td>
<td></td>
</tr>
<tr>
<td><strong>ADD/focus</strong></td>
<td>16%</td>
</tr>
<tr>
<td>vyvanse</td>
<td></td>
</tr>
<tr>
<td>methylphenidate/ritalin</td>
<td></td>
</tr>
<tr>
<td><strong>anxiety</strong></td>
<td>10%</td>
</tr>
<tr>
<td>CBD oil</td>
<td></td>
</tr>
<tr>
<td>sertraline</td>
<td></td>
</tr>
<tr>
<td>paroxetine</td>
<td></td>
</tr>
<tr>
<td><strong>migraines</strong></td>
<td>10%</td>
</tr>
<tr>
<td>acetaminophen/paracetamol</td>
<td></td>
</tr>
</tbody>
</table>

#### 28% have used rescue meds for seizures
- diazepam/diastat/stesolid
- midazolam

#### 16% have used rescue meds for movement episodes
- diazepam/diastat/stesolid
- lorazepam
- clobazam
- CBD oil

n=184
## Other Treatments

### ketone supplements
- 20% have tried
- 60% found it effective

### CBD Oil
- 9% have tried
- 53% found it effective
*movement, anxiety*
- 20% have tried
- 60% found it effective

### VNS
- 5% have tried
- 12.5% found it effective

### corn starch
- 6% have tried
- 40% found it effective

### C7 oil/triheptanoin
- 14% have tried
  - how effective?
- 48% of those have tried both KDT and C7

### Energy/Stamina

<table>
<thead>
<tr>
<th></th>
<th>very</th>
<th>somewhat</th>
<th>not at all</th>
</tr>
</thead>
<tbody>
<tr>
<td>cognition</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>attention</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>seizures</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>speech</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>movement</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>behavior</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>headaches</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

### Use both together
- 16.7%

### Equal effectiveness
- 16.7%

### Most effective
- KDT 50%
- C7 oil 16.7%

n=184
Patient & Family Research Priorities

- 46% for new and better treatments
- 27% for basic science for better understanding disease
- 22.3% for changes in adulthood
- 21.8% for long term KDT effects
- 21.4% for changes in puberty
- 19.3% for potential impact on other body systems
- 14% for different ketogenic diets/which best?
- 12% for newborn screening development
- 10% for better understand genetic mutations
- 3.4% for improved diagnostic testing
- 2.9% for role of inflammation
- 2.5% for unusual eye/head movements
- 2.5% for other

n=260
Priority Outcomes for New Treatments

*choose top 3*

- able to eat a normal diet: 35%
- improved cognition: 27%
- better speech/communication: 25%
- eat an easier or less restrictive keto diet: 23%
- greater independence: 23%
- more typical development: 16%
- fewer movement issues: 14%
- improved coordination/balance: 12%
- fewer seizures: 10%
- improved fine motor skills: 9%
- better sleep: 5%
- improved gross motor skills: 4%

n=260
Types of New Treatments Willing to Try

- pill or tablet: 65%
- oil or liquid: 58%
- dietary therapy: 56%
- IV or injection into skin/muscle: 42%
- infusion pump: 25%
- IV or injection into spinal fluid: 23%
- IV or injection into brain: 17%

Willing to do clinical trials?
- Yes: 60.4%
- Maybe: 34.7%
- No: 5%

n=260
Family Burdens
choose top 3

financial burden
- cost of ketogenic diet: 32%
- gave up career for caregiving: 27%
- out of pocket therapies: 23%
- out of pocket medical costs: 20%
- travel costs for medical care: 15%
- more expensive insurance plans: 9%

overall burden
- overall family life: 43%
- social life: 42%
- emotional/mental: 40%
- finances: 32%
- sibling life: 30%
- career: 24%
- relationship spouse/partner: 24%
- other: 4%

- significant sibling impact
- long term care & financial planning often needed

n=260

COLLECTIVE VOICES PROJECT
Please join the Natural History Study to help tell the full story of Glut1 Deficiency across the lifespan.

Thank you!

COLLECTIVE VOICES PROJECT