



www.G1DFoundation.org

Glut1 Deficiency
COLLECTIVE
VOICES
Project



COLLECTIVE VOICES PROJECT

Goals

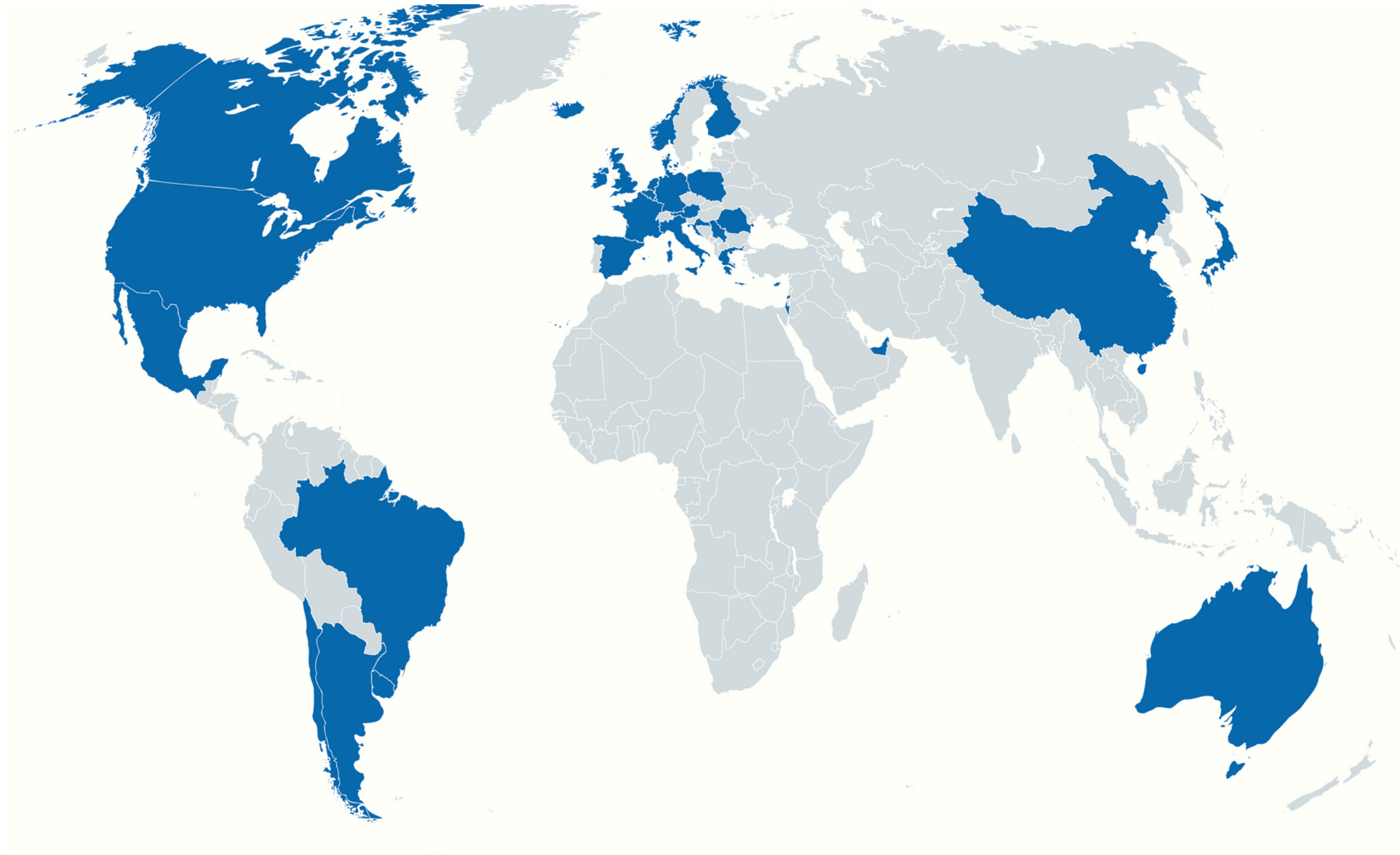
- **better understand the patient and family experience**
- **better define the range of symptoms**
- **identify gaps in treatment and patient care**
- **identify gaps in knowledge and understanding of this disease**
- **better understand the burdens of this disease on the patient and the family**
- **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



260 responses

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



Demographics

female 57% **male 43%** n=242

caregiver or parent 95% **patient 5%** n=250

age and distribution



youngest
infant



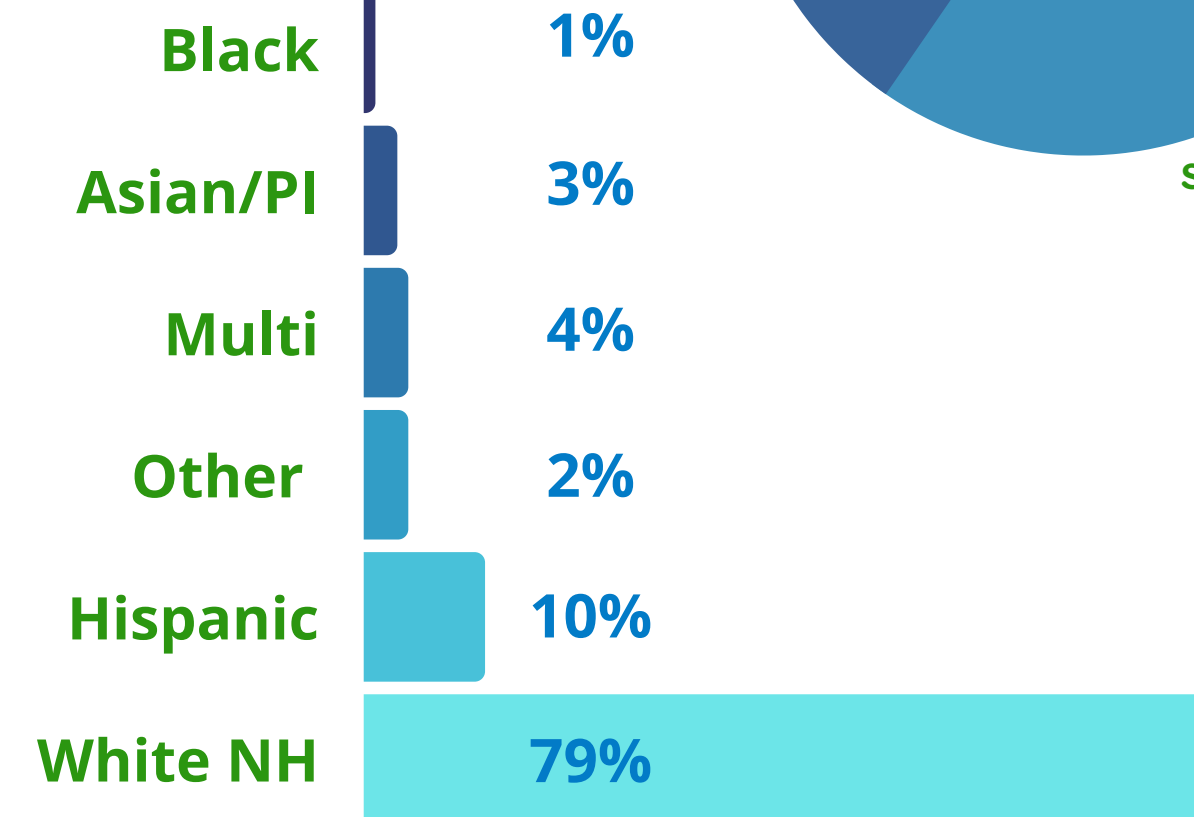
average
22 years



oldest
53 years

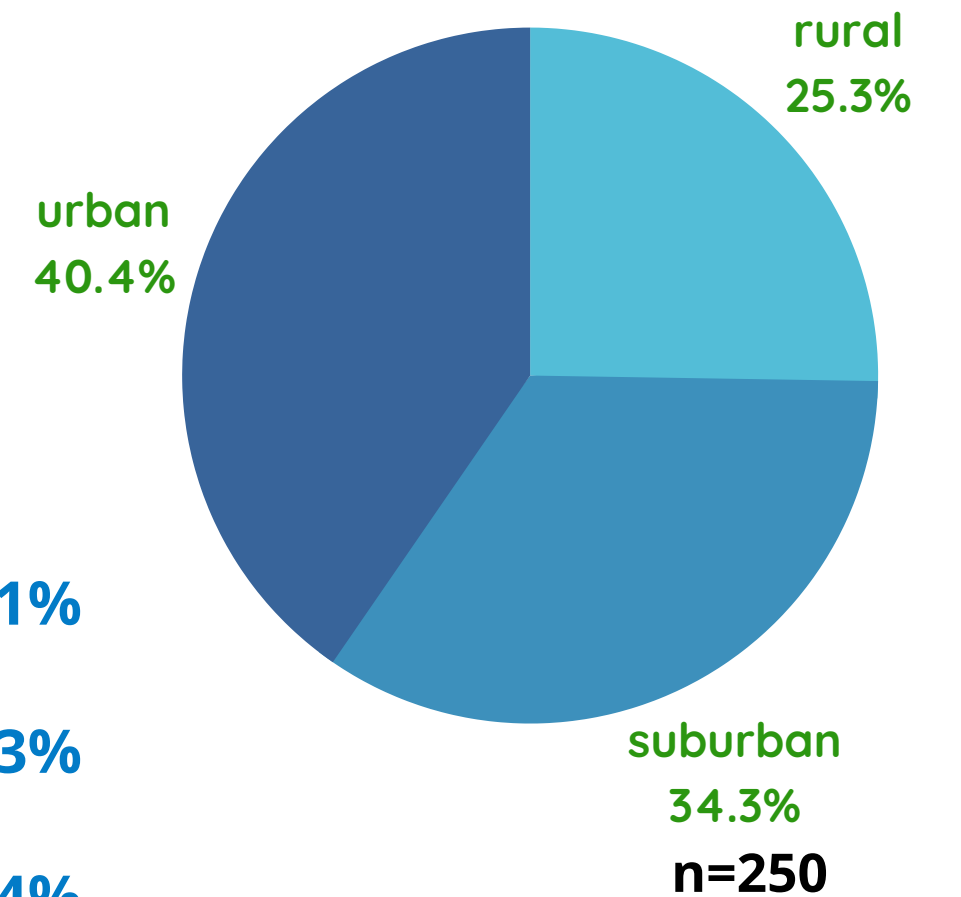
birth to 6 yr	23%
7-12 years	37%
13-17 years	17%
18-53 years	23%

n=238



identity groups

n=242



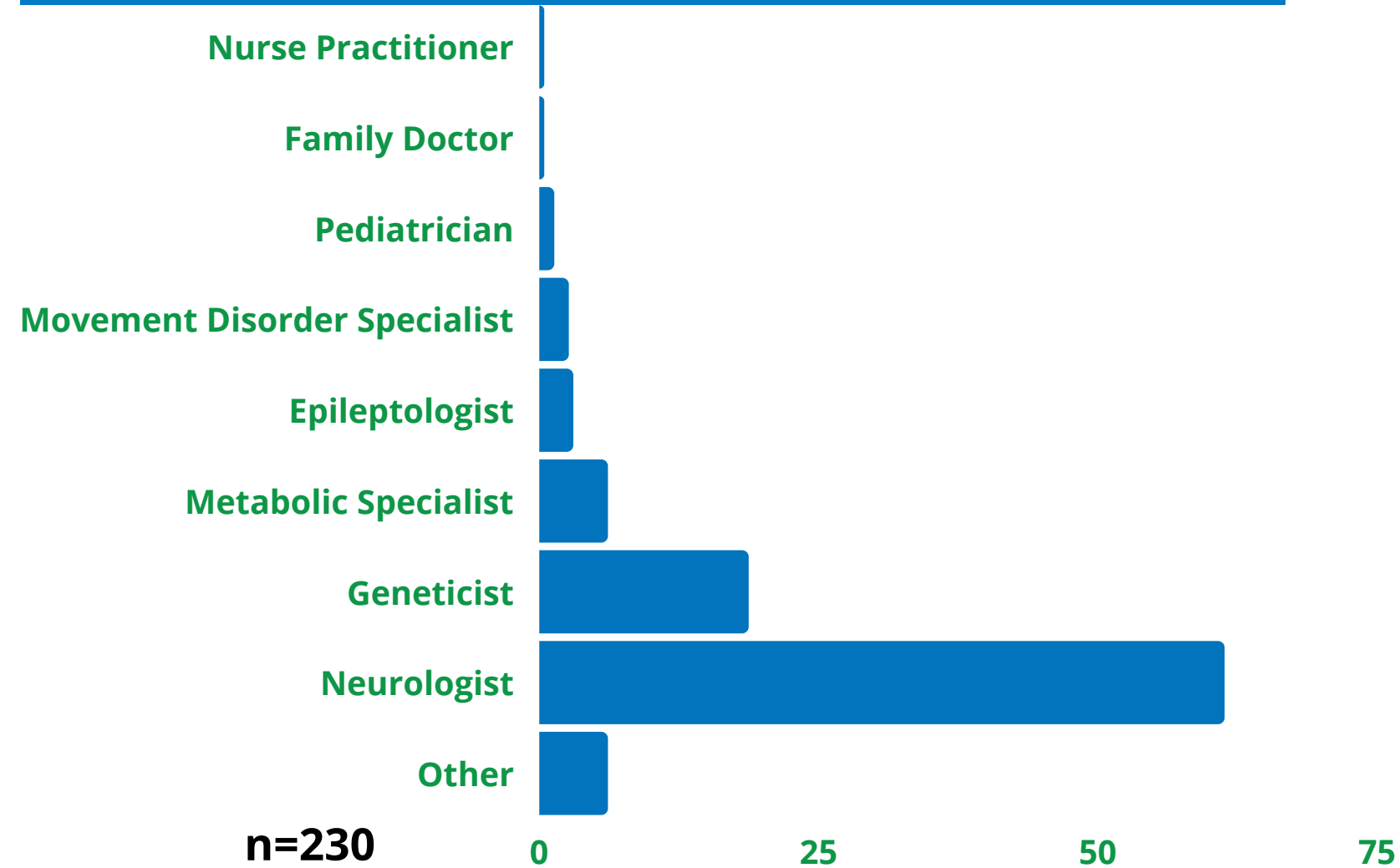
Diagnosis

average # of physicians
seen to get diagnosis

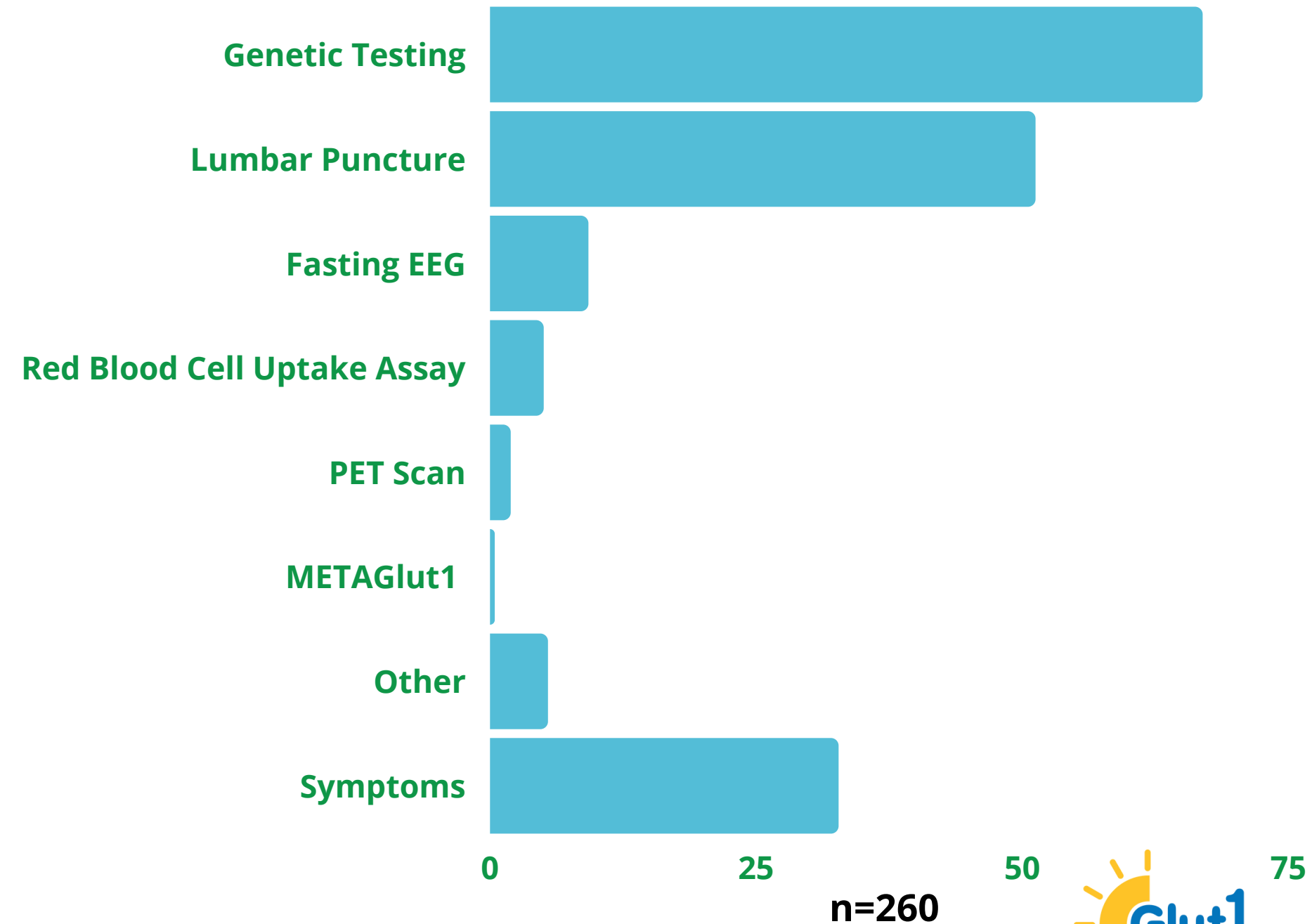
8

n=230

who made the diagnosis?



diagnostic tools used



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Diagnosis

additional family members

10%

n=230



first symptom to diagnosis

shortest 1.5 weeks
longest 34 years
average 2.8 years

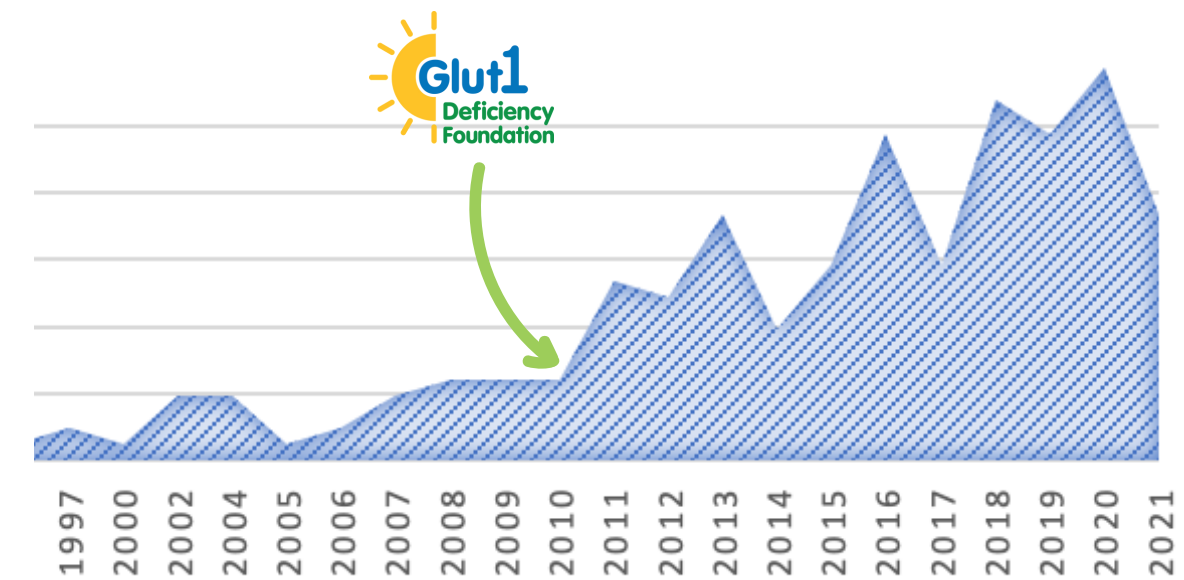
n=203

0 to 11 months	18%
1-4 years	44%
5-9 years	22%
10-14 years	9%
15-35 years	7%

age at diagnosis

youngest 12 days
oldest 39 years
average 6 years

year of diagnosis



n=227



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Diagnosis

biggest obstacles to diagnosis

lack of knowledge by healthcare team

treated symptoms rather than looking for cause

misdiagnosed

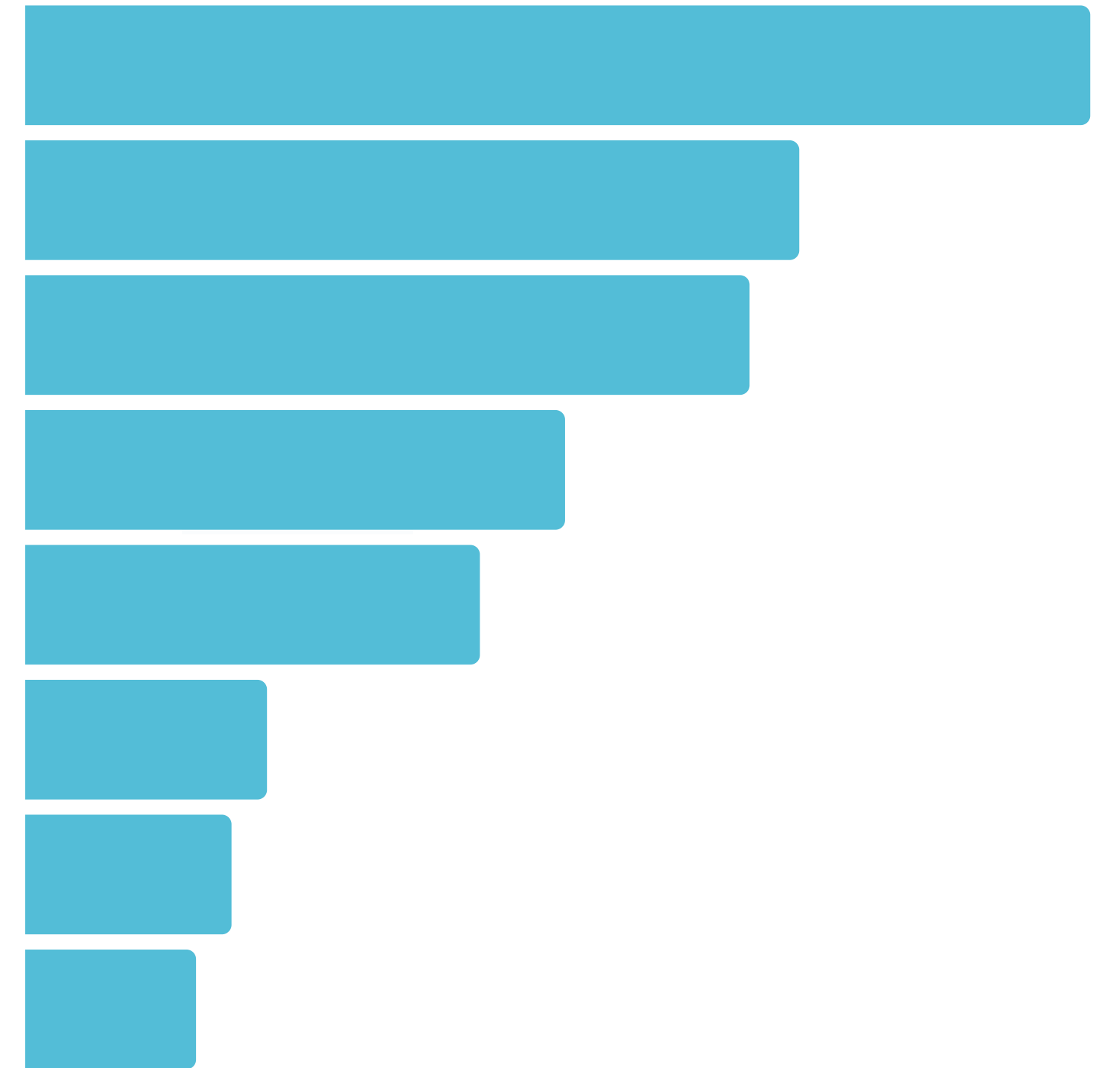
didn't listen to family concerns

misinterpreted or missed test results

other

lack of access to specialty care/testing

no obstacles



n=260

0

50

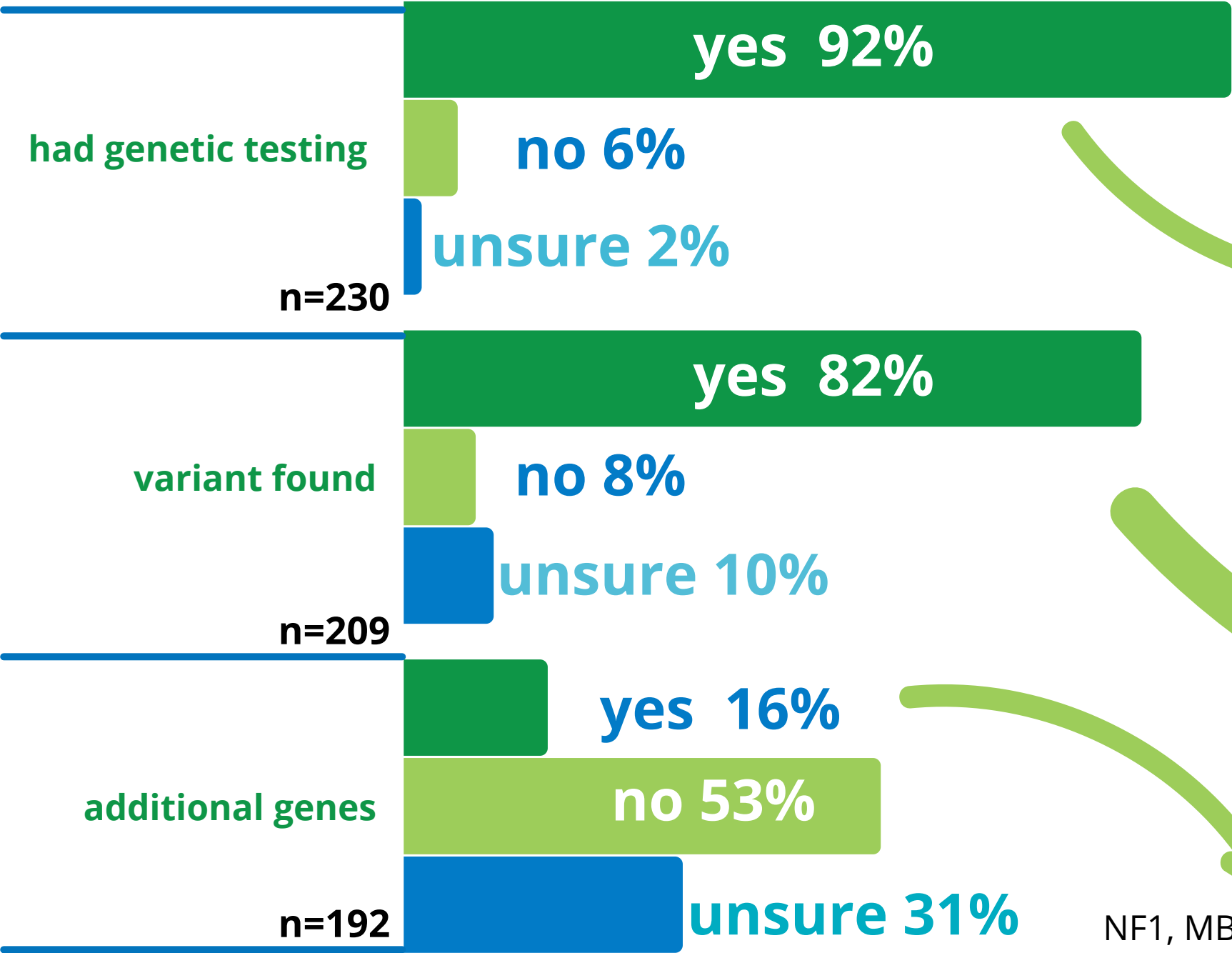
100

150



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Genetics



type of testing

	%
unsure	30
single gene	20
epilepsy panel	16
WES	12
WGS	10
movement panel	6
other	6

type of variant

	%
unsure	49
other	13
missense	13
whole deletion	9
small deletion	8
splice site	4
nonsense	4
duplication	1

NF1, MBD5, TUBA1A, HCN4, SCN4A, SHH, CHD2 -
CHRNA2, GPR98, TANGO2, USP7, IFIH1, PIGG, EPM2A,
MTOR, SCN1A, BRAT1, BRCA1, CENPJ



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

roughly 1/2 eligible to transition to
adult care have done so

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

worked with a dietitian

90%

quality of care rating average
(0-10)

7

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

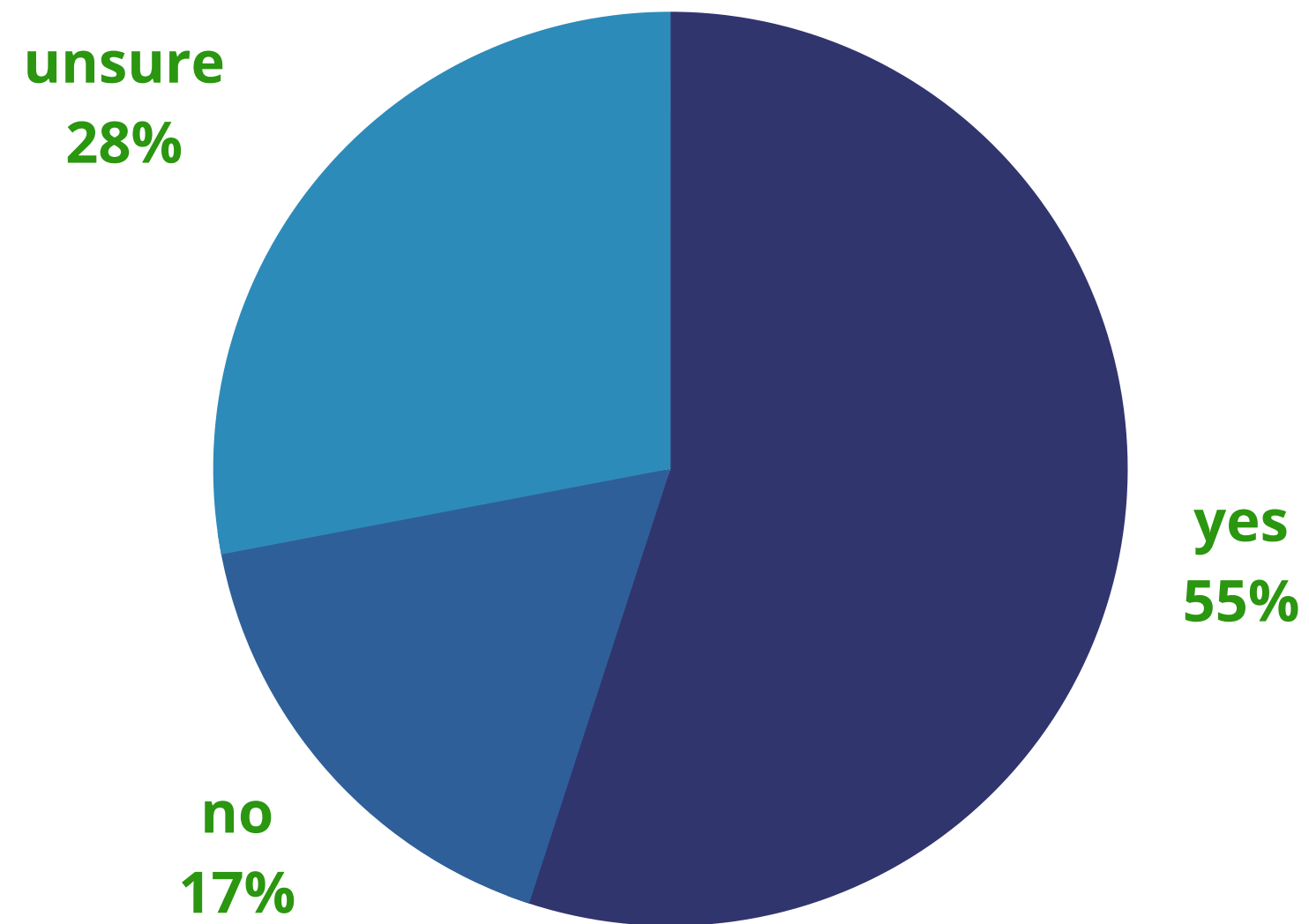


n=209

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Medical Care

are your medical professionals
proactive in learning about Glut1
Deficiency



n=209

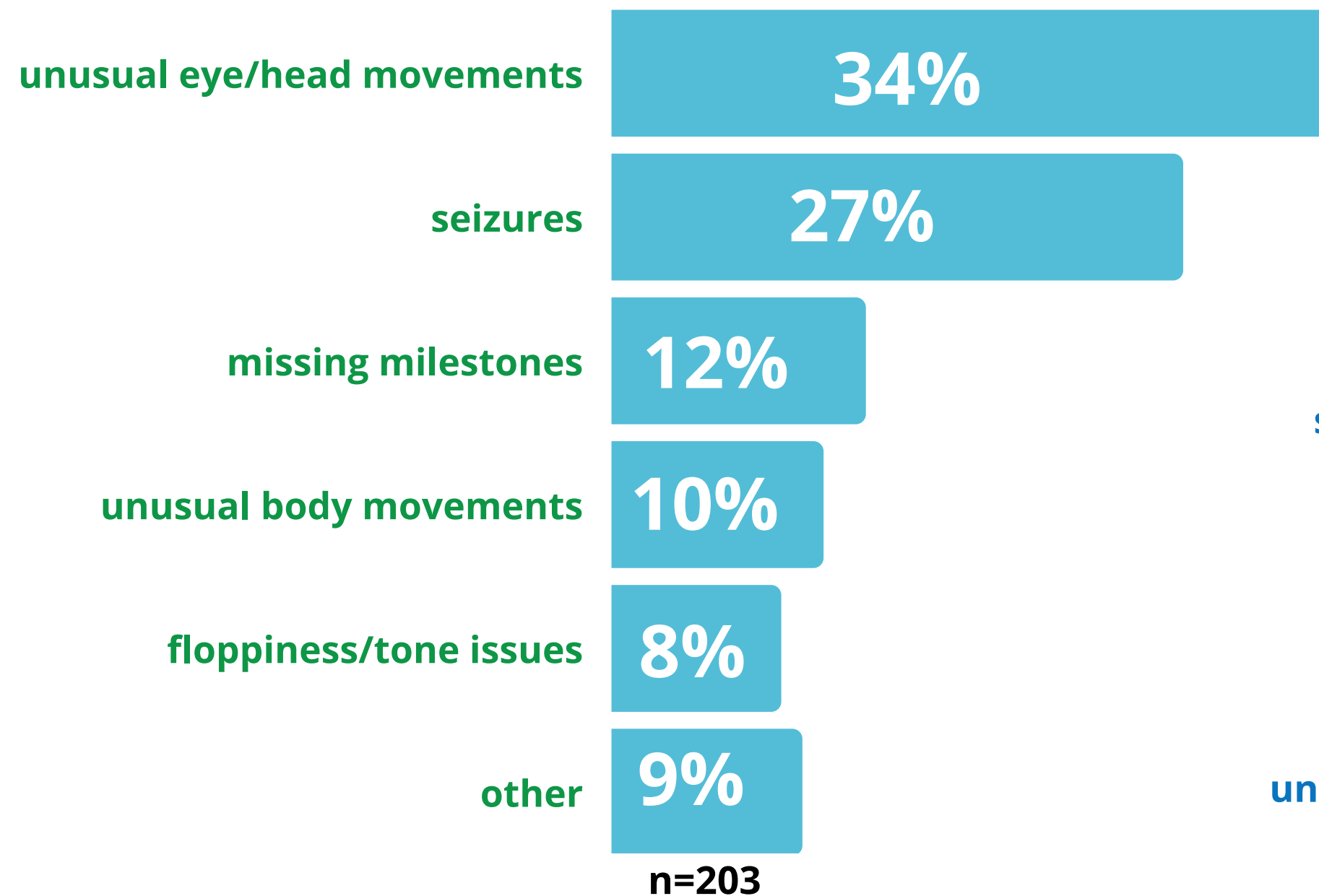
68% report their healthcare
providers have other
Glut1 patients

71% of those feel it
results in better care

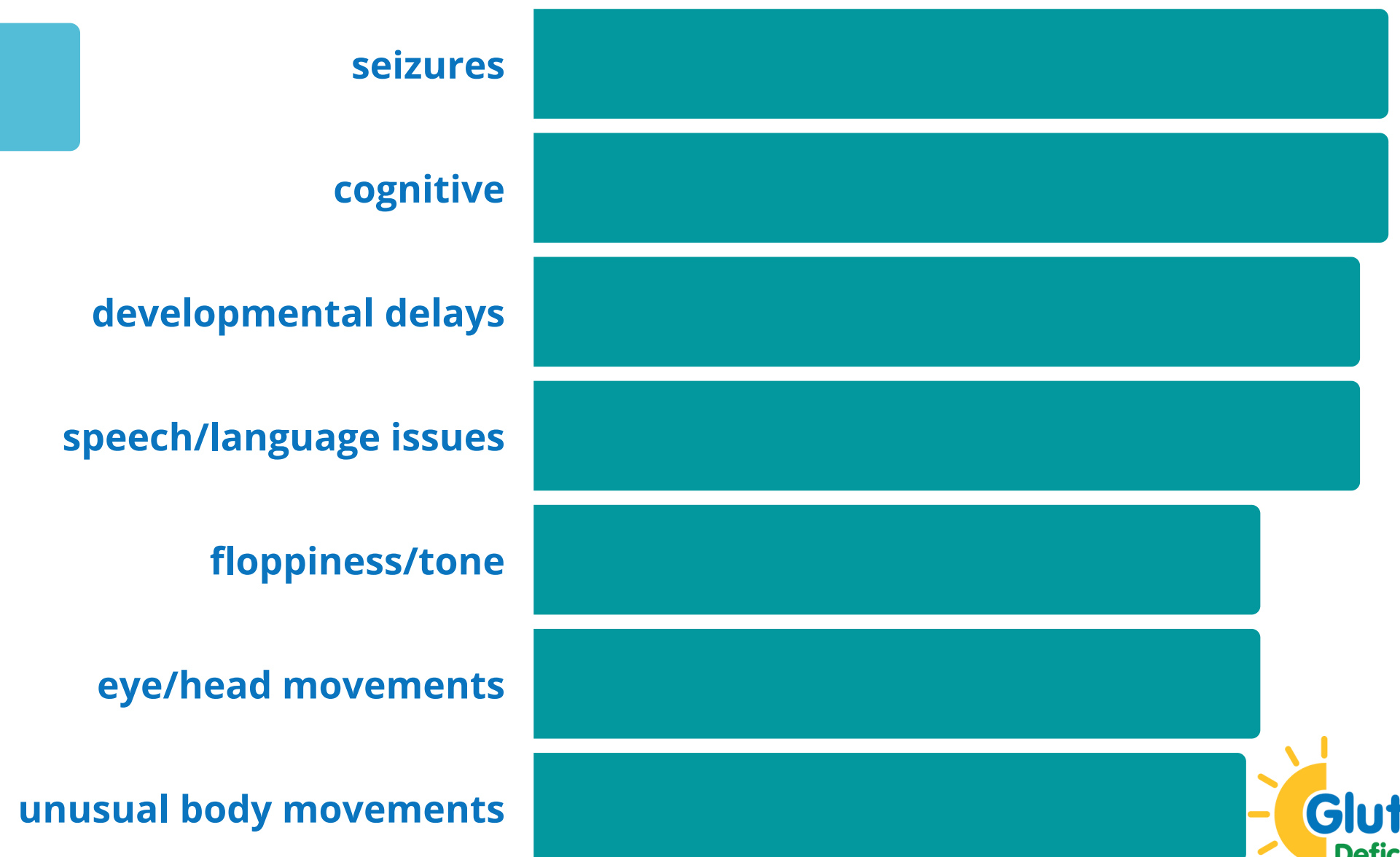


Symptoms

first symptom

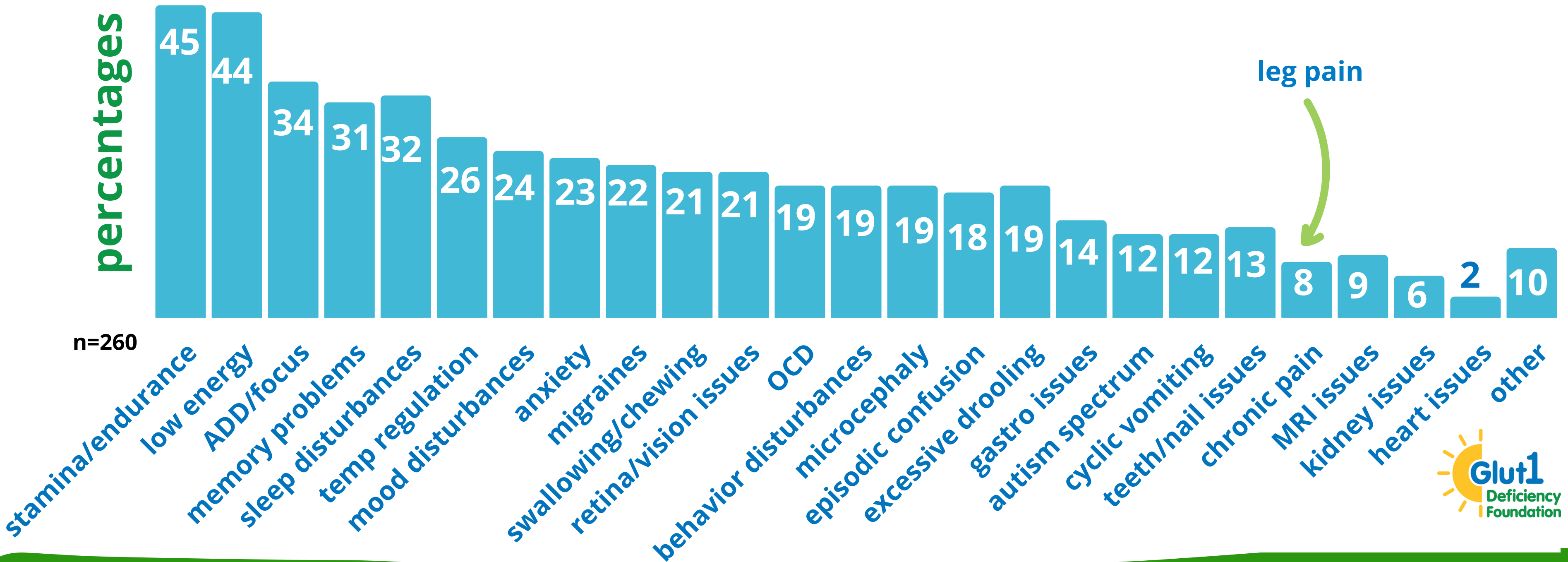


"classical" symptoms

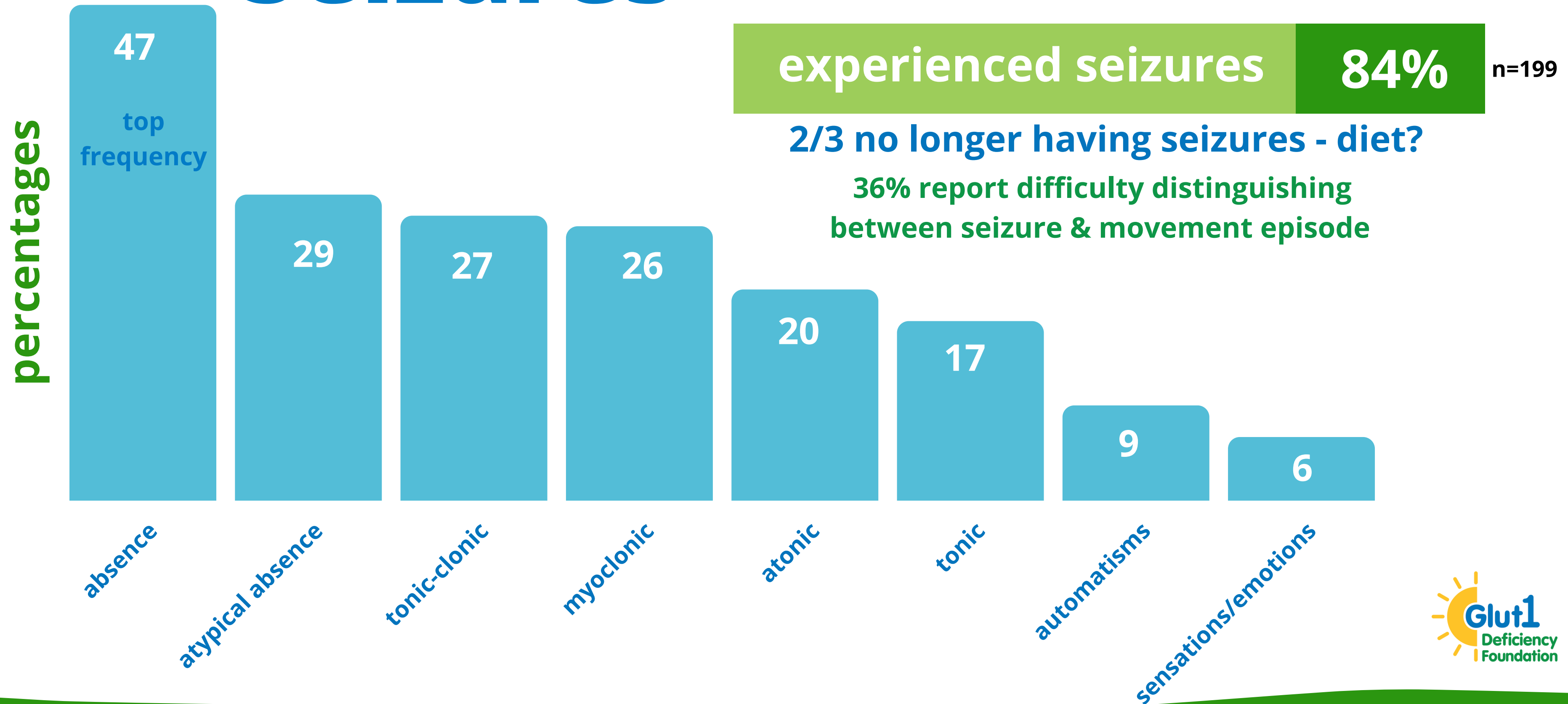


Symptoms

other symptoms



Seizures



Movements

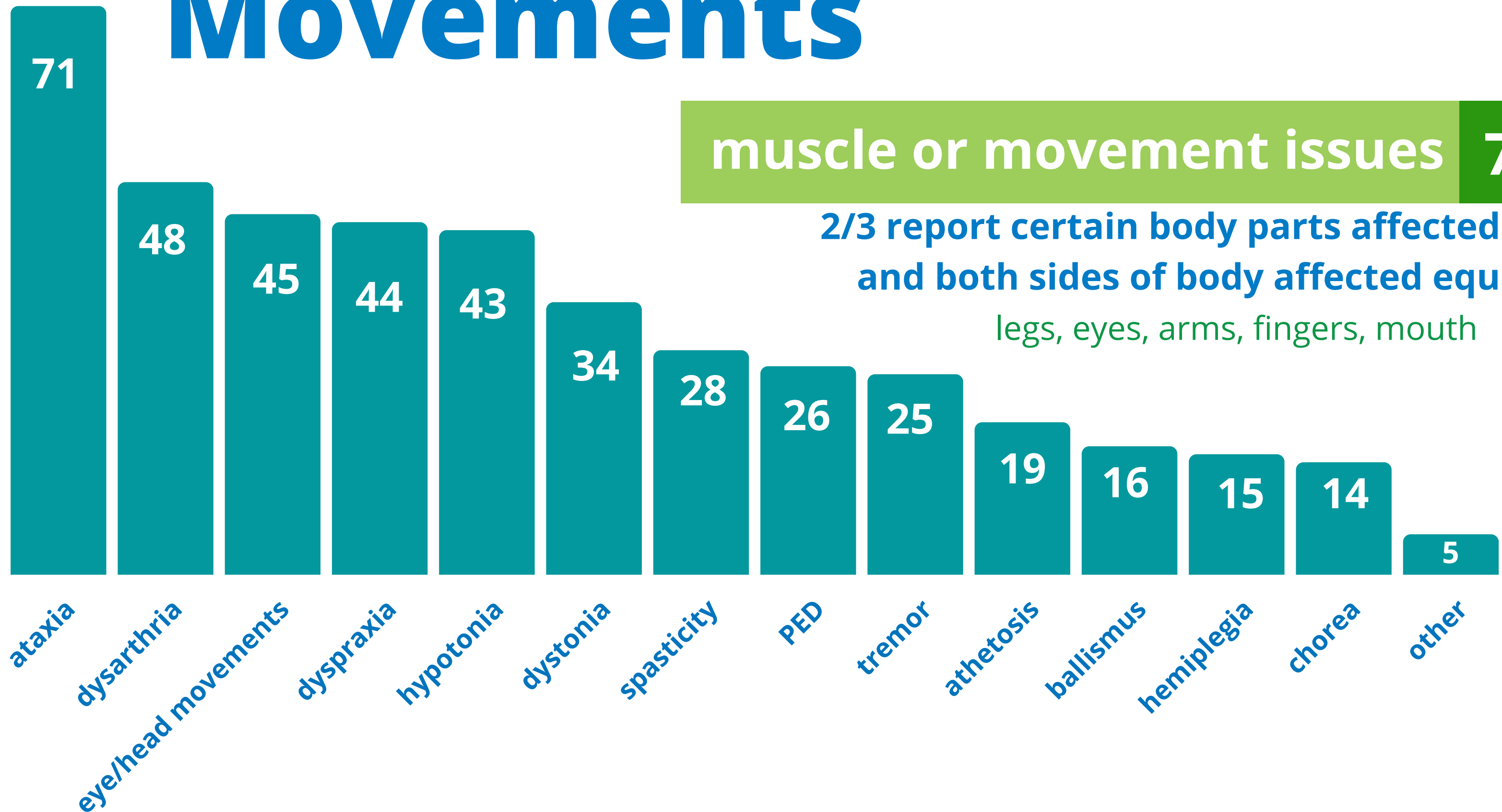
percentages

muscle or movement issues **77%**

n=202

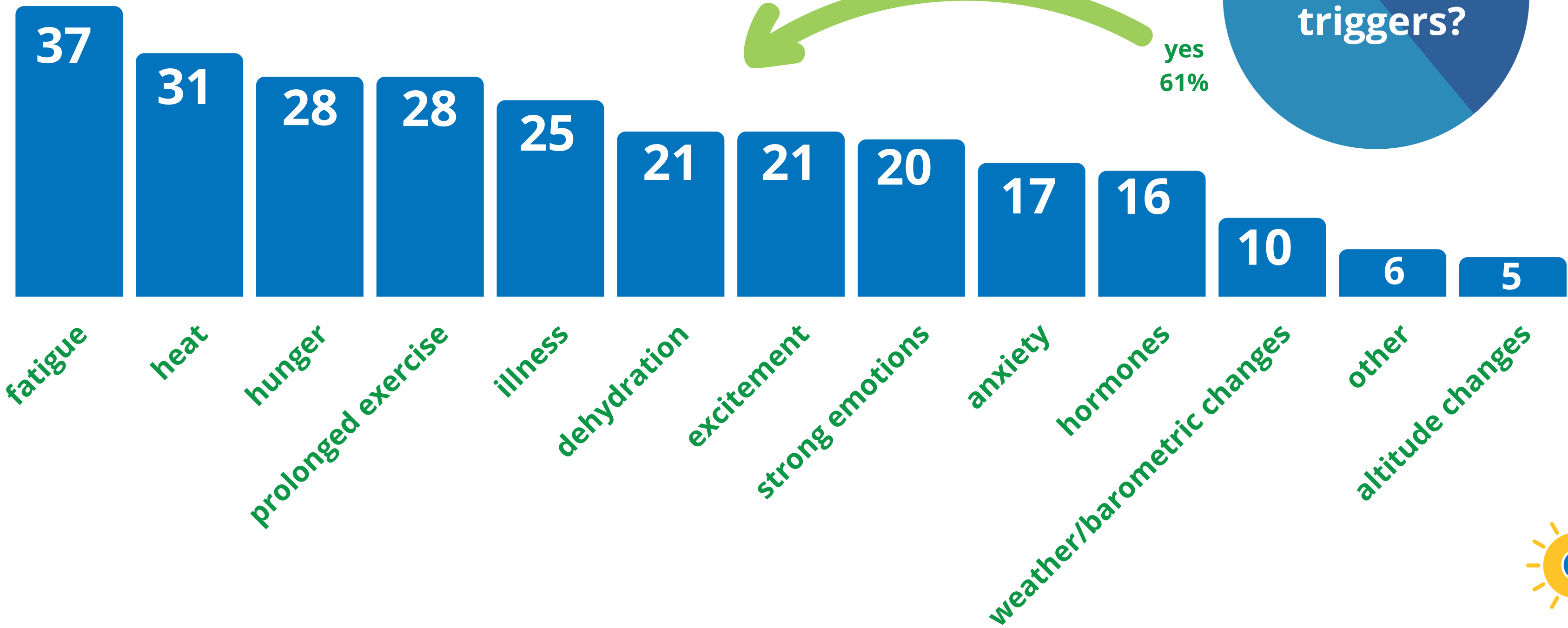
2/3 report certain body parts affected most
and both sides of body affected equally

legs, eyes, arms, fingers, mouth



Symptom Triggers

percentages



Other Symptoms & Behaviors

n=203

high pain threshold 44%

sensory seeking 27%

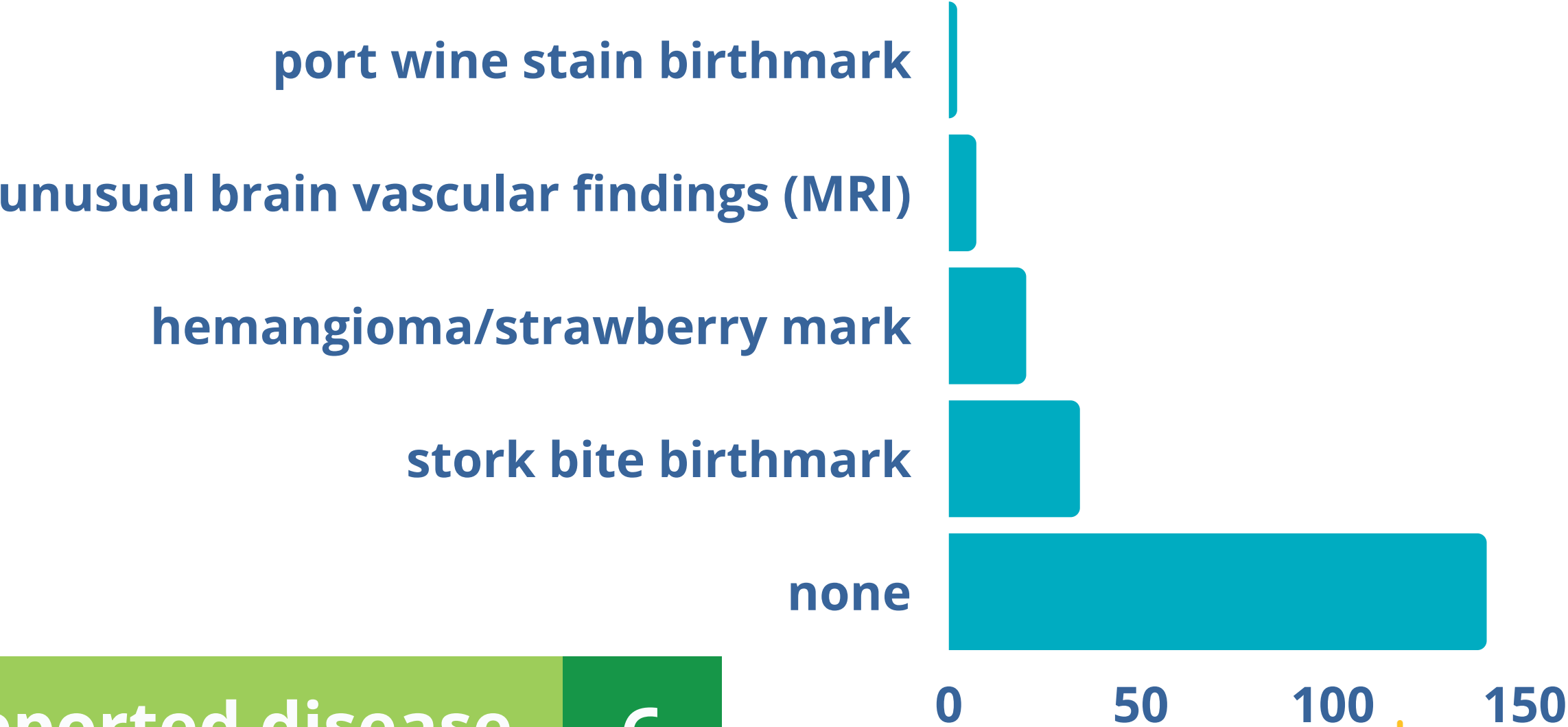
sensory avoidance 19%

self-stimulatory 25%

occasionally 66%
frequently 33%

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

vascular issues

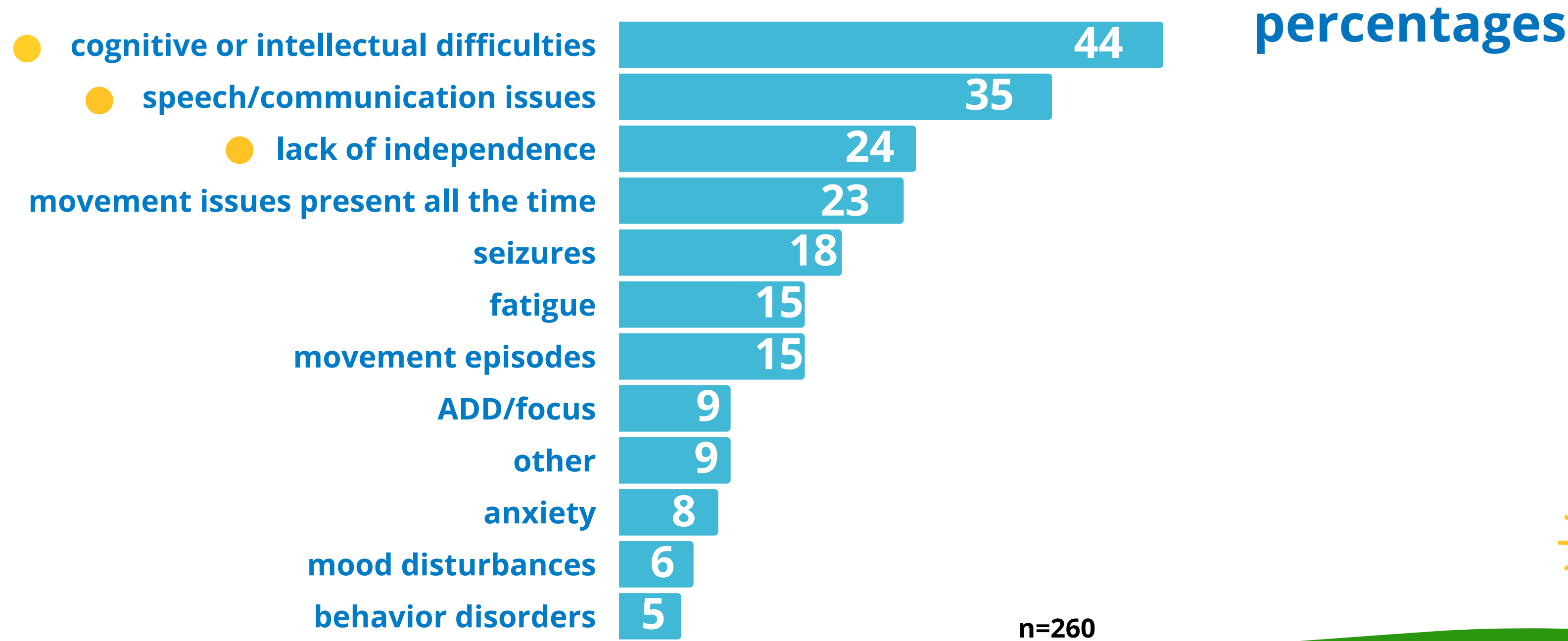


n=211



Symptoms & Quality of Life

top 3 symptoms negatively impacting quality of life

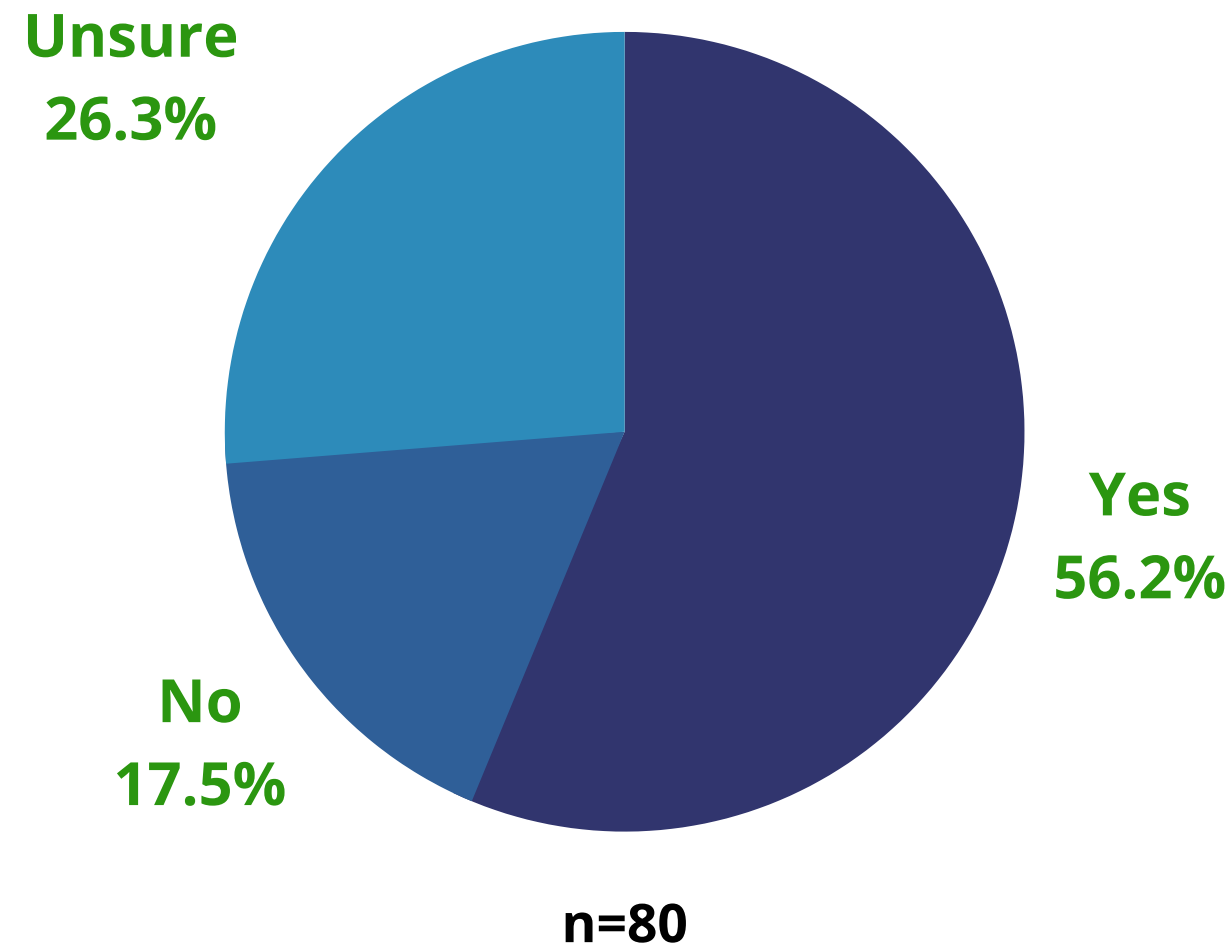


Puberty

41% have experienced or are currently in puberty

n=202

Did symptoms change in puberty?



How did symptoms change?

improved
none

stayed same
cognitive
speech/language

worsened
movements
stamina/energy
seizures
anxiety

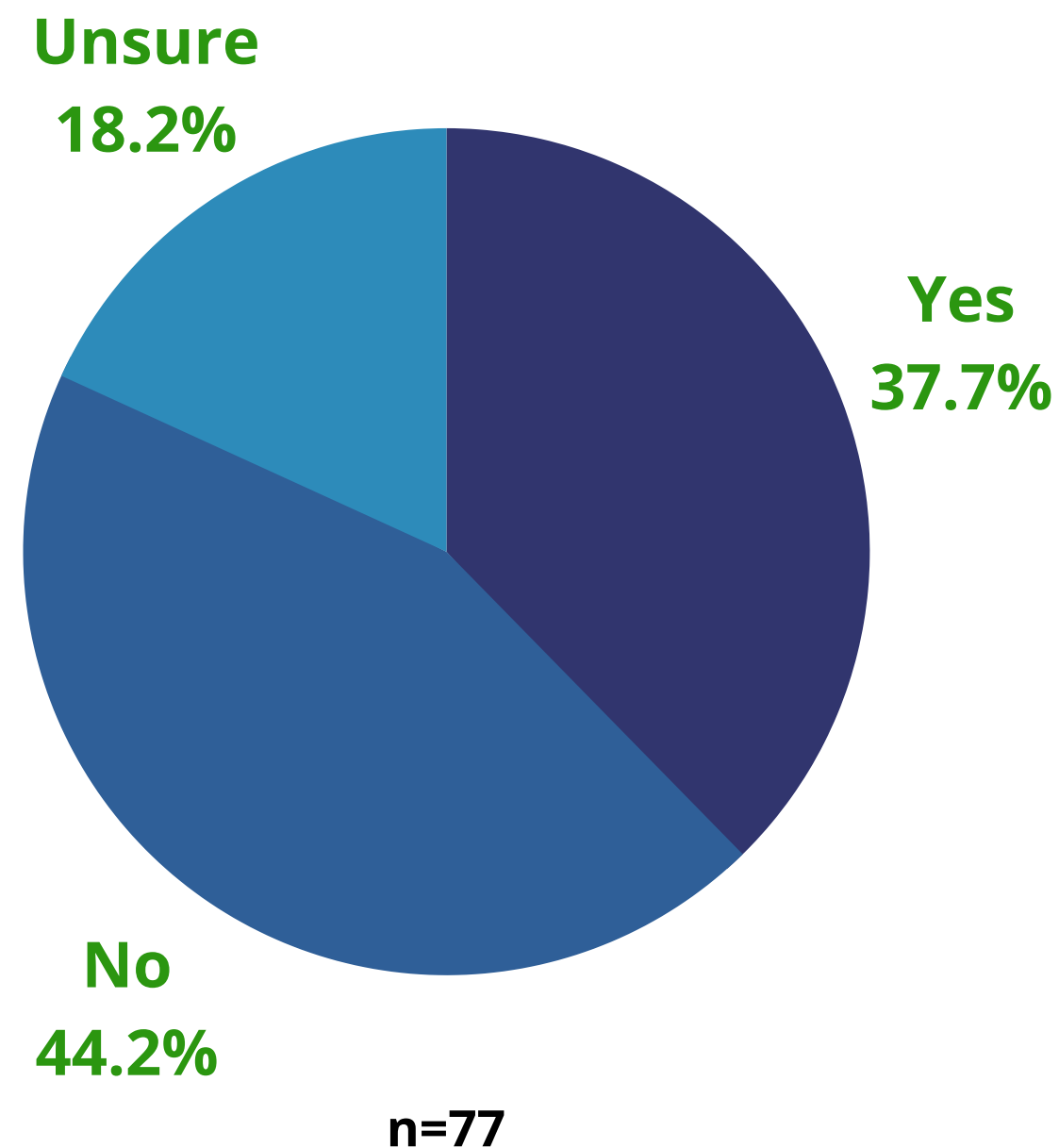
Did treatments need to change?
45% yes

How did treatment need to change?
diet changes 49%
new meds 36%
new therapies 8%
other 8%

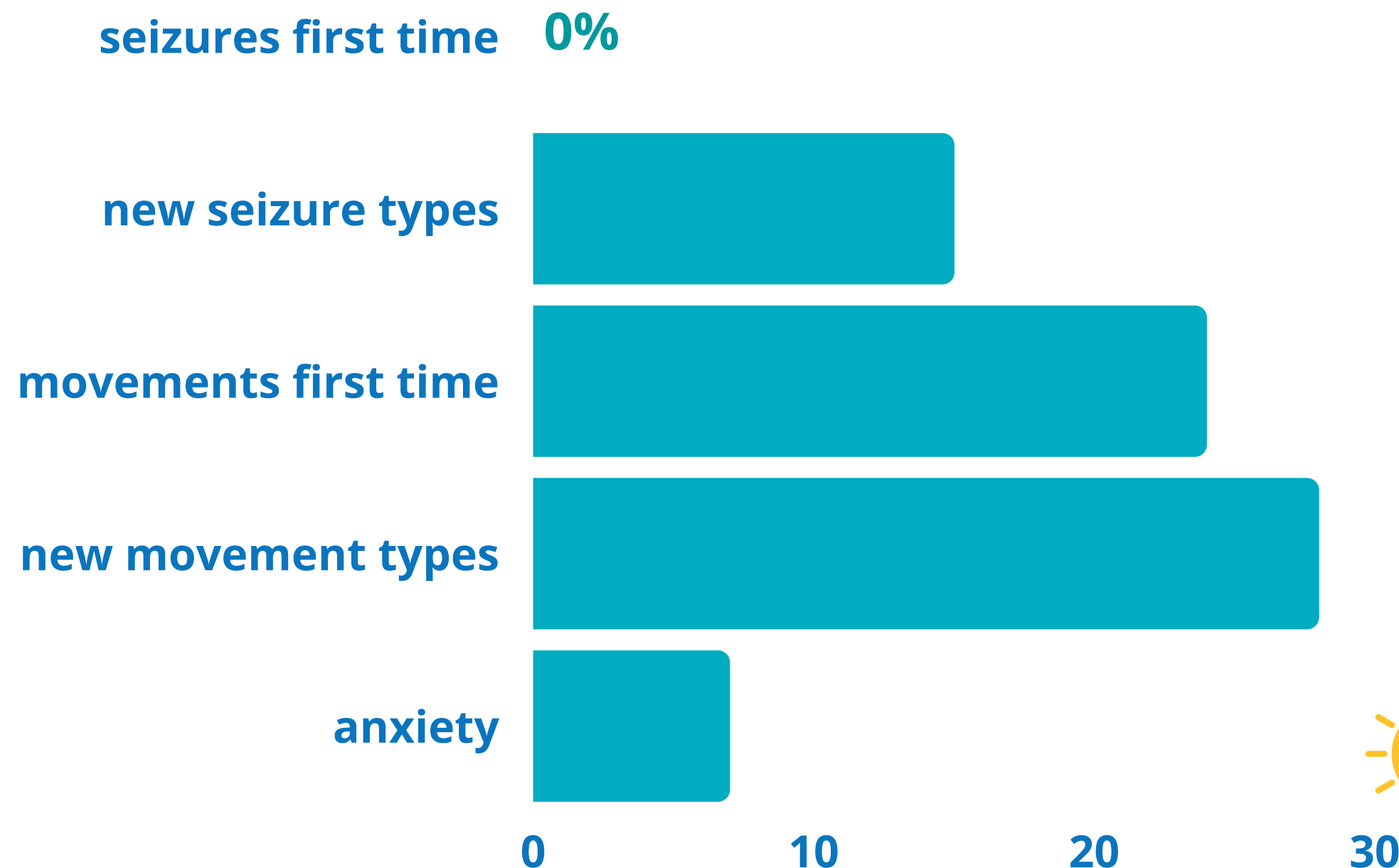


Puberty

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



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

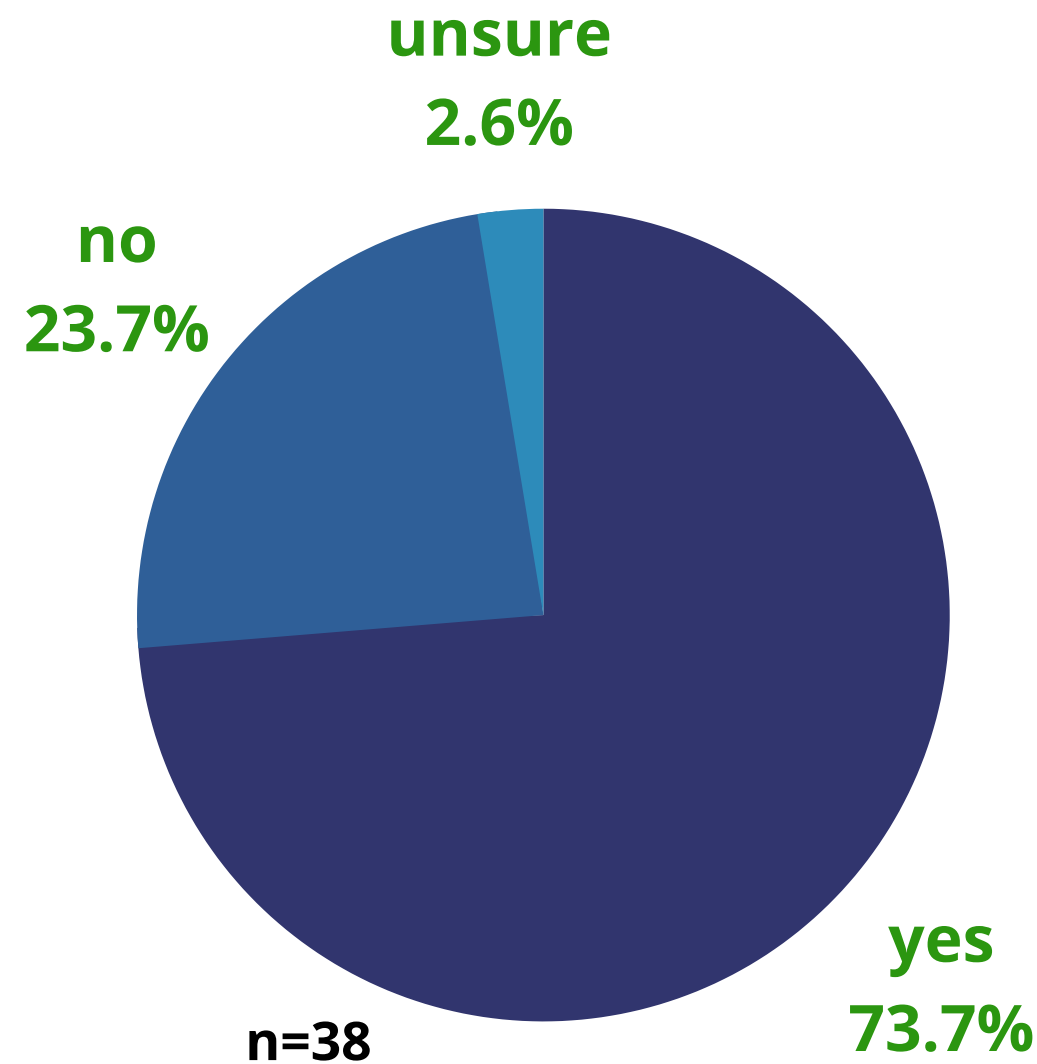


Adulthood

20% of responses

n=191

Did symptoms change
in adulthood?



n=38

quality of life in adulthood

improved

45%

same

34%

worsened

21%

n=38

How did symptoms change?

improved
seizures

stayed same

cognitive
speech/language
memory

worsened
movements
stamina/energy

on KDT

57%

able to manage
diet independently

20%

10% have children of own

75% of children also have Glut1 Deficiency

How do symptoms compare?

similar

67%

more severe

33%

less severe

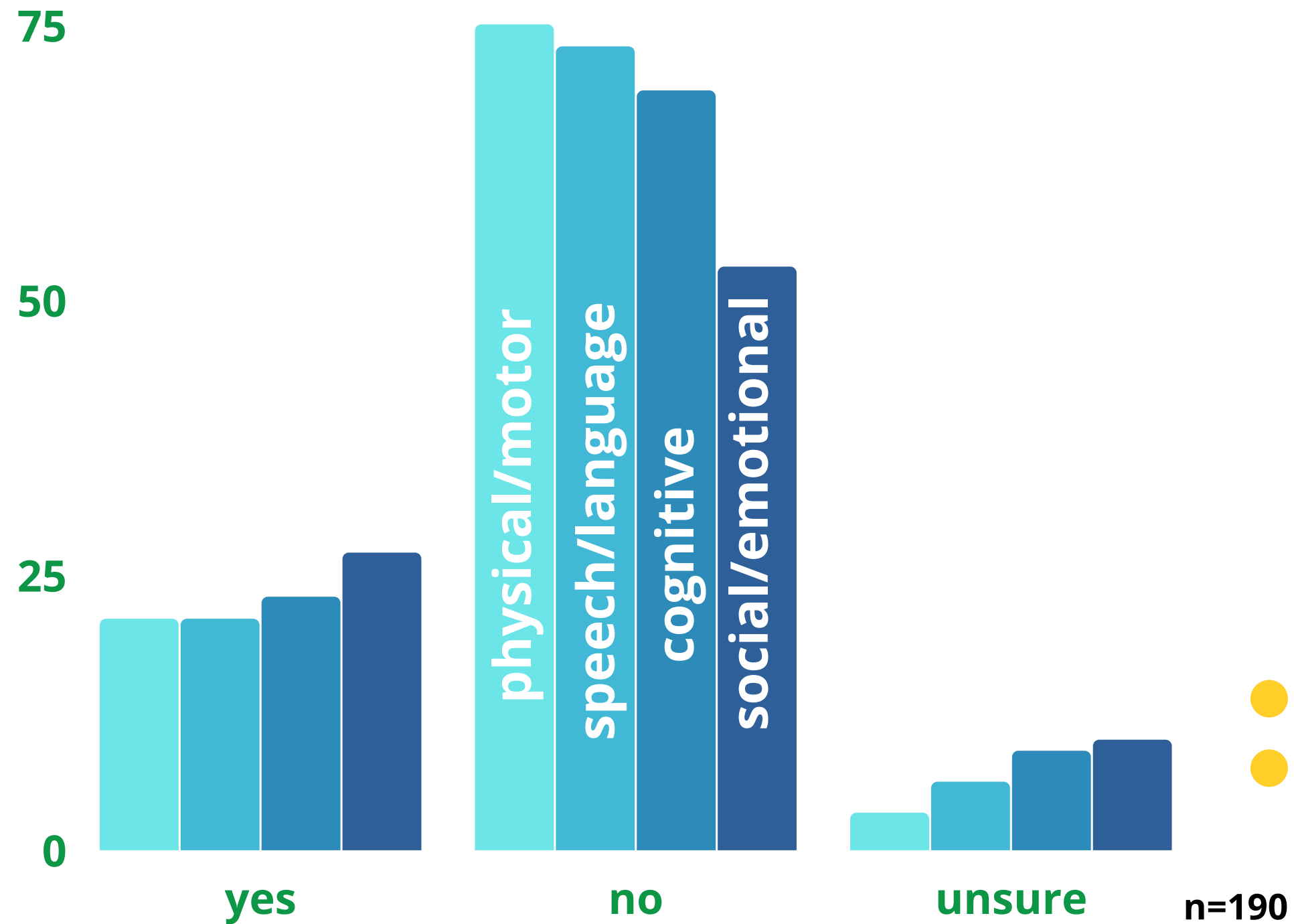
0%



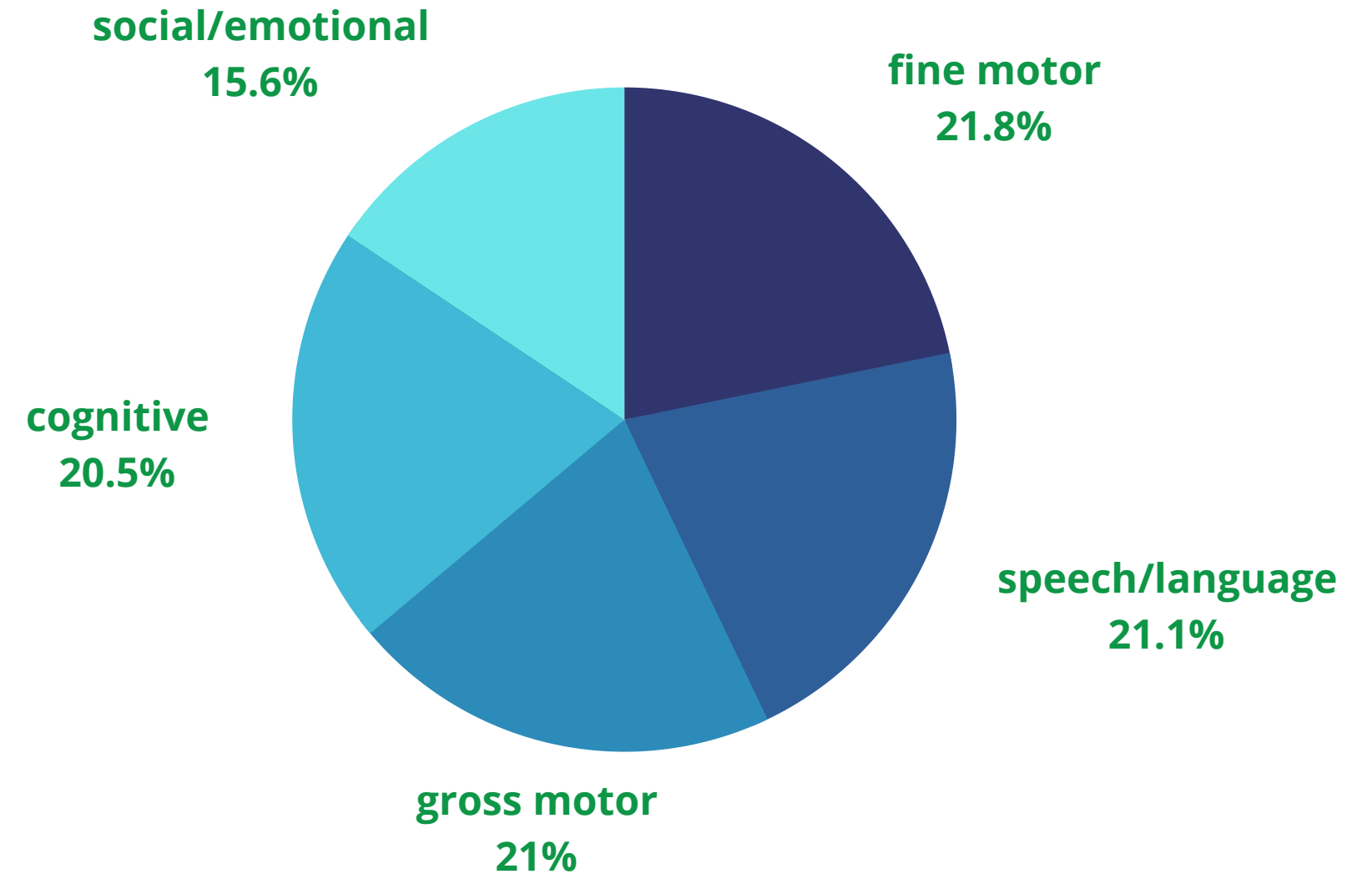
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Development

meet developmental milestones on time?



experience challenges in any of these developmental domains?



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



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% age?

average age for walking

2 years

oldest: 9 years
youngest: 8 months



n=190

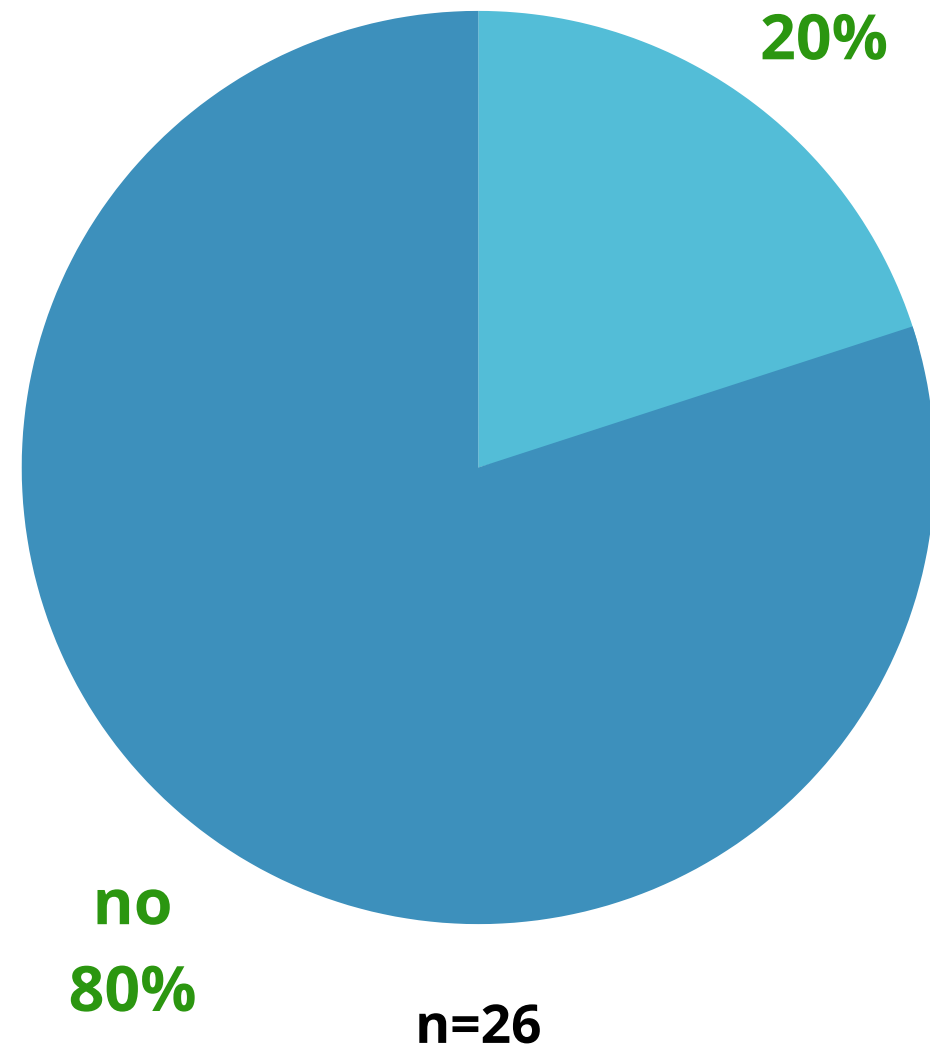
COLLECTIVE VOICES PROJECT

Development

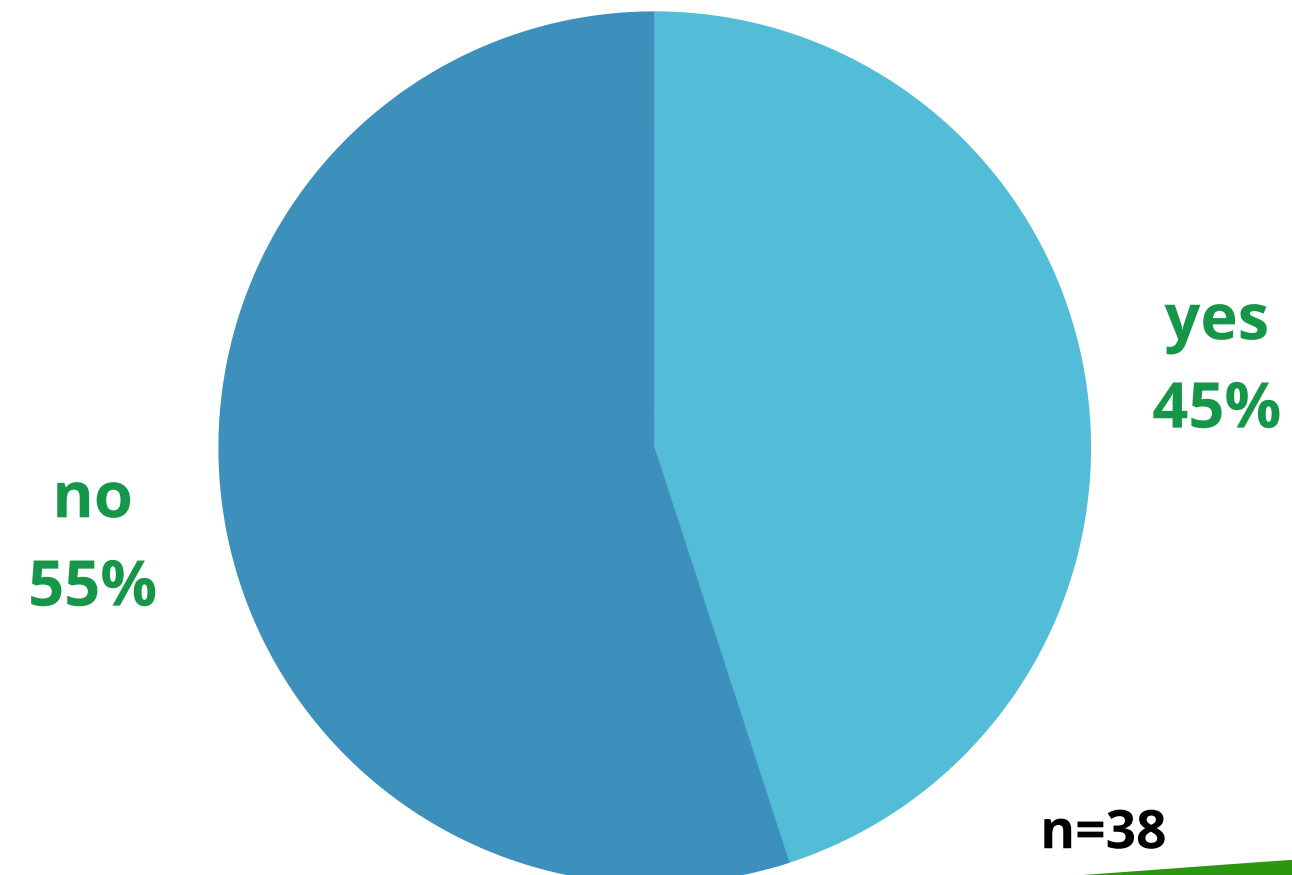
early intervention services **73%**

n=190

adults able to manage
KDT independently



adults able to manage
medications independently



toileting

reached on time 30%

reached but delayed **51%**

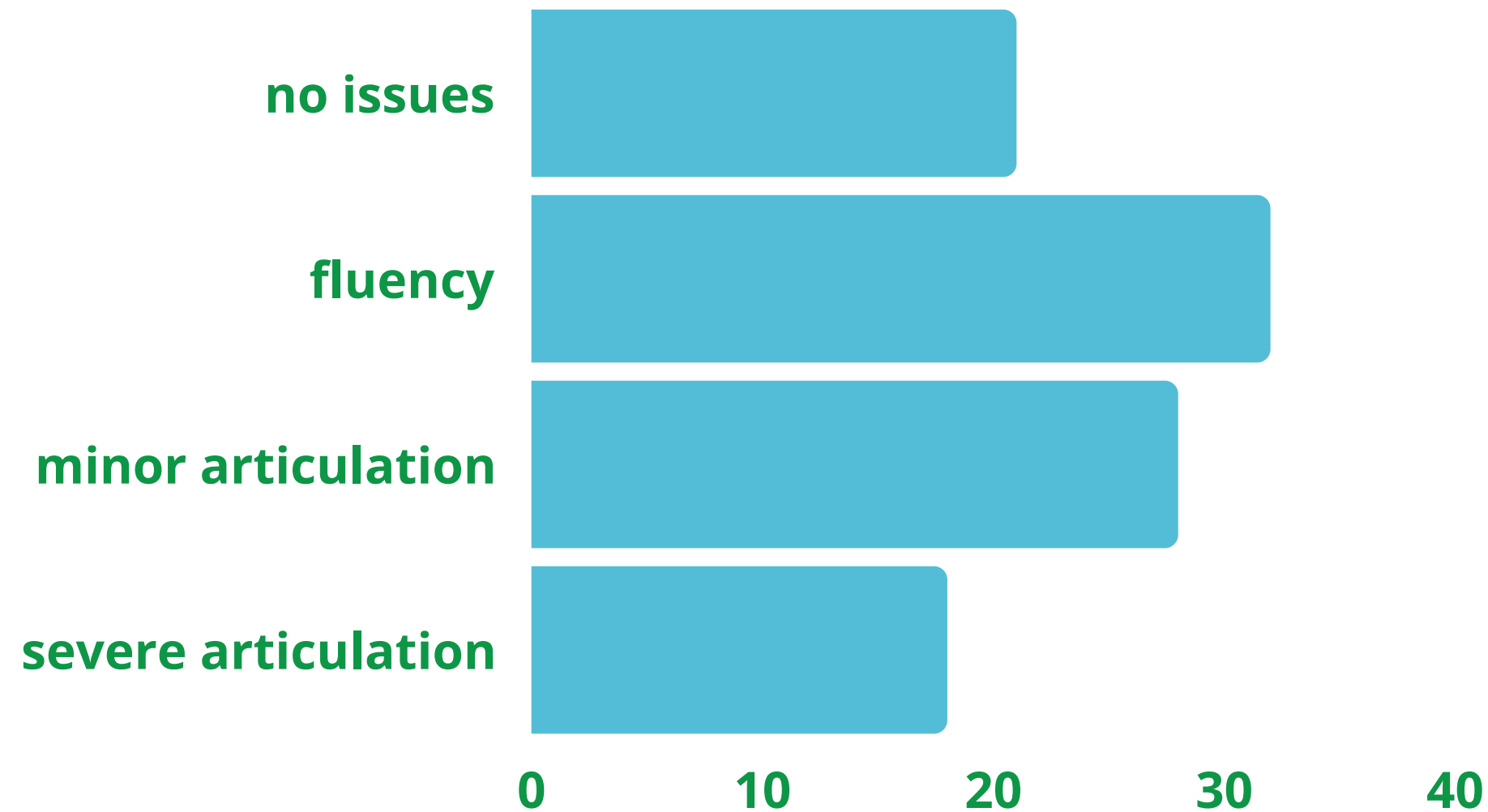
n=190



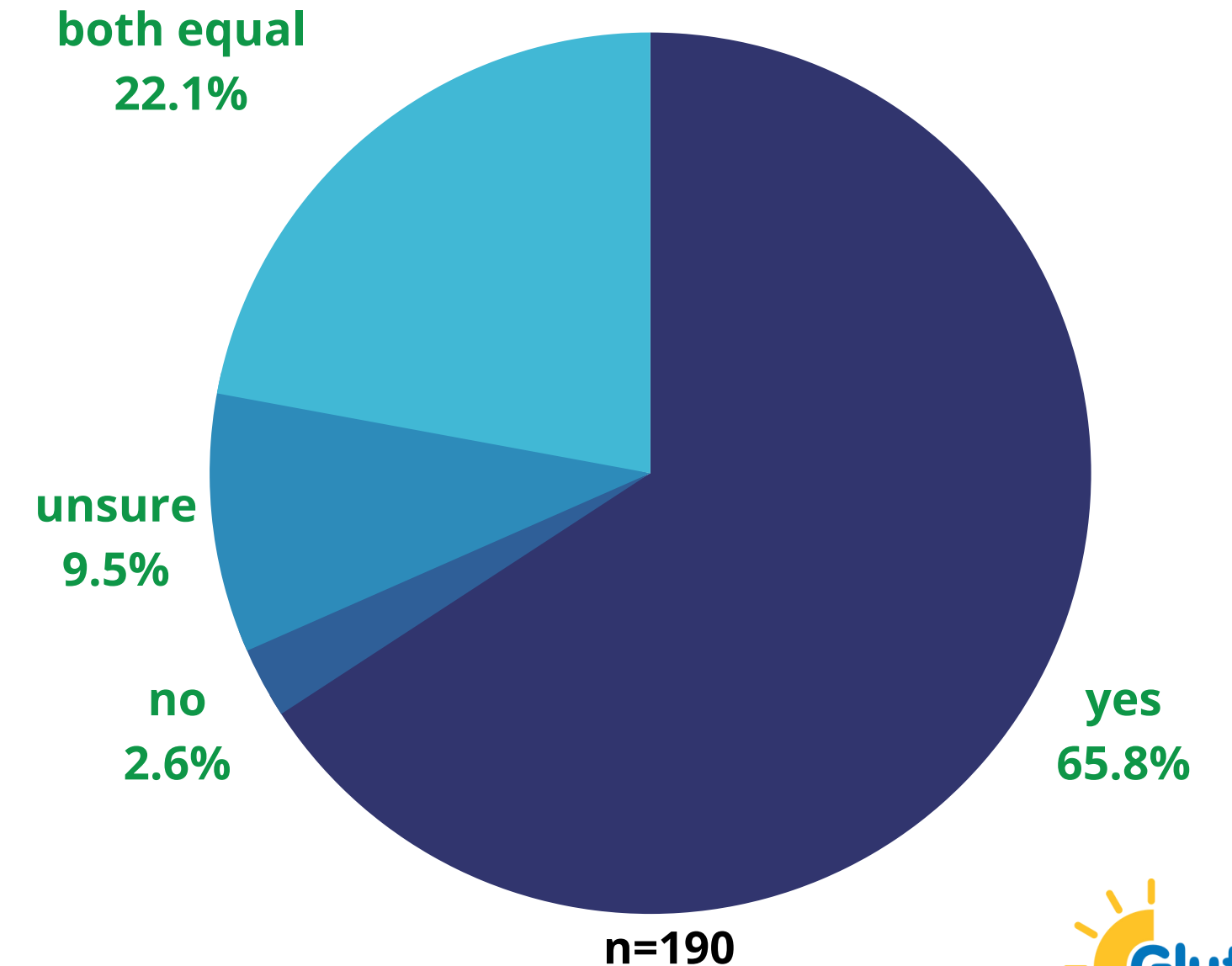
Speech & Language

average age to speak well enough to communicate 3.5 years

able to communicate using own voice 84% n=190



receptive language better than expressive?



- half feel speech issues make them appear less capable/intelligent than they are
- 2/3 experience frustrations around communication issues

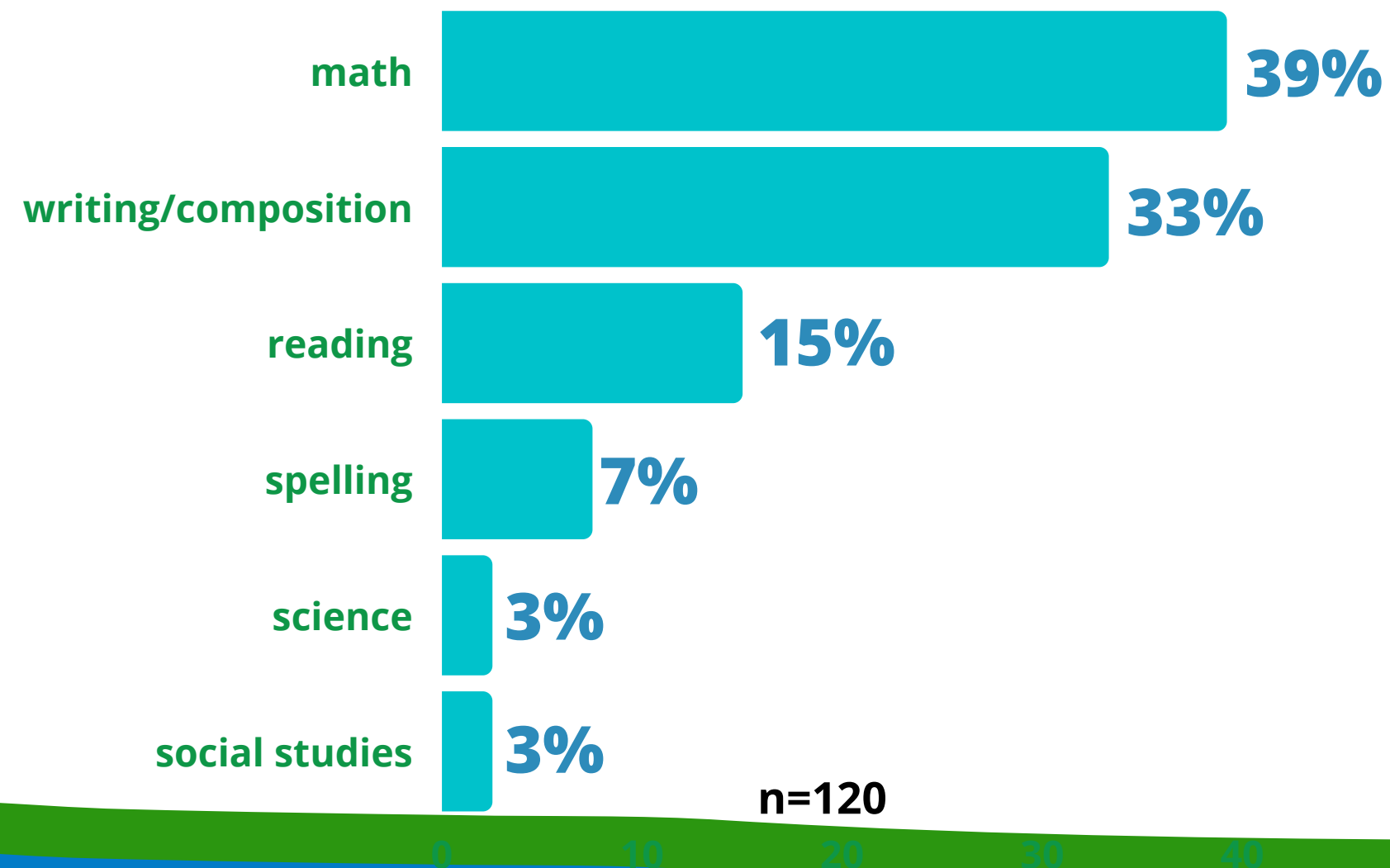
Cognitive

only 56% reported neuropsychological testing

special education services

74% n=145

83% report certain subjects harder



87% reported deficits

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

only 25% knew IQ score

- majority reported 60-70 range
- average of all scores reported was 79



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Cognitive & School

performing at grade level 33% n=145

college/vocational 27%

beneficial supports

IEP 66%

1:1 aide 60%

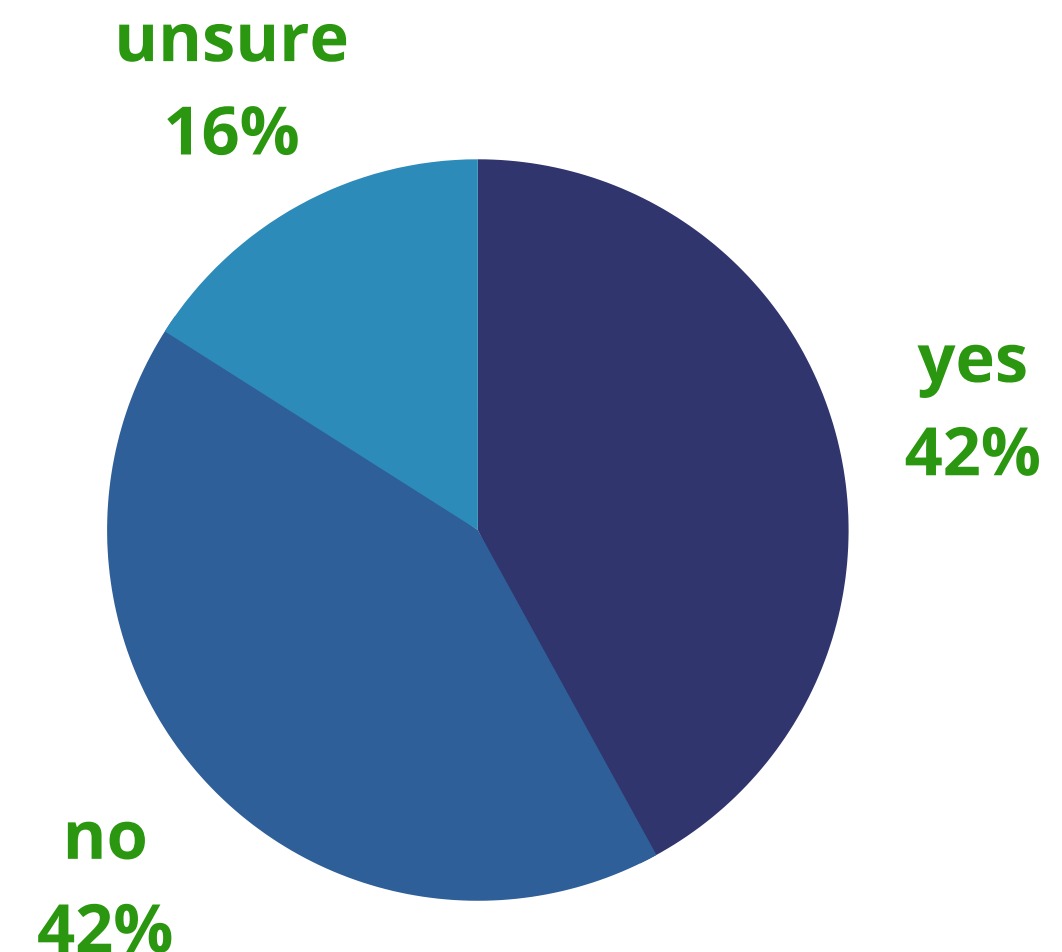
504 plan 18%

special transportation 31%

shortened school day 20%

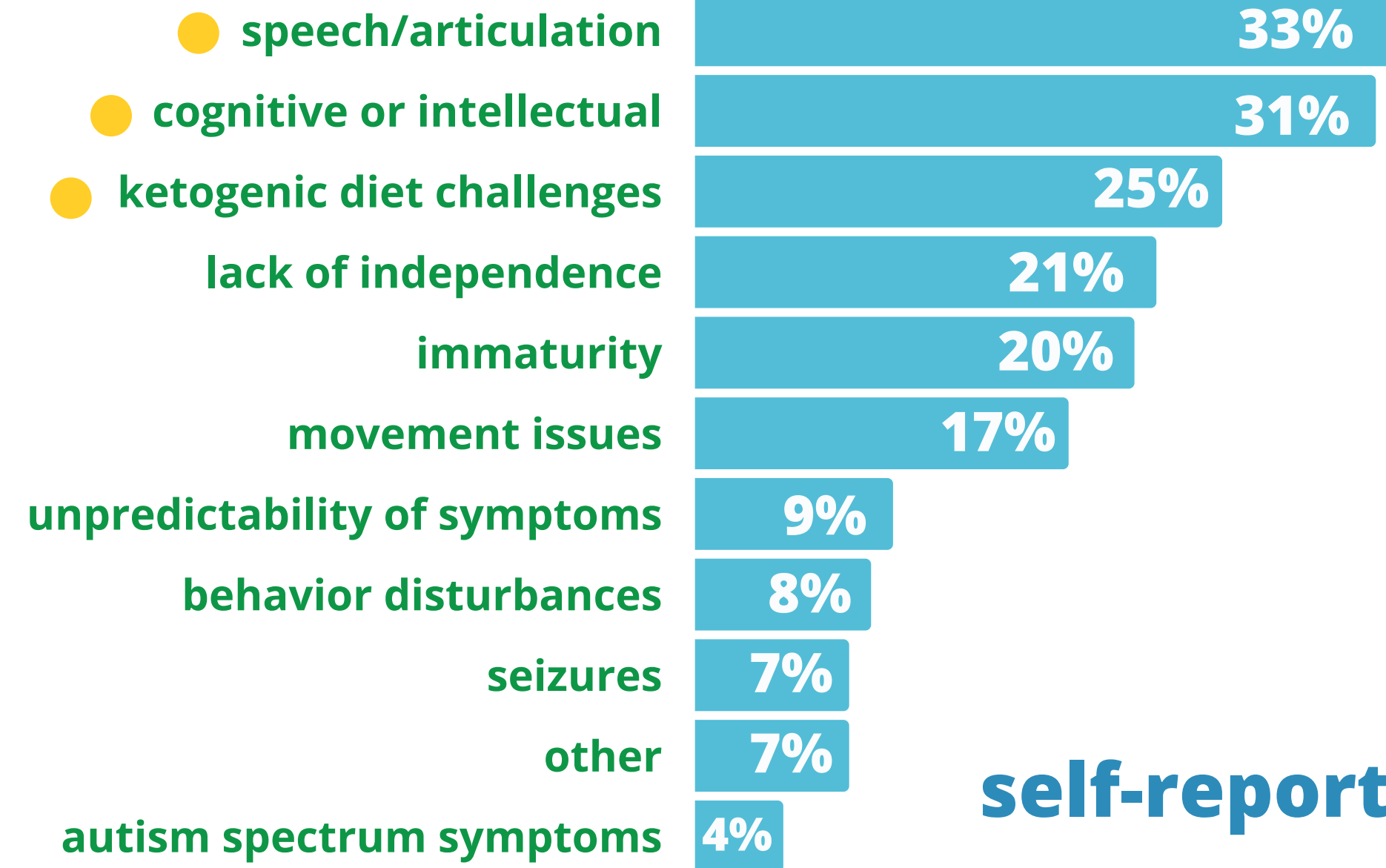
all forms of school based therapies were reported as highly beneficial

trouble getting good curriculum fit?



Social & Emotional

impacts on social life choose top 3



n=260

77% describe self as social

2/3 report having close friendships

27% report mood disturbances

20% report behavior disturbances

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



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Social & Emotional

sibling impacts

close friendship with Glut1 Deficiency sibling

missed out on traditions and food celebrations due to KDT

taken on caregiving responsibilities

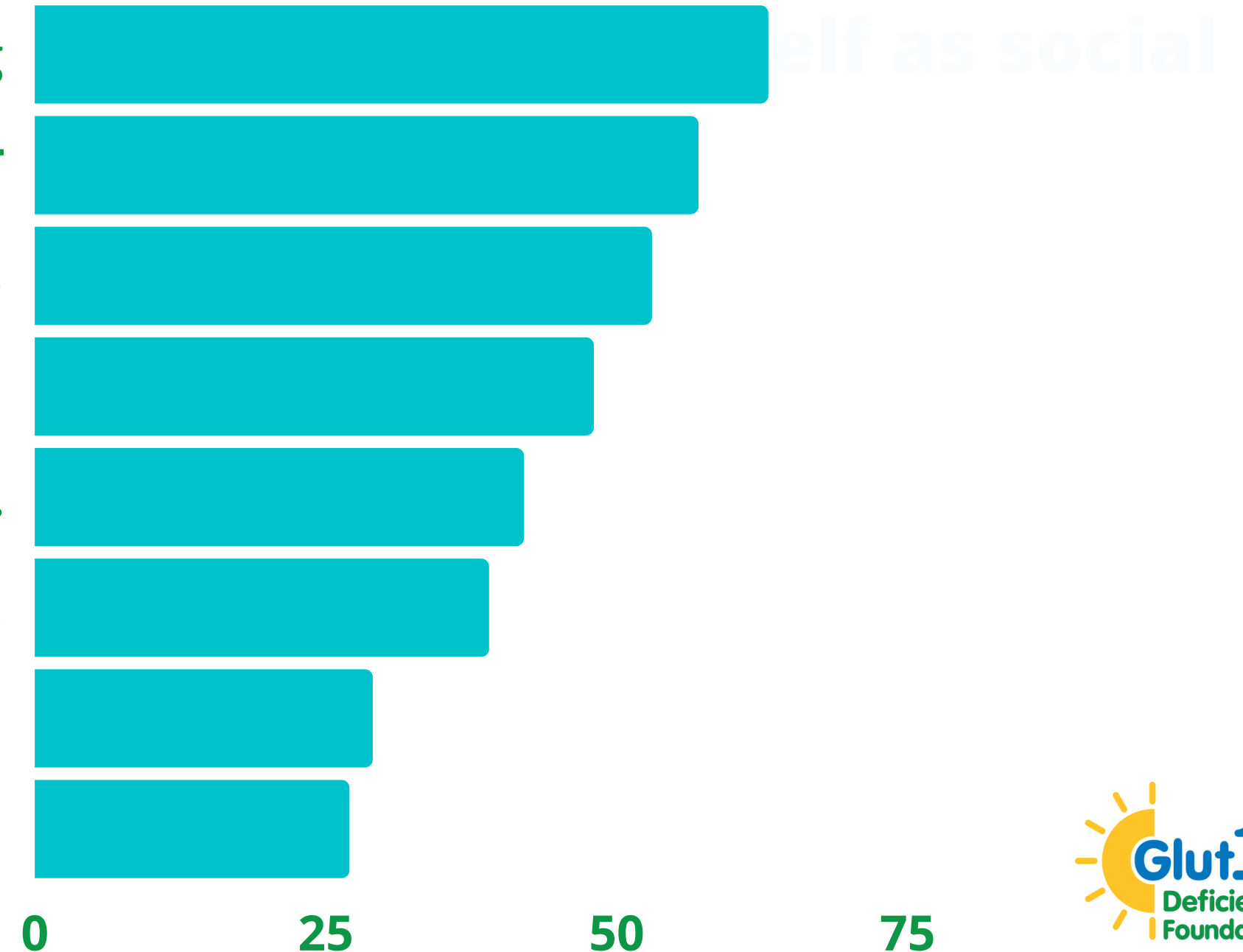
missed out on social opportunities

significant time at medical appointments, therapies, etc.

extreme jealousy or resentment

taken on advocacy role

other



n=260



Ketogenic Diets

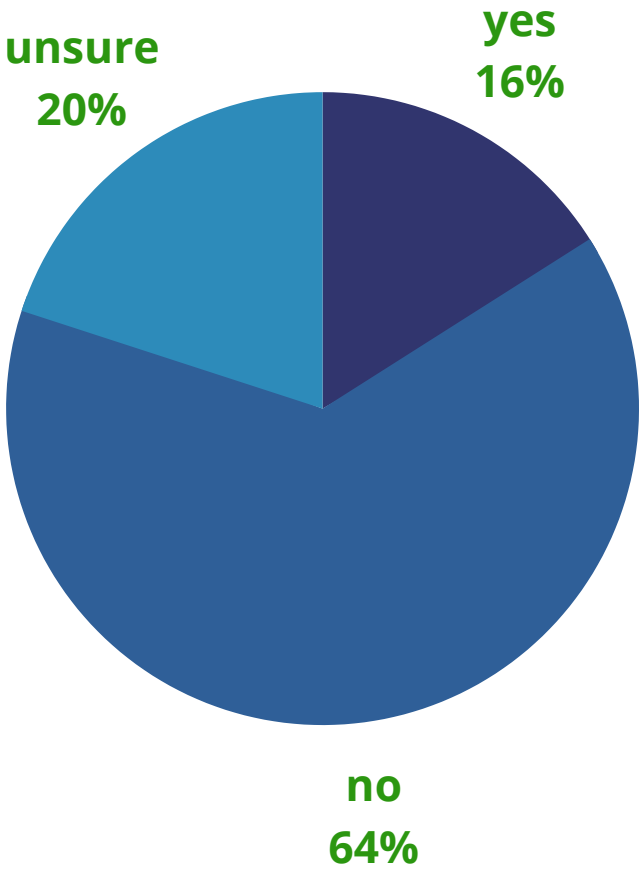
91% have tried ketogenic dietary therapy



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

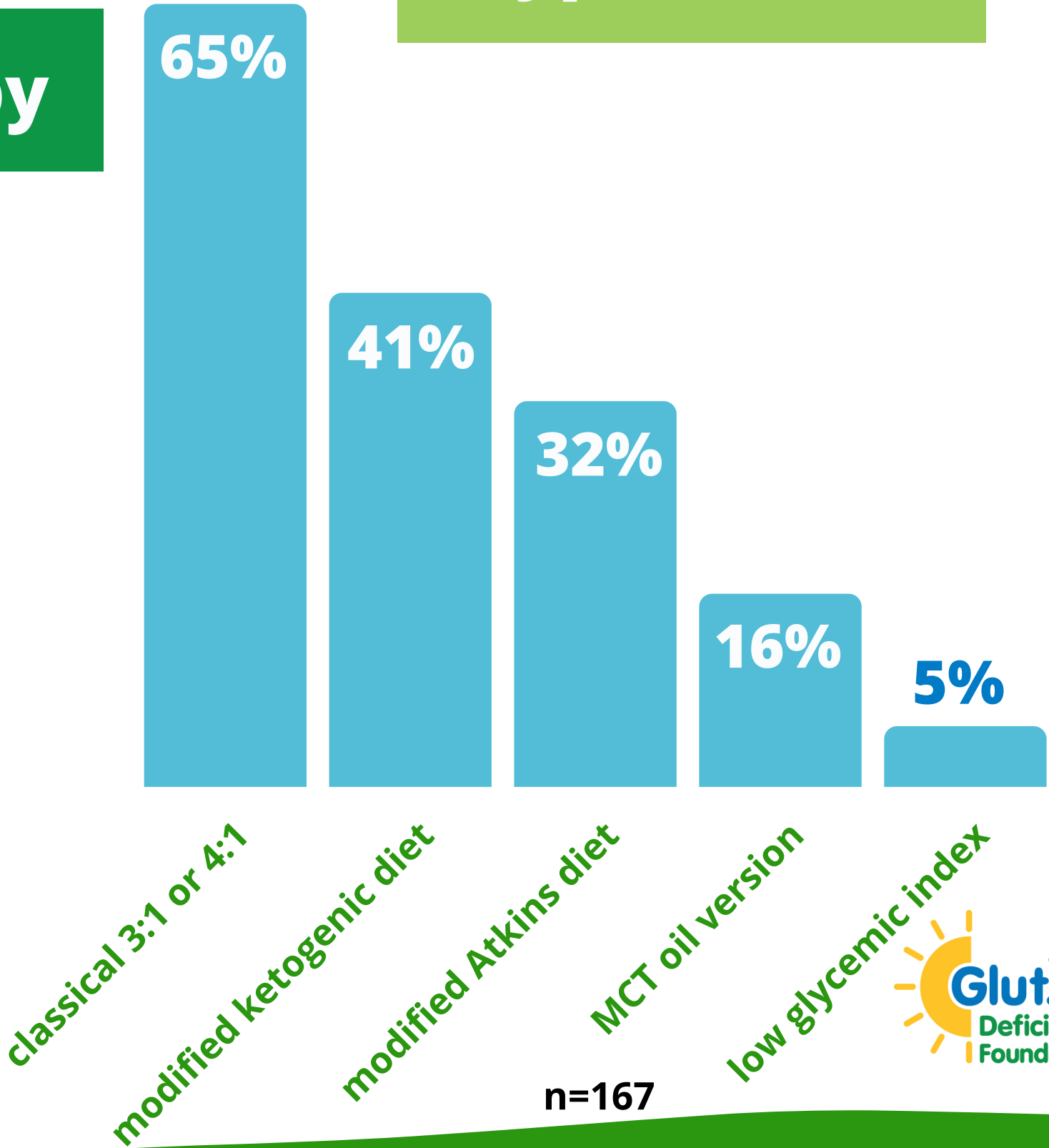
has KDT lost effectiveness
over time?

7.5% have stopped
KDT due to lack of
effectiveness



n=260

types used



n=167



Ketogenic Diet Benefits

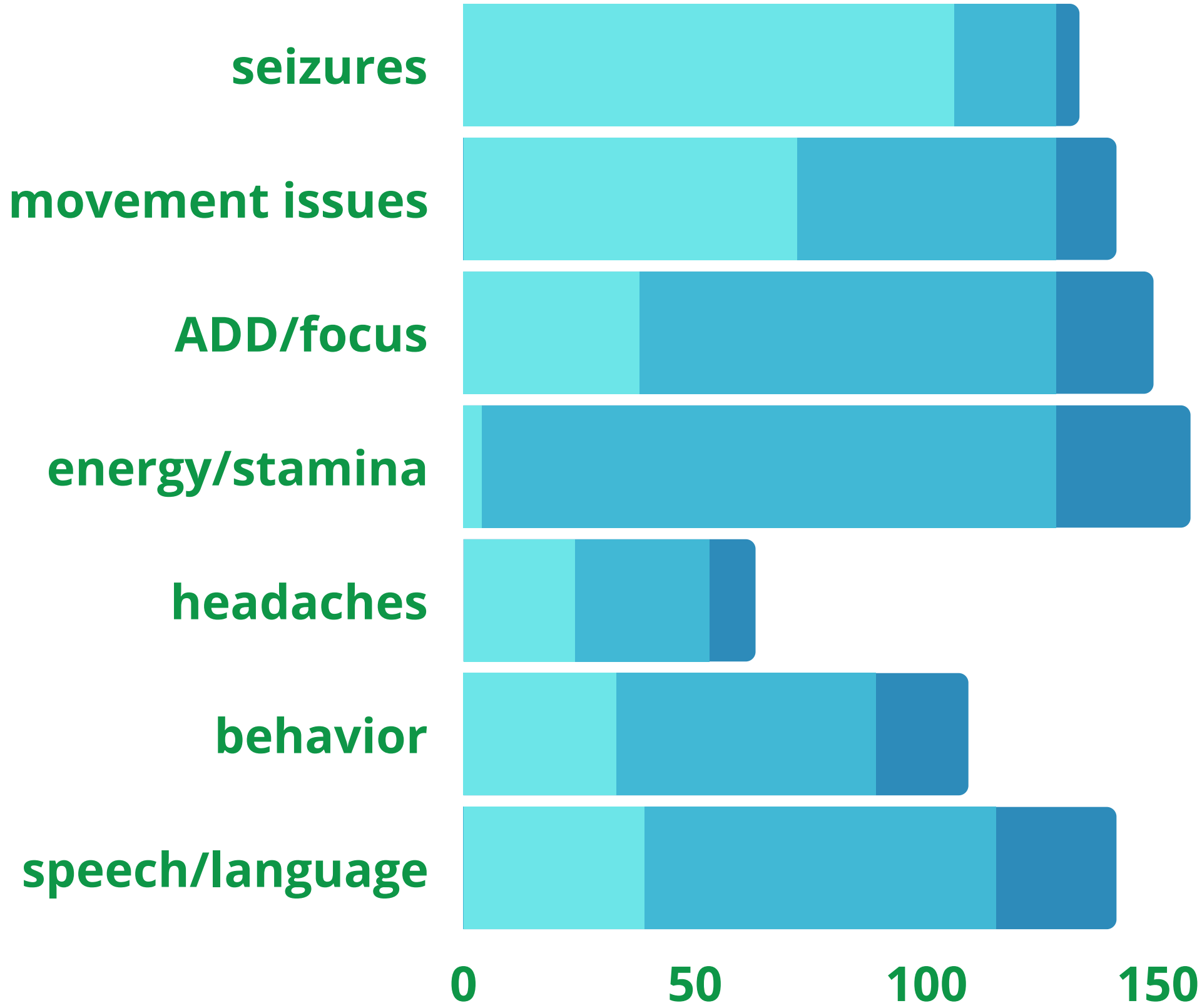
overall

very effective

somewhat effective

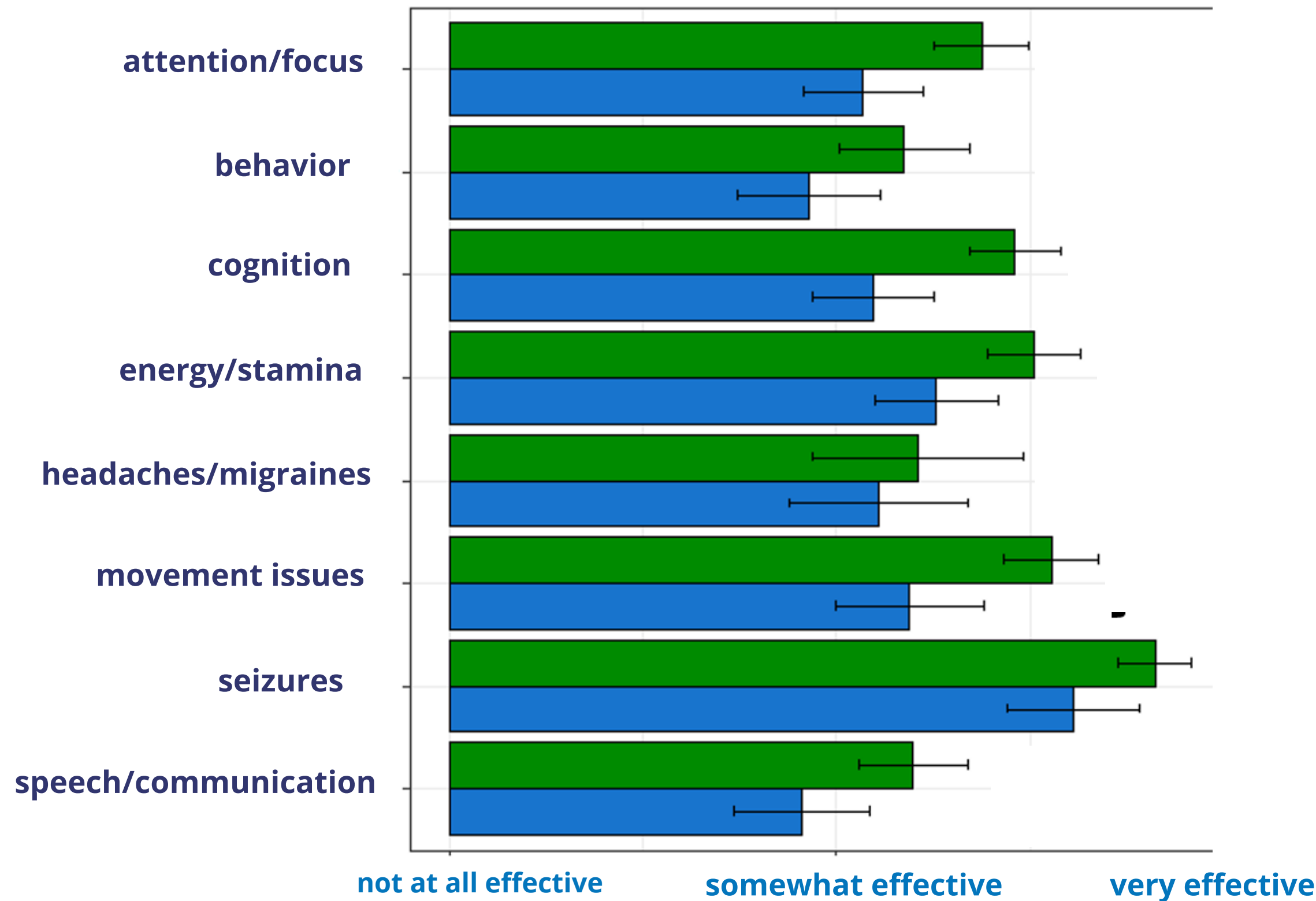
not at all effective

n=167



Ketogenic Diet Benefits

by age



Age Group
0-12
13 and Over

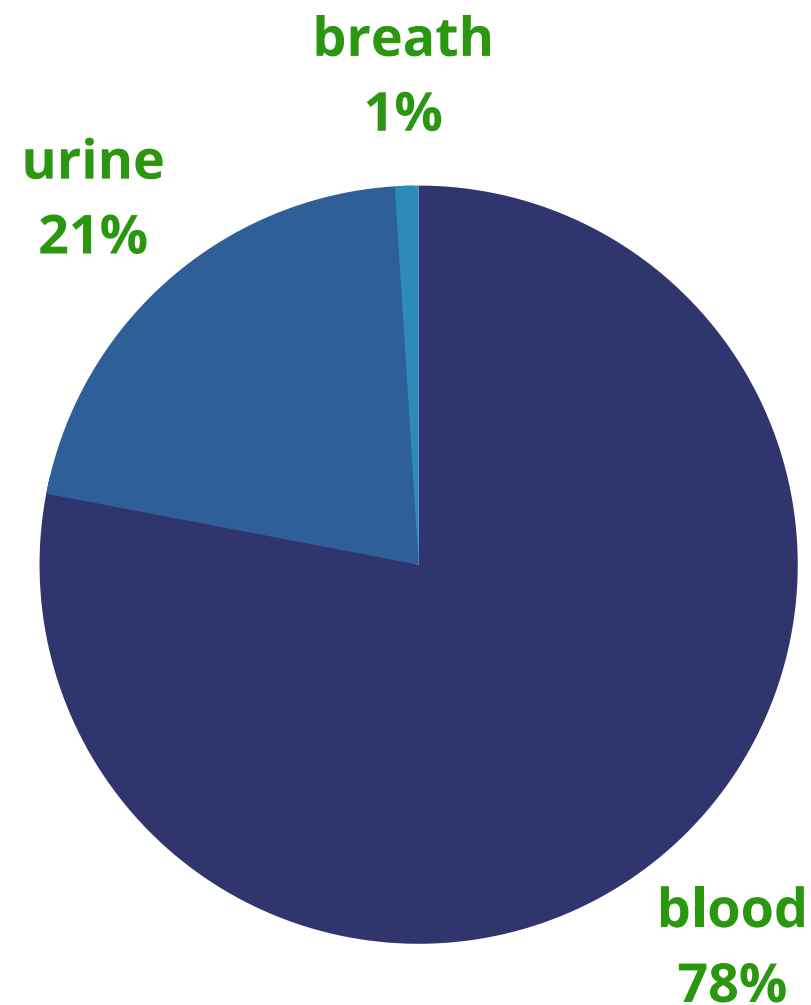
n=167



Ketogenic Diets

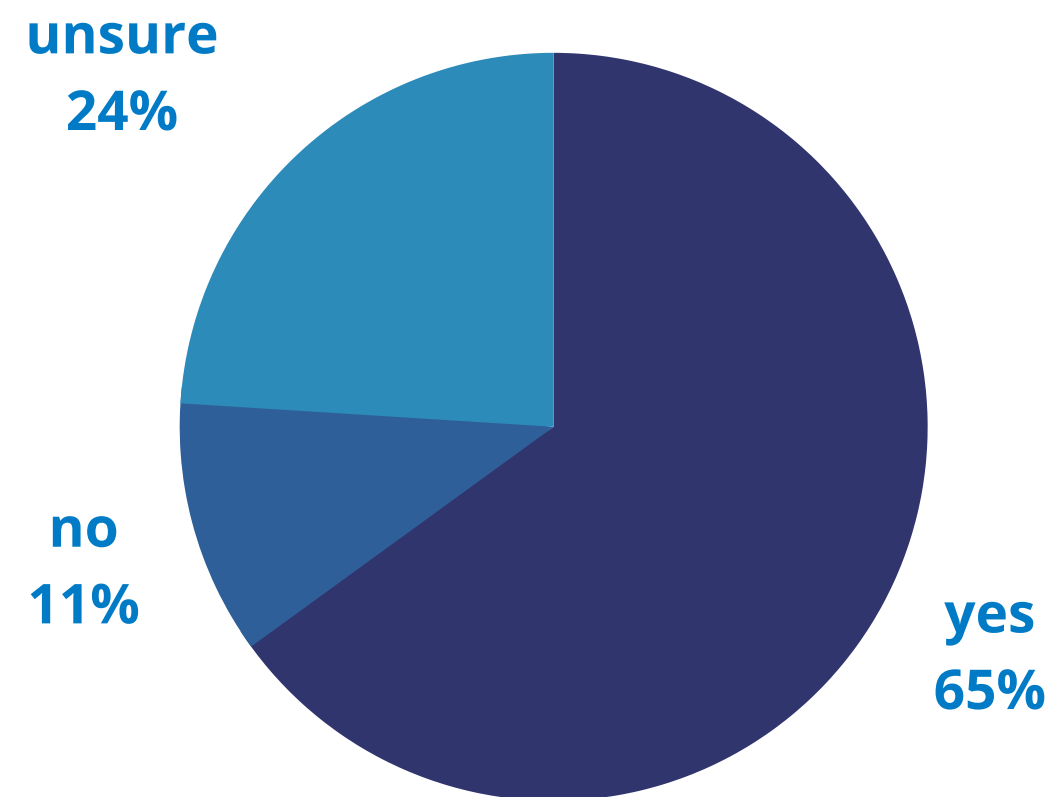
83% measure ketones

method



n=167

correlation to symptoms?



ideal ratio 3:1 44%

"just right" ketone level 3-5

53% report using MCT oils

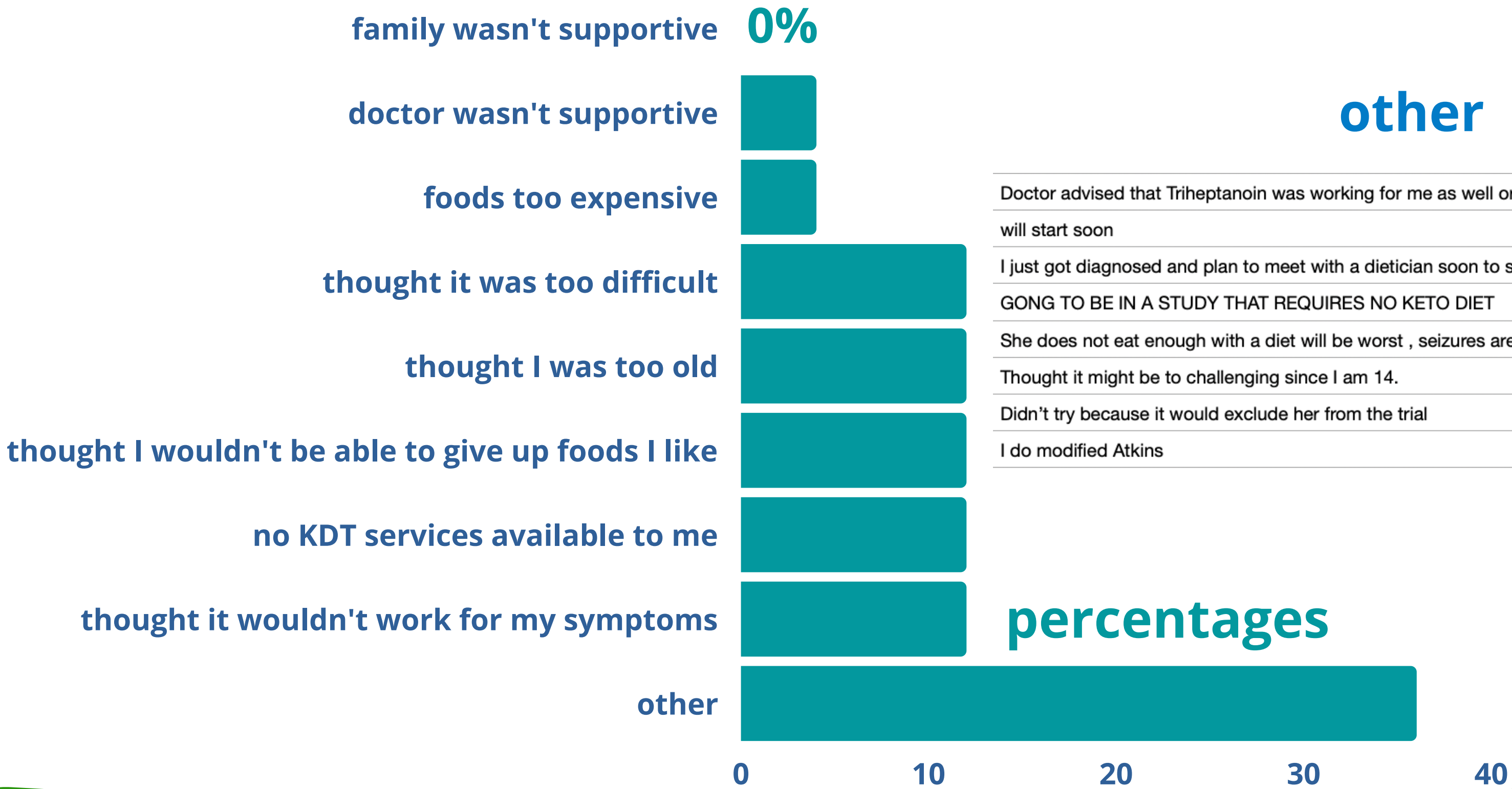
65% of those feel MCT oil makes the diet more effective

8% report using feeding tube



Ketogenic Diets

reasons for not trying KDT



other

Doctor advised that Triheptanoin was working for me as well or better than I could expect with a ketogenic diet will start soon

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 kepra

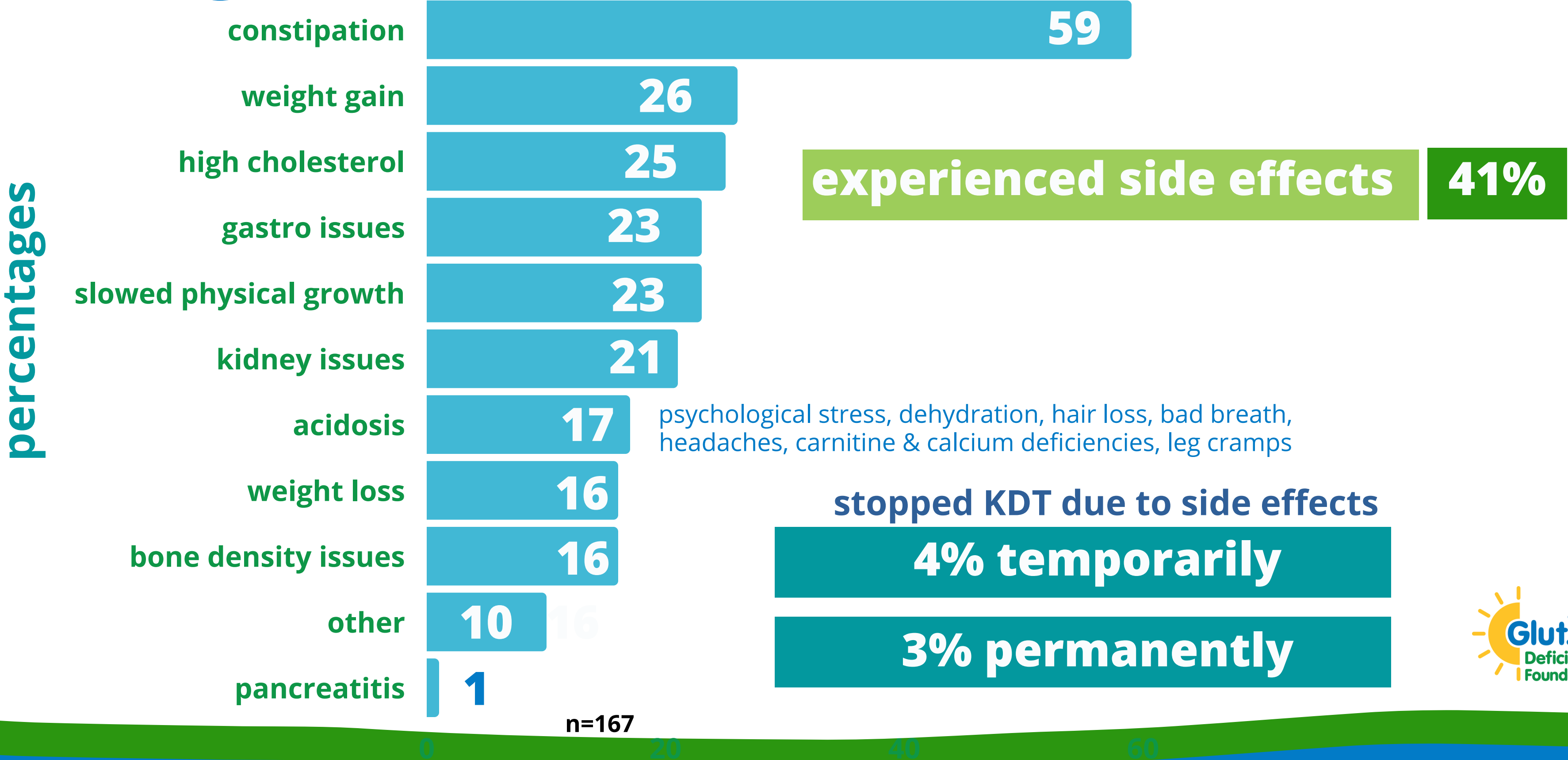
Thought it might be to challenging since I am 14.

Didn't try because it would exclude her from the trial

I do modified Atkins



Ketogenic Diet Side Effects



Ketogenic Diet Challenges

choose top 3

level of difficulty (0-10)

patient 6.4

family 6.6

● social: feeling different

● social: impact on family celebrations/holidays

● lack of opportunities for spontaneity

time required

costs of food and supplies

cooperation/compliance

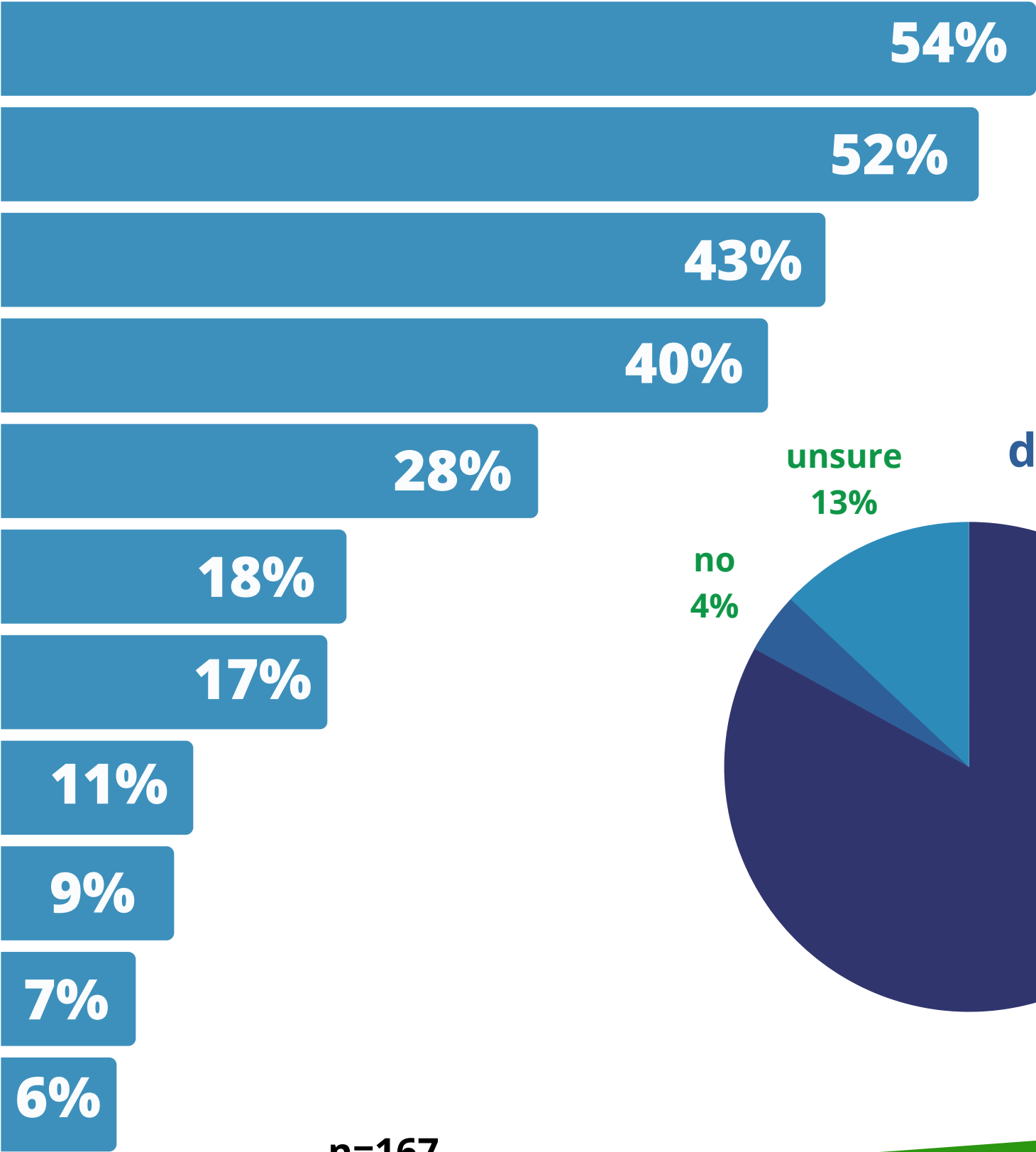
managing complexities of diet (calculations, etc.)

lack of effectiveness for all symptoms

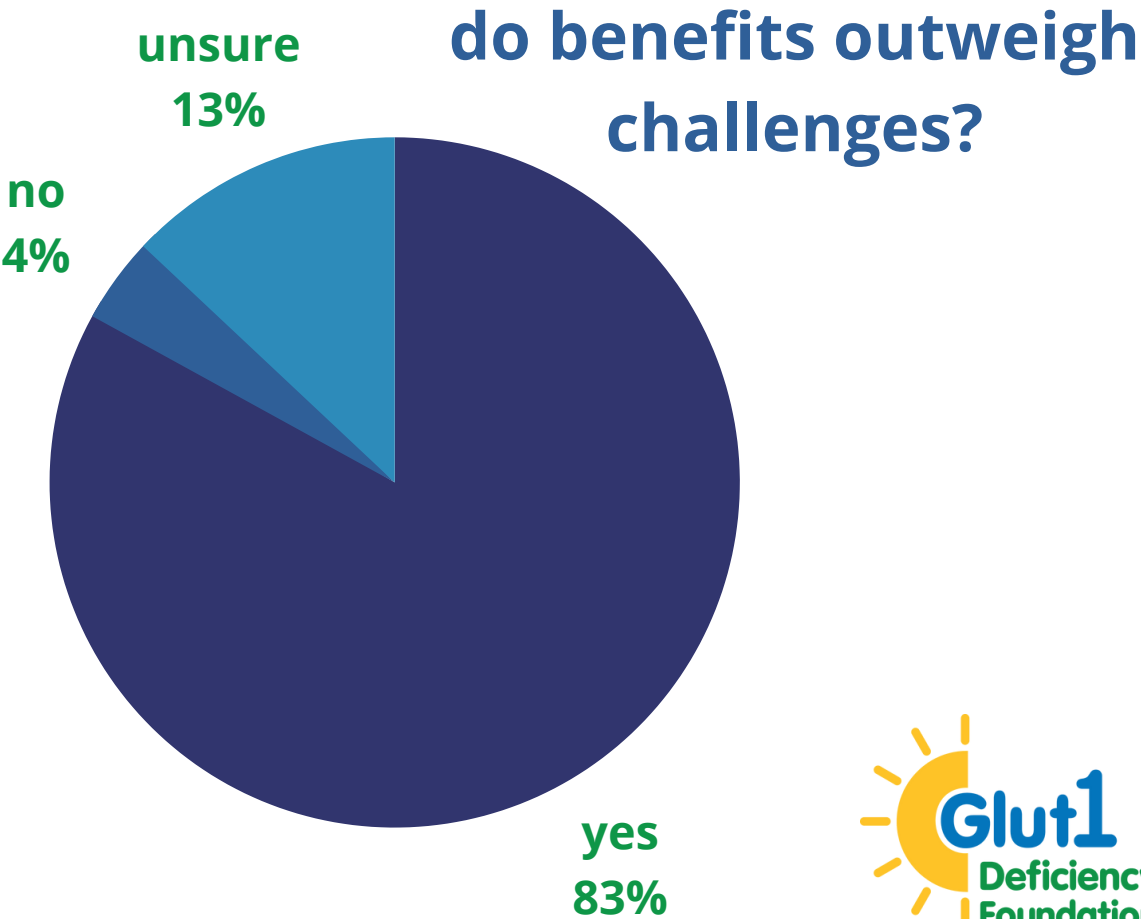
other

lack of medical support

managing side effects

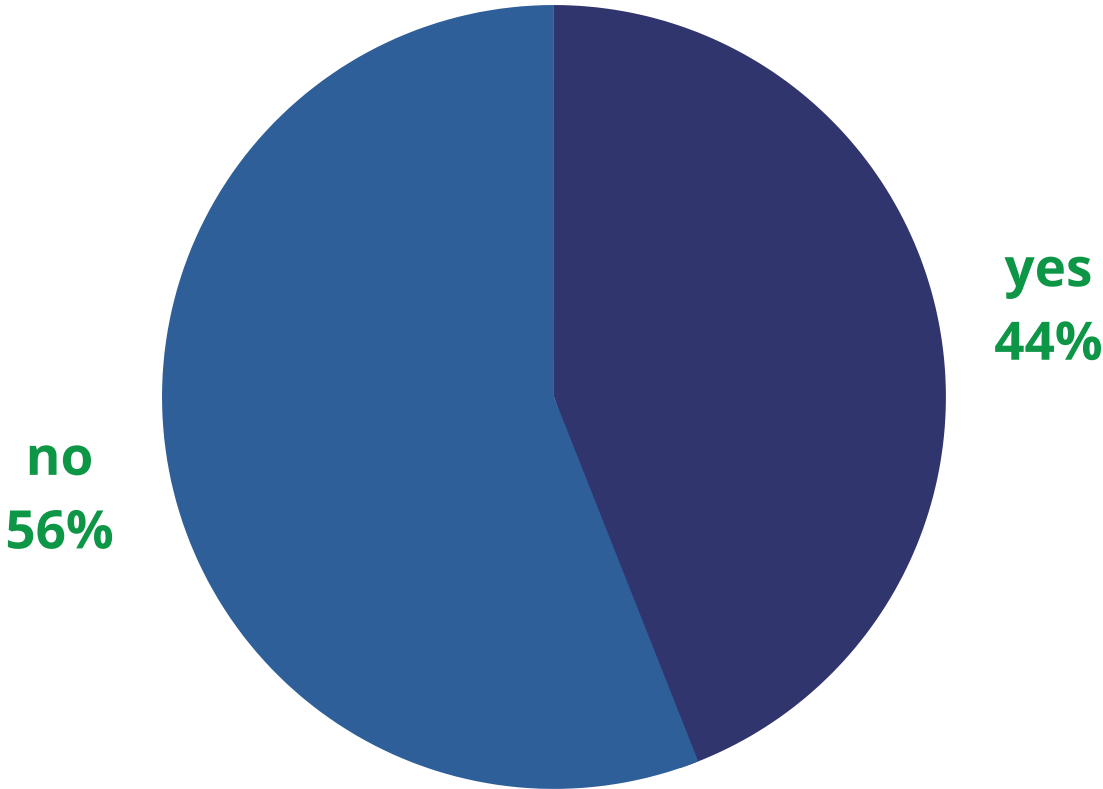


n=167

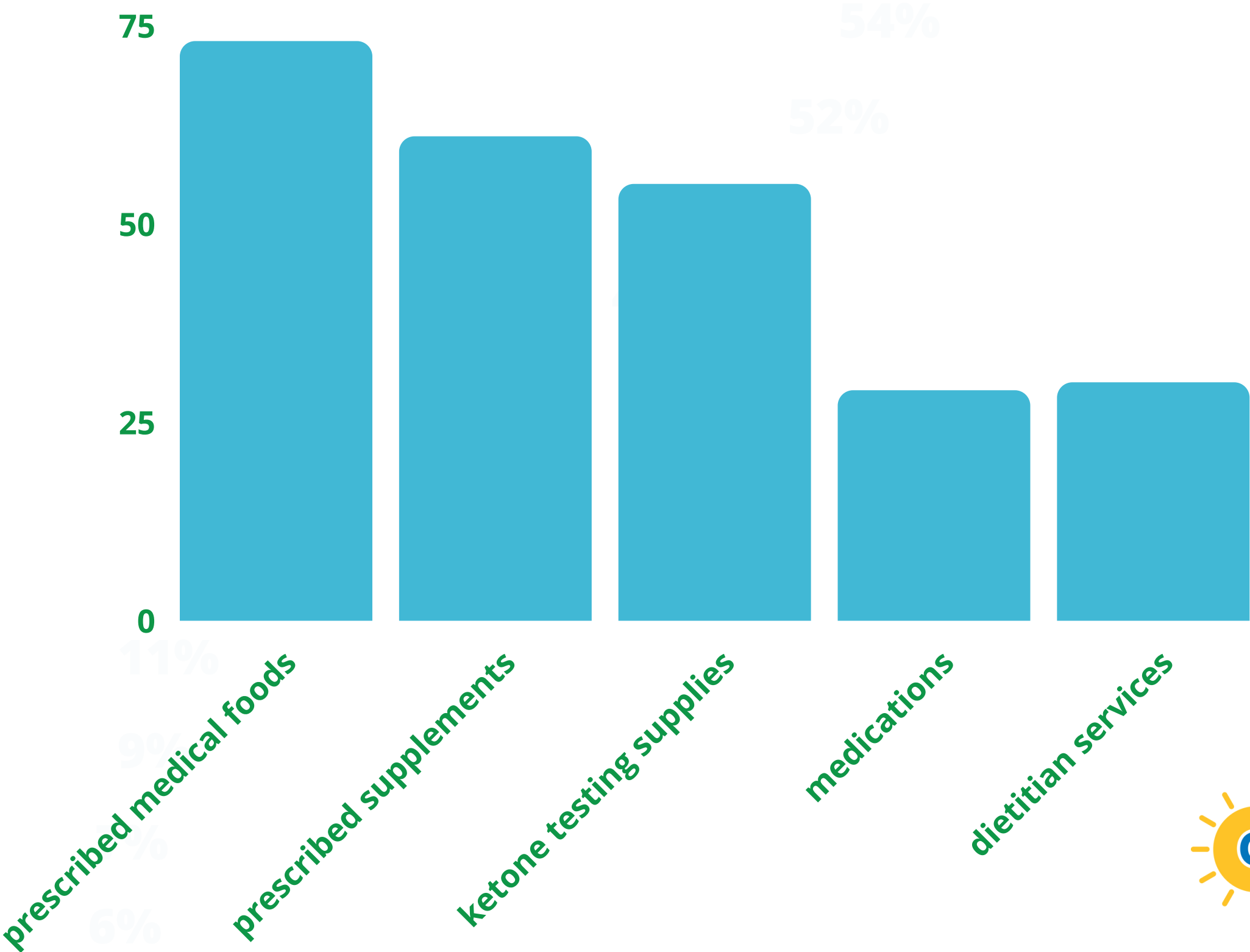


Ketogenic Diet Challenges

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



n=167



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



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Other Treatments

Have you tried other treatments that help?

seizures 35%

keppra/levetiracetam - 22% or 14 total
sodium valproate/depakote - 11% or 7 total
acetazolamide/diamox - 8% or 5 total
ethosuximide - 8% or 5 total
lamotrigine/lamictal - 6% or 4 total

movements 17%

baclofen
acetazolamide/diamox

ADD/focus 16%

vyvanse
methylphenidate/ritalin

anxiety 10%

CBD oil
sertraline
paroxetine

migraines 10%

acetaminophen/paracetamol

rescue meds

**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



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Other Treatments

ketone supplements

20% have tried
60% found it effective

CBD Oil

9% have tried
53% found it effective
*movement, anxiety

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

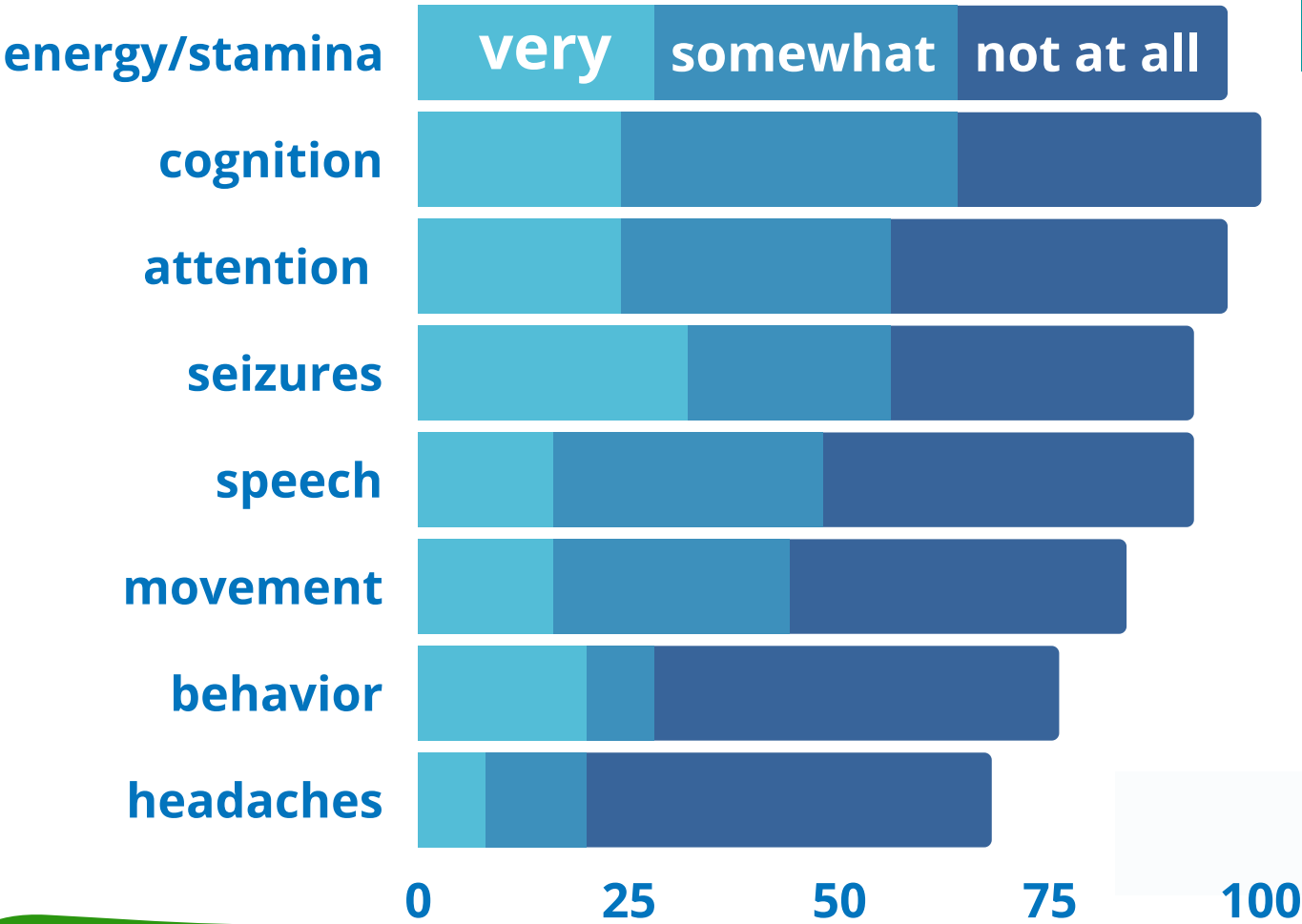
which is most effective?

use both together
16.7%

equal effectiveness
16.7%

C7 oil
16.7%

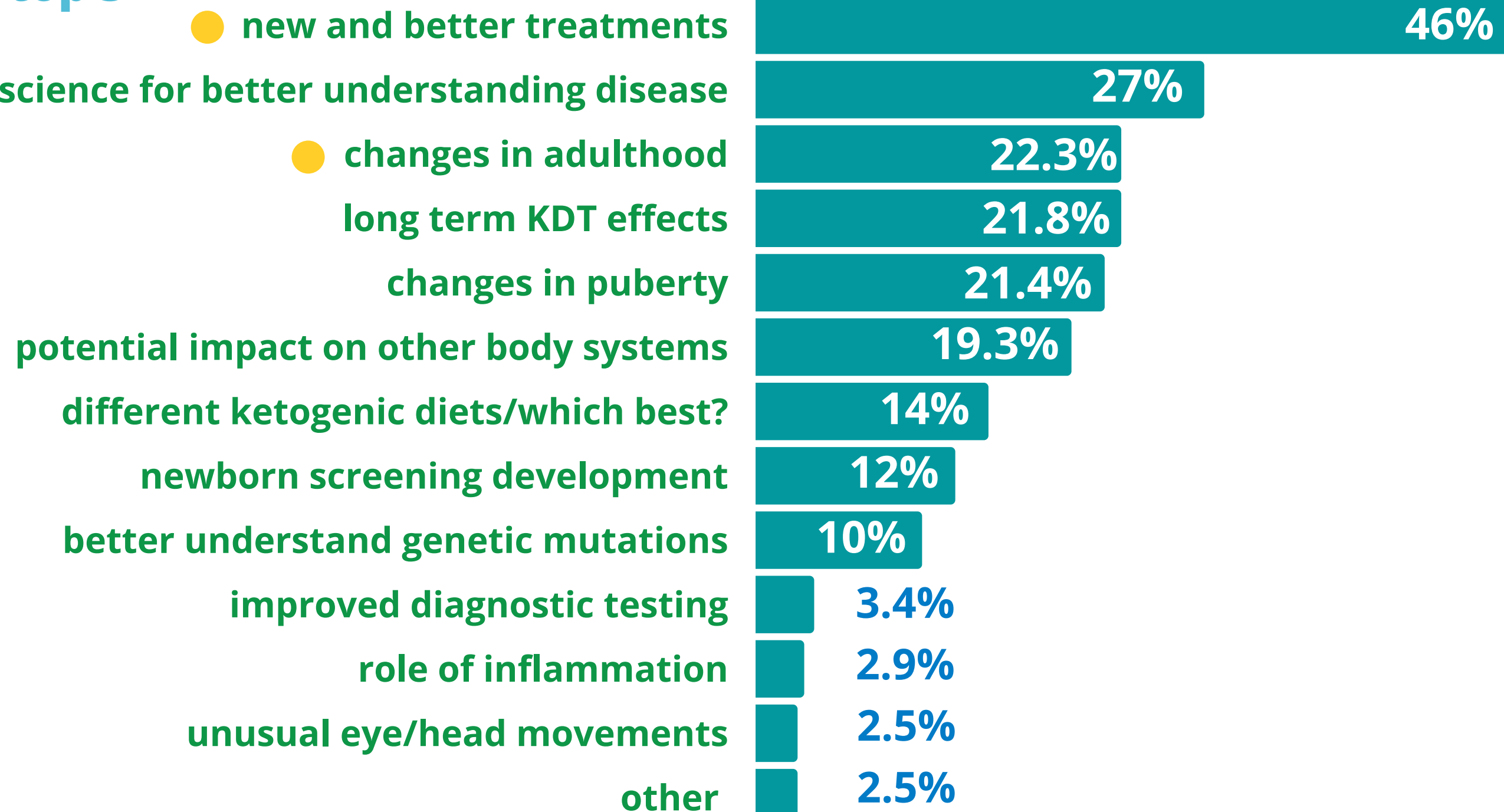
KDT
50%



n=184

Patient & Family Research Priorities

choose top 3



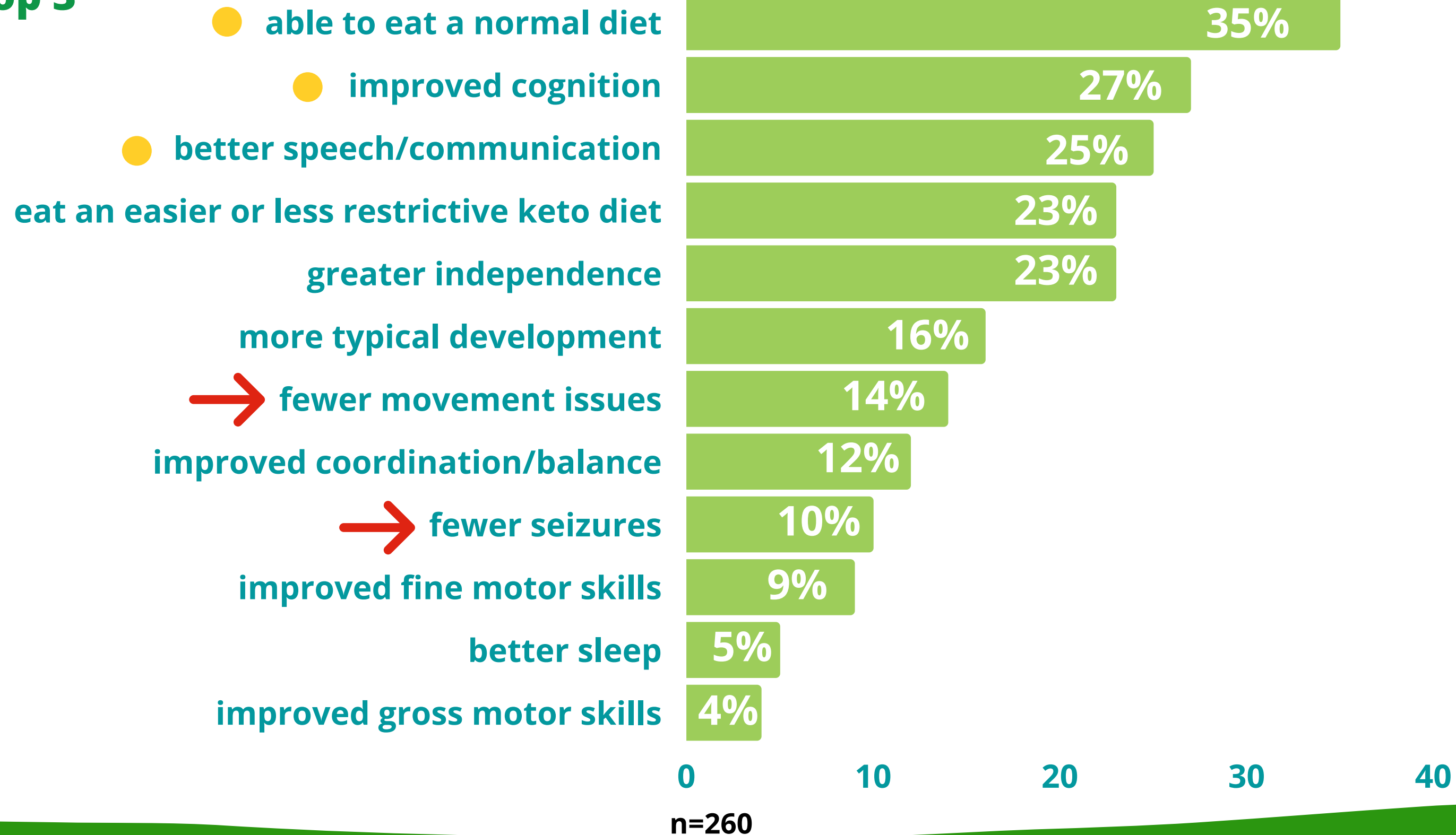
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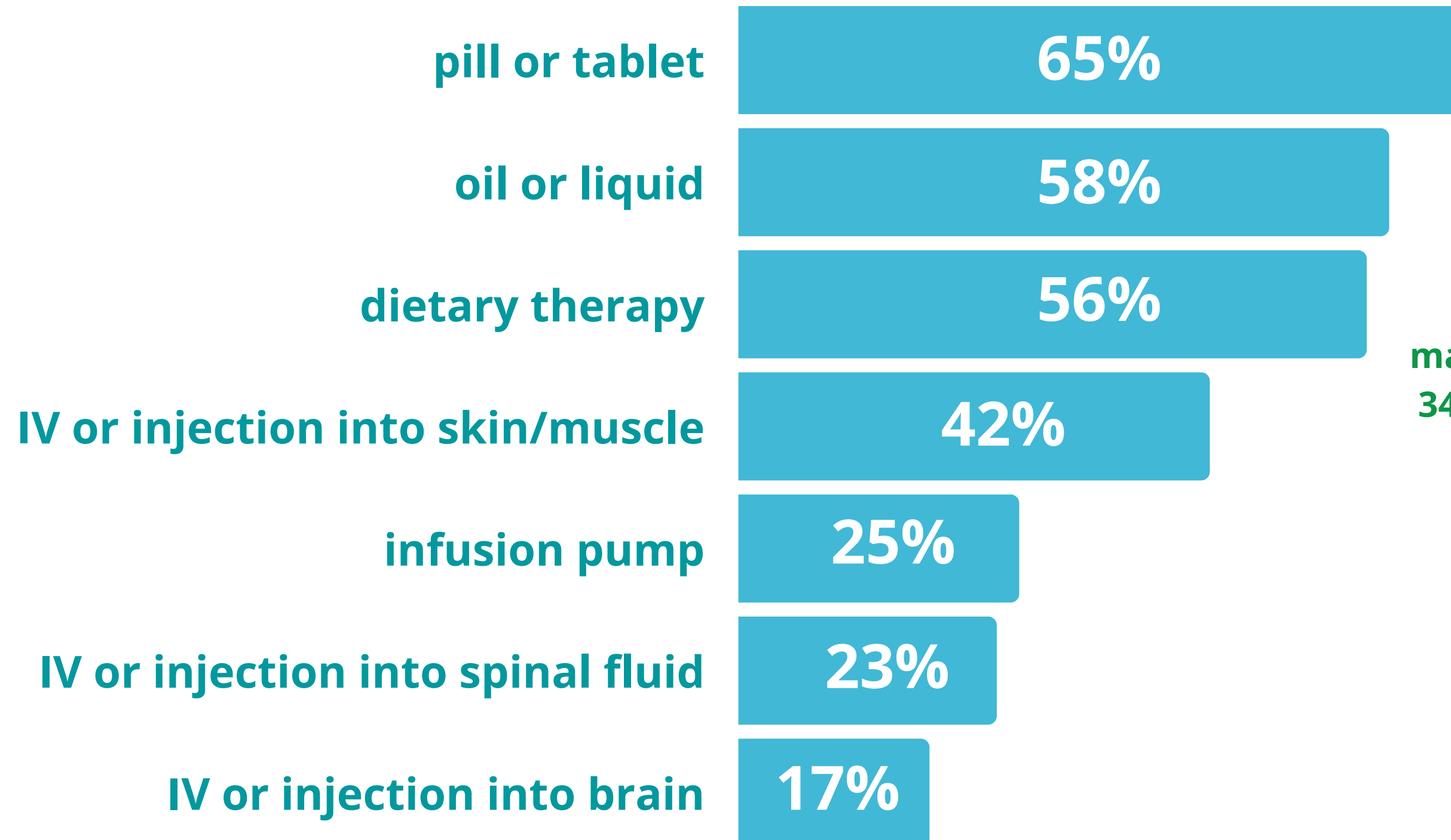
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Priority Outcomes for New Treatments

choose top 3



Types of New Treatments Willing to Try



n=260



Family Burdens

choose top 3

financial burden

- cost of ketogenic diet
- gave up career for caregiving
- out of pocket therapies
- out of pocket medical costs
- travel costs for medical care
- more expensive insurance plans

32%

27%

23%

20%

15%

9%

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

overall burden

- overall family life
- social life
- emotional/mental

43%

42%

40%

finances

32%

sibling life

30%

career

24%

relationship spouse/partner

24%

other

4%

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!



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