



research



roundtable

FALL EDITION

*welcome!*



# purpose

To provide a forum for scientists and clinicians to share updates on current projects, engage in discussions, and foster collaborations.

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accelerate *progress* toward our vision:

A brighter future where Glut1 Deficiency will be easy to diagnose early, treat effectively, and cure completely.



# zoom courtesy

- Zoom name
- video on if comfortable
- muted unless talking
- chat any questions or comments
- no recordings or screenshots without permission
- reach out if you'd like to further connect

**introduce yourself in chat - where you're from,  
what is your interest in or connection to Glut1?**



# agenda

- family experiences and perspectives
- adult experiences
- tips and challenges - group discussion
- gaps in resources and care - group discussion
- C7 implementation and dosing



# agenda: *special guests*



**Juan Pascual, MD, PhD**  
Weill Cornell



**Kelly Faltersack, MS, RD, LDN, CD**  
University of Wisconsin-Madison



## We're focusing on 8 major areas:

> OPEN SOURCE RESEARCH TOOLS

> CELLS

> GLUT1 - THE TRANSPORTER

> METABOLISM

> GENETICS

> PATHOGENESIS

> THERAPY DEVELOPMENT

> CLINICAL TOOLS



research  
*compass*



Help us drive *progress*

We need faster, easier diagnosis and deeper, clearer understanding of this rare disease so we can find better ways to treat it. The burdens are many, and the patients are waiting.

CONTRIBUTE IDEAS OR RESOURCES >





research



roundtable

WINTER EDITION

TUESDAY, JANUARY 21, 2025

11:00-12:00 Eastern



Asheville  
NORTH CAROLINA



SCIENTIFIC AND FAMILY  
**SUMMIT**

JULY 13-15, 2026





thank  
you!





# Ketogenic Diets

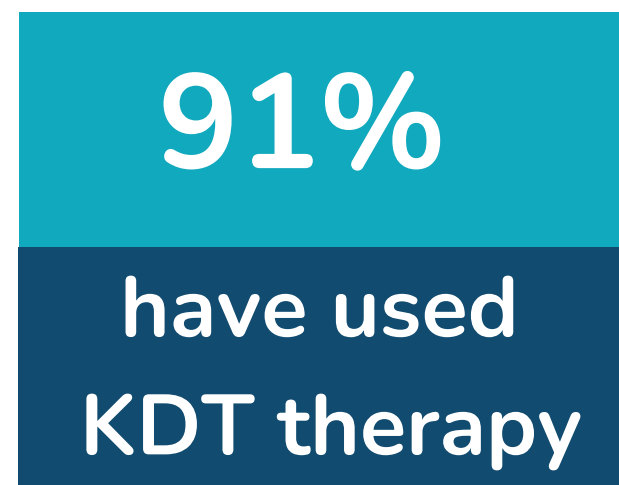
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family *perspectives*



RESEARCH ROUNDTABLE  
OCTOBER 22, 2024

# survey demographics



## age distribution

birth to 6 yr	20%
7-12 years	22%
13-17 years	15%
18-70 years	43%

# ketogenic diet *usage*

## KDT use by age group

age in years	KDT use by age group	most used KDT by age group	# of KDT tried by age group
0-6	90%	modified KD	1.2
7-12	97%	classical KD	1.4
13-17	90%	classical KD	1.9
18 and older	*83%	classical KD	2.1

## types of KDT used overall

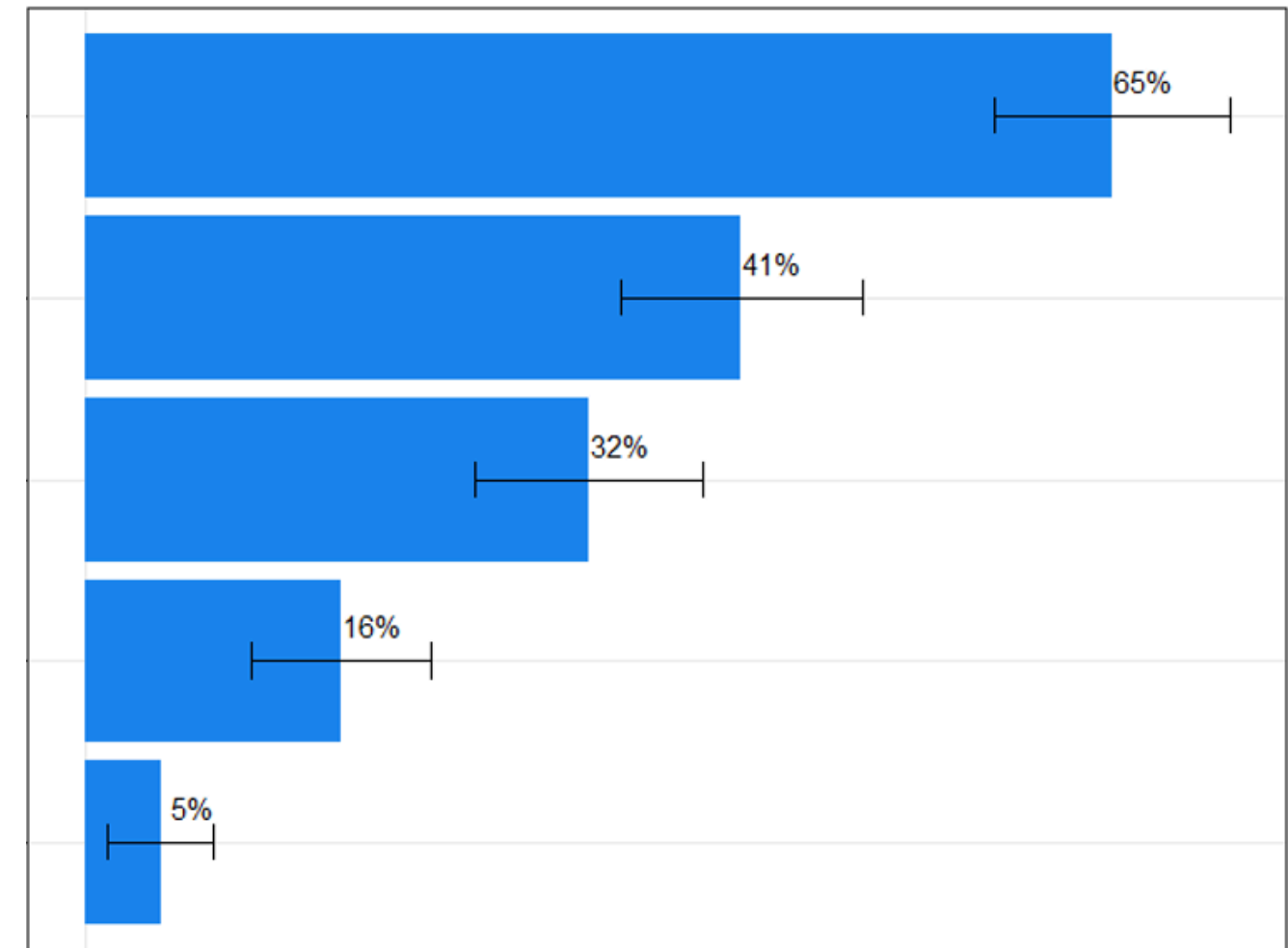
Classical Ketogenic  
(3:1 to 4:1 ratios)

Modified Ketogenic  
(other ratios)

Modified Atkins

MCT

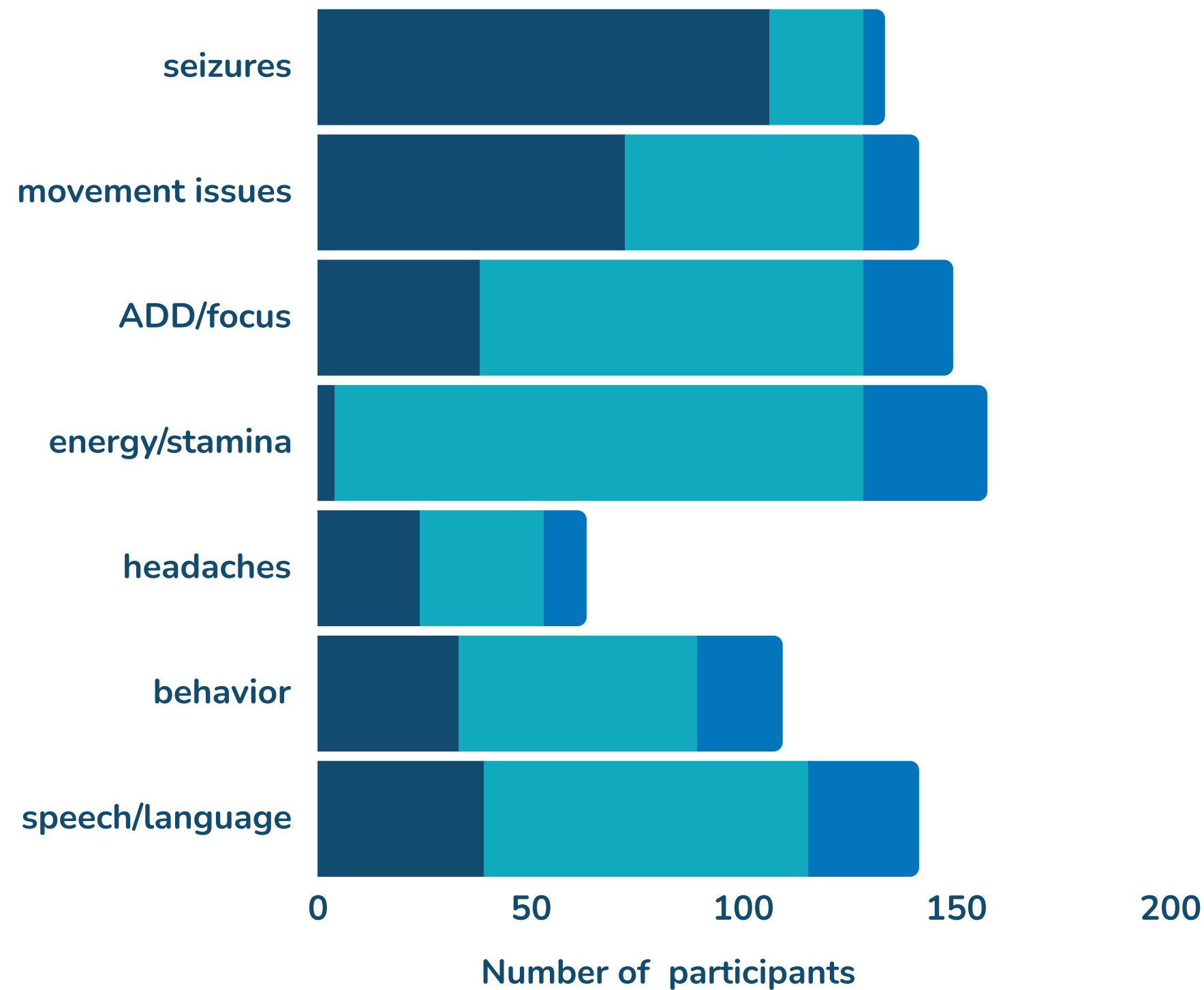
Low Glycemic  
Index Treatment



Percentage

n=167

# ketogenic diet *benefits*



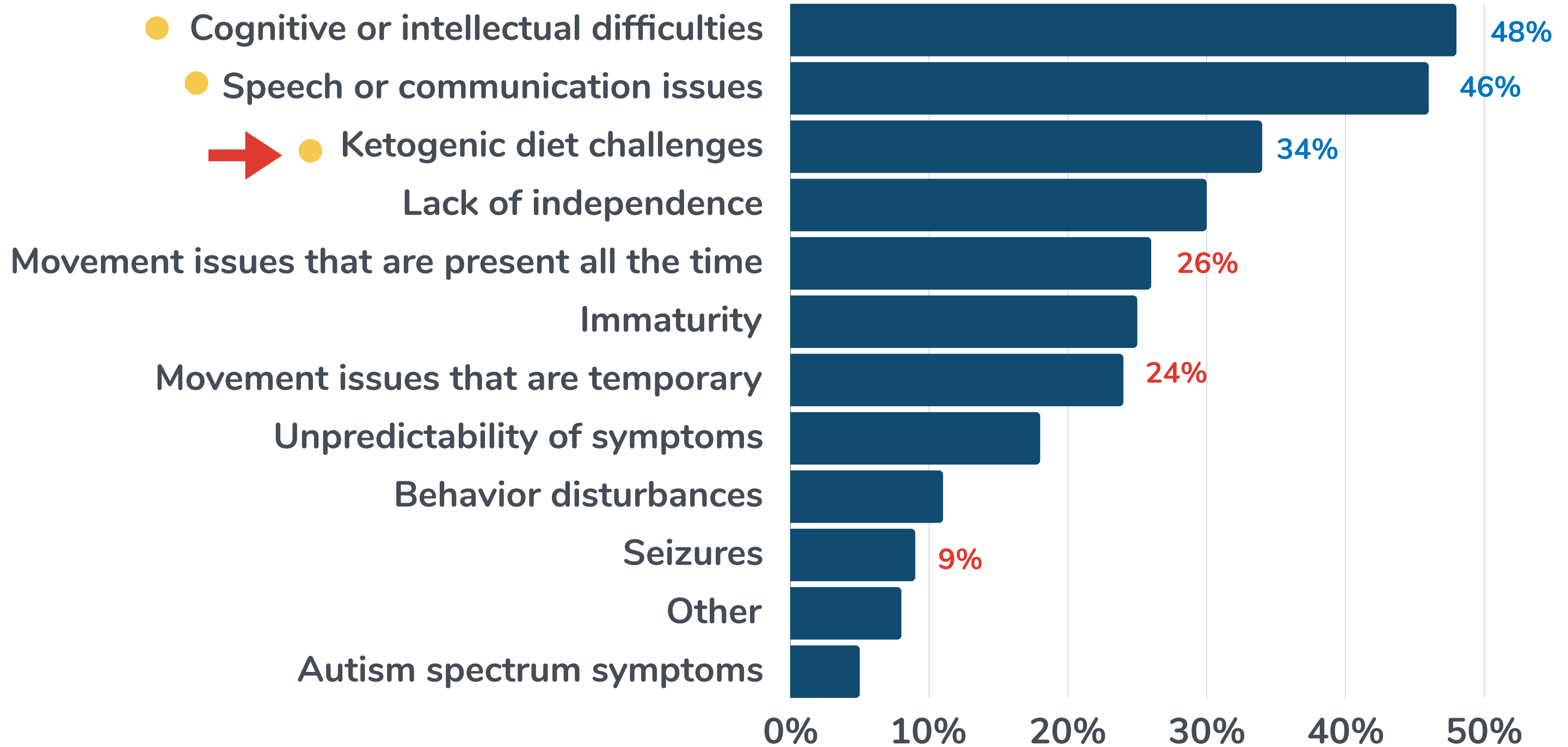
very effective

somewhat effective

not at all effective

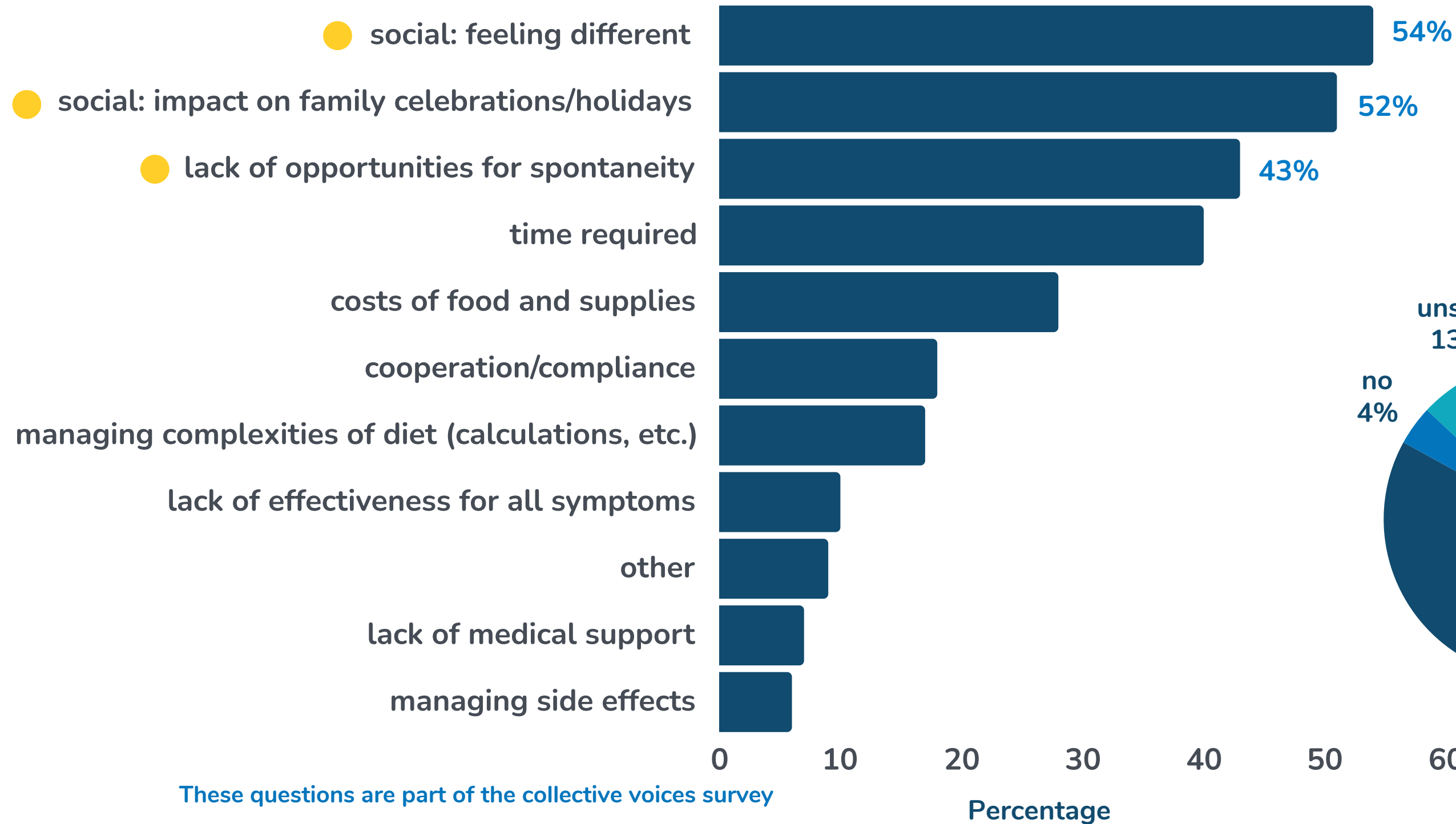
choose top 3

## impacts on social life



# ketogenic diet challenges

choose top 3

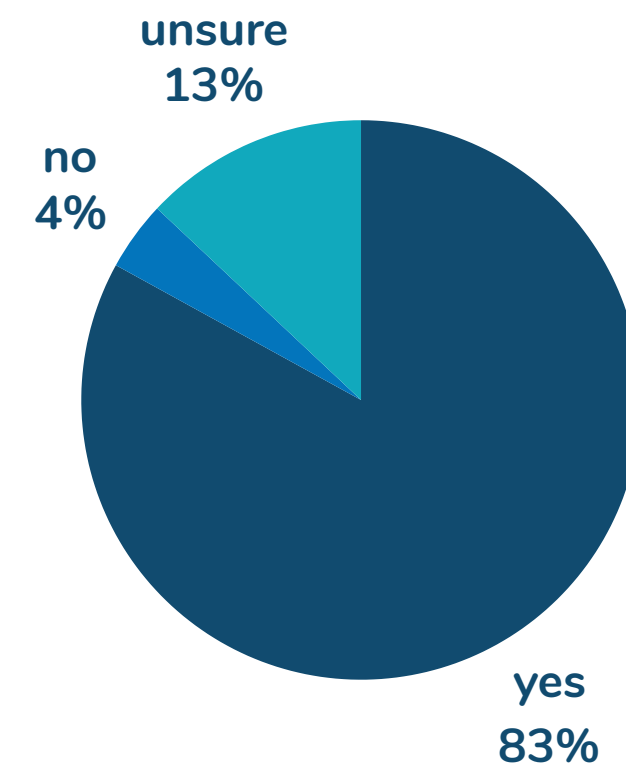


These questions are part of the collective voices survey

level of difficulty (0-10)

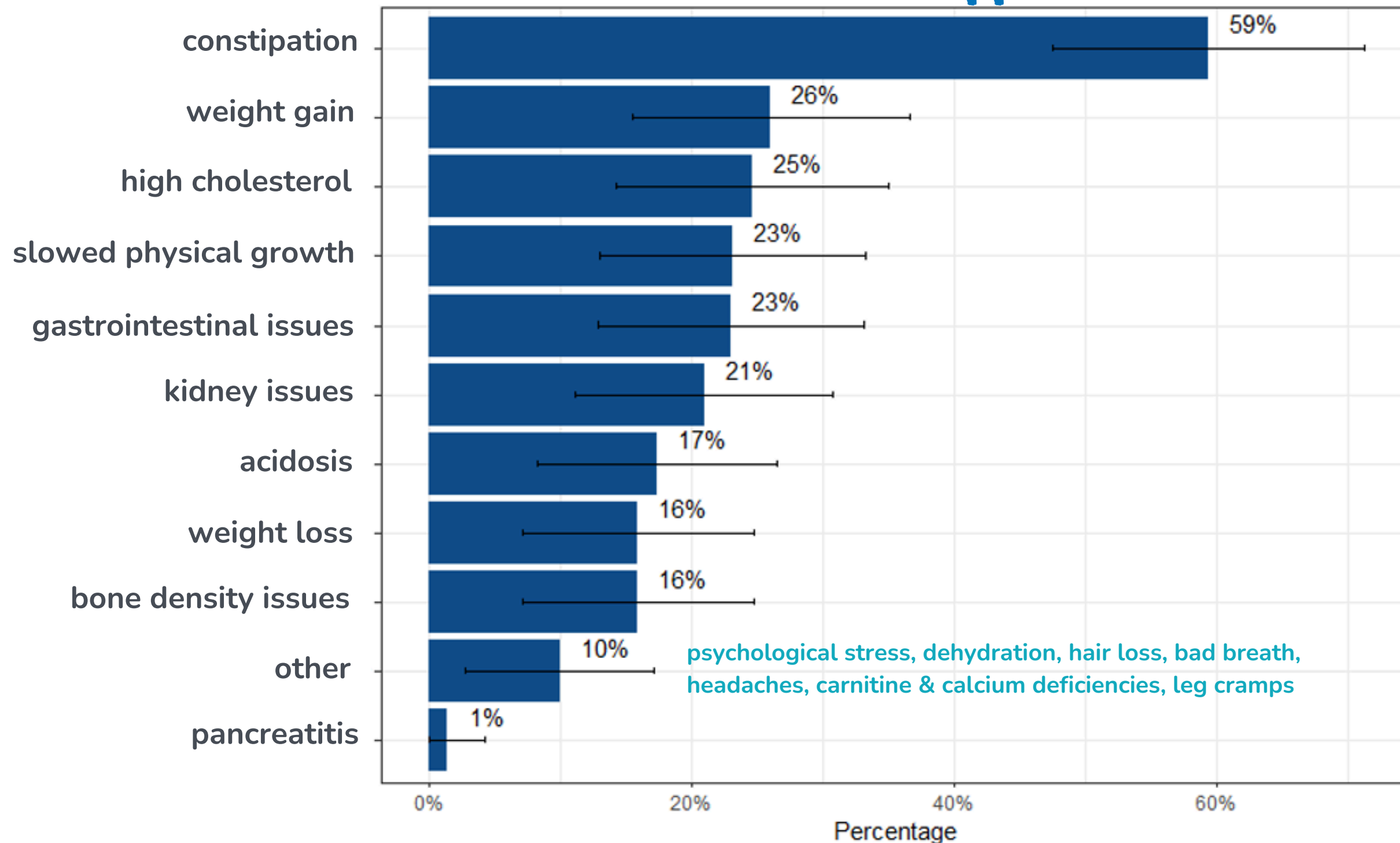
patient 6.3

family 6.5



do benefits outweigh challenges?

# ketogenic diet *side effects*



n=167

**41%** report experiencing side effects on KDT

**stopped KDT due to side effects**  
4% temporary      3% permanent

**7.5%** stopped KDT due to lack of effectiveness

**10%** patients on KDT use a feeding tube

**37%** report difficulties implementing KDT at school

**80%** adult patients are unable to manage a KDT independently

# ketogenic diet experiences *summary*

- Most of the participants in the survey report they have used the KDT to treat Glut1 Deficiency.
- KDT is very effective for seizure control but not as effective for other symptoms such as movement issues or speech and language.
- The KDT brings many challenges that impact the patients' QoL with an impact on social life.
- KDT benefits outweigh its challenges.

PARENT AND CAREGIVER

ketogenic diet care

*survey*

52 responses

OCTOBER 2024

# ketogenic diet *care*



Biggest benefits of using ketogenic diet therapy for you or your loved one with Glut1 Deficiency:

developmental gains    attention    no brain fog  
quality of life    mood    focus    feels better  
happier  
brain power  
**seizure freedom**  
fewer symptoms    motor skills    coordination    health    behavior  
more alert  
chance at normal life    **increased energy**  
**fewer seizures**    off medication  
started speaking  
fewer headaches  
cognitive    easier movements    increased endurance  
started running    brighter    growth    balance    started walking

# ketogenic diet *care*



Biggest challenges of using ketogenic diet therapy for you or your loved one with Glut1 Deficiency:

adequate supplementation    measuring food    adding own foods to calculator    too rigid

**compliance**    **eating different food than others**

**taking food everywhere**    fine tuning challenges

lack of acceptance    lack of respite    socially isolating    **travel**    weight gain

**prep time required**    nutritionally inadequacies

limits on food amounts    **avoiding carbs**    change in symptoms

side effects    **cost**    getting in enough fat

menstruation    balancing glucose/ketones    lack of dietitian support

**socially isolating**    **lack of variation**    **special events**

too low protein    slowed physical growth    **lack of snack options**    no social support

**limitations of eating out**    **lack of spontaneity**    picky eater

**restrictive**    burden on caregiver    **lack of ready made foods**    avoiding processed foods

gluten confusion

# ketogenic diet *care*

How would you rate the quality of the ketogenic diet support you receive from your care team?



# ketogenic diet *care*

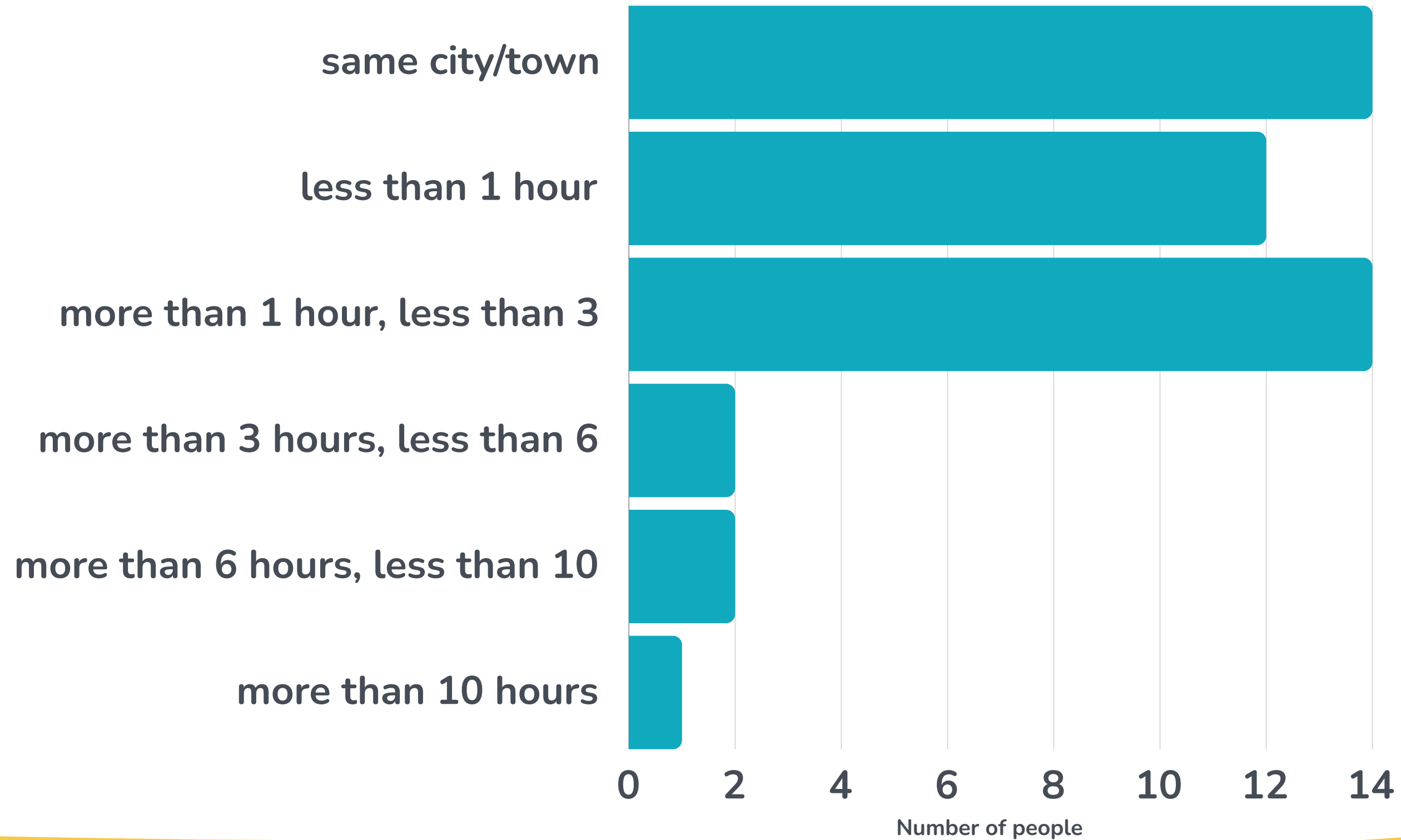
How would you rate the quality of the ketogenic diet support you receive from your care team?



**3.8 out of 5 stars**

# ketogenic diet *care*

How far do you have to travel by car for your ketogenic diet care?



# ketogenic diet *care*

## Part 1

### Challenges you've experienced with the care you receive from your ketogenic diet team:

- They have been excellent and very responsive. Just wish they had a keto calculator rather than needing to calculate exchanges ourselves.
- Problems with transitioning into adult care from Paediatric's- no provision available.
- One size does not fit all. What works for one patient doesn't work for my situation. Also, blood work results aren't always reflective of how well or poor my loved one is doing.
- Understanding the individual needs/tastes of my child.
- Lack of flexibility with meal suggestions (but this was many years ago)
- Dietician never heard of it 25 years ago
- Sometimes the suggestions I get for brands or foods in the diet are foods I don't want to give him because they are unhealthy for anyone. For example, mayonnaise and ranch dressing, which is made out of unhealthy seed oil. Foods or drinks with artificial sweeteners. Deli meats with nitrites/nitrates.
- follow up and response time when problems arise
- Having to travel across the U.S. to see them at our own expense.
- Not responding, hard to reach out, feel like doing it on my own
- Being able to get advice in an emergency. Advice when need to give additional meds ie antibiotics when ill.
- They want labs to be done in their institution
- poor call back communication, poor follow through, our dietician doesn't seem to be in the know.
- Lack of diet support. Just here is the Ketodietcalculataor and good luck contacts us with any questions.
- Lack of response or charging a co pay to talk or ask a question.

# ketogenic diet *care*

## Part 2

### Challenges you've experienced with the care you receive from your ketogenic diet team:

- We are very fortunate to have an excellent ketogenic diet team that is supportive and responsive and collaborates well with other specialists helping us manage our son's care so we are maximize therapeutic benefits while minimizing side effects. Traveling an hour each way for in clinic follow up and testing is probably our biggest challenge.
- My challenge was getting to the care team...trying to get the best way by public transport, where my daughter didn't have to walk far, taxis were out of our budget
- Recently our care team is splitting GLUT1 patients into Ketogenic and Non-Ketogenic patients. This is very new. I didn't pick this team because of their keto diet expertise. Ask me again in a few months. 😊
- We have worked with at least 10 different dietitians over the last 8 years. They will have different approaches and most we don't feel hear us or our concerns. When we have asked to change the ratio or try new meals, one dietitian will begin the process and a few months later a new dietitian will delete the meal or change it back. Our 22 year old son is still under the care of a pediatric program because there is no adult program in the state.
- Slow response times, particularly in terms of not having control of adding foods to the calculator so I can make new meals
- Generalization. Not every kid is the same, yet the diet is the same for their patients
- Menu selection is very limited for a child and most choices are time consuming to prepare. Honestly a lot taste really awful also.
- Our team is amazing. Our dietitian works with our GI doctor and Neurologist.
- transitioning from classic keto to MAD - we haven't made the transition yet but the team has not done this with others before
- Lack of understanding and minimal direction. At times, we felt as if we were helping our dietitian to understand more about the keto diet.
- I think they (fair enough) lack the lived experience of being in a family with a person on the ketogenic diet.
- Dietician turnover over a 15 yr. period. This has been minor issue for us.

# ketogenic diet *care*

## Part 3

### Challenges you've experienced with the care you receive from your ketogenic diet team:

- Our center has only 2 cases of Glut-1 so not much experience. Dietitian staffing limited.
- We only had one 40 minute meeting with the dietitian and the rest we learned on our own. She wasn't very supportive and we never returned
- Biggest challenge is we do not have a ketogenic care team at all. That option was missing in the previous question. There are still places without support.
- We love our ketogenic diet team but we are the only GLUT1 patient at our hospital so we have had to do some more research and trial&error on our side
- We do most sessions virtually and wish they followed up a bit more. I could use more guidance on day to day meals as we get bored of same routine
- We did not see eye to eye with our care team early on because they refused to agree with me continuing to breastfeed. After arguing over it for a few months we parted ways and have worked on our own ever since...24 years now. (And of course now we know that breastfeeding is neuroprotective...vindicated!) Back then we created our own master template in excel that we continue to use today and update regularly to create all our own menus. It's worked well for us over the years and our son is quite healthy. Although we've had a couple of rough patches during puberty and during a change in fats, we've weathered them and our son has mostly been seizure free since the first day after starting the ketogenic diet. It's been a great blessing for us.
- Keto team and neurologist appointments are done together via a video visit. I find the appointment is usually based on the question of, "how are things going" vs anything educational. They put a lot of time in upfront when getting started and now the focus is more ongoing support.
- Updating recipes and keeping up with staff turnover
- The lack of cooking demonstration and preparation.

no challenges = 10

# ketogenic diet *care*

## Part 1

### Highlights or most helpful parts of the care you receive from your ketogenic diet team:

- Keto chef, keto recipes and videos of cooking the recipes , keto dietitians and nurse, list of products available to use that are most keto friendly.
- They care about our child and our family. They help trouble shoot problems, provide feedback on different keto products, foods, etc.
- Being able to call, state the worry and getting an answer asap
- Regular video and phone call follow ups, quick response to email requests.
- The smallest tweaks can make a big difference. We discover things simply by talking about every day stuff. We feel heard by our team & that matters a lot!
- Very thorough with information
- Helpful response when asking for advice, trusting that we are capable to manage the diet ourselves mainly
- We have a keto chef who helps do private classes for my child when things are hard. We have access to a psychologist that specializes in keto kids.
- My husband did all calculations
- Very responsive. Very encouraging. Lots of great suggestions for diet variety and ways to add healthy fats and reduce carbs. Great team problem-solving.
- providing add ons
- They have a good deal of knowledge about Glut1 and the keto diet which is wonderful. Our team is supportive and encouraging too.
- How to use the meter and how to log recipes for nutritional info
- Suggestion of a new option, a trial I might be able to go through.
- Encouragement and their joy in how diet has helped our son. The continuous weekly support for the first three months was immeasurably helpful. Could NOT have managed without it!!
- a book about the diet

# ketogenic diet *care*



## Part 2

### Highlights or most helpful parts of the care you receive from your ketogenic diet team:

- keep us in care even when our son has aged out, listened to our input after going to the Glut 1 conference and learned new things.
- Necessary bloodwork and paperwork for traveling ect.
- Recipes or helpful shortcuts with the diet
- Our keto team is highly responsive to any of our questions and concerns and they monitor our son closely. When we are unfortunately inpatient they meet with us to work through our needs and provide support while in hospital. They help us with plans/paperwork for school.
- They have provided excellent training and resources for planning food/meals including providing a flexible range for macronutrient targets for meals and a keto calculator for meal planning. They closely monitor his bloodwork so we can adjust supplements as needed. They collaborate with nephrology to ensure renal side effects are minimized. They provide out of the box solutions to help us manage other side effects. They have helped us access programs that provide some of the nutritional supplies we need (Glycosade, MCT, KetoVie).
- Definitely the support with starting out on the keto journey, being at the end if a phone and the info they gave us was invaluable
- Switching to MAD and just being their for questions.
- We are grateful to know that there is at least one Keto team in our state and that they have continued care for our son after he turned 18.
- The well-rounded care we receive. The kindness and willingness to allow us to speak into our care
- Having access to ketocal
- She is always there for us either by phone or email. The first meeting we had she sat with us for 3 hours leading us through each step.
- The knowledge they possess
- Always being available to talk to through email. Them explaining things we don't understand
- The dietician and most definitely the keto calculator
- Very patient and thorough in answering all our questions
- We were provided with a clinical weighing scale
- The approach that they have with the neurologist . During the same visit

## Highlights or most helpful parts of the care you receive from your ketogenic diet team:

- They always promptly respond to email questions.
- Access to the keto calculator to create our own meals and measurements of a variety of foods.
- We were in hospital for the initiation week and a lot of effort was put into making sure we knew about calculations and blood levels and we felt very confident on leaving the hospital. They also gave us a few meals to take home with us.
- Lots of good recipe ideas and flexibility to work with us. Emergency diet collaboration with oncology dietitians during cancer treatment.
- Orientation to diet was overwhelming but practical (tools and resources required to be set up for success). Couldn't survive without the database from the Charlie foundation. Advice re supplements required based on bloodwork results. Our province provides funding for special products. Would be helpful to have access to products for diets with a lower ratio than 4:1.
- We know how many carbs she can have and how to count them
- Very understanding and helpful with answering our questions or concerns
- They are responsive and accommodating. They understand my daughter's needs.
- The preparation meetings before we started the diet were so helpful. They were accessible to us on a daily basis and so supportive in the beginning when things were all new to us. This support has continued but as we've become more use to the diet, we've needed their daily/weekly support less and less.
- Support and encouragement, problem solving about ingredients and substitutions, helping me understand labels and nutrition, providing guidance with keto calculator
- New recipes
- They had an in person educational meeting of the keto diet and glucose checking. Meeting included handouts of the diet, how to figure net carbs. Diet goals broken down with snack and meal allotment. Also gave a handouts of recipes, how to handle sick days, and letters to caregivers, TSA, and hospitals.
- The support and care is always available within reason.



# Keto *Care* PROJECT

Not all patients have access to ketogenic clinics or high quality ketogenic diet support.

The goal of the Keto Care Project is to provide expert ketogenic dietitian care to underserved patients and families.

Baszucki Group - Metabolic Mind

# by the *numbers*

**24** patients and families receiving at least one consultation

**3** countries

**12** states

**3** expert dietitians providing virtual consultations



**Keto** *Care*  
PROJECT

**age distributions**

**age range:** 4 to 35 years

**average age:** 16 years

# dietitian *feedback*

any recurring themes or challenges?

**NO!**

useful tips or strategies?

- increasing resources available to families
- additional support options
- more recipes
- ketodietcalculator access
- make meals as similar for person w/ Glut1 as rest of family  
*i.e. protein and vegetable are the same*

Some of the families want to provide the least restrictive diet as possible and aren't willing to try stricter diet to get better results.



**Keto** *Care*  
PROJECT



*new* resource

# KETOGENIC DIETS AND **Glut1 Deficiency**

GLUCOSE TRANSPORTER TYPE 1 DEFICIENCY SYNDROME

Medically supervised ketogenic diets are the current standard of care treatment for Glut1 Deficiency, but there are special considerations for their use in this disease.





Thank you!