Non-Seizure Symptoms & Quality-of-Life in Epileptic Encephalopathies

Why are you being asked to take part in this study?

You are being asked to take part in this research study because you have been identified as caring for someone who has epilepsy and/or a genetic neurodevelopmental disorder.

What is the purpose of this research study?

The researchers recognize a need for proper diagnosis and treatment of symptoms associated with neurodevelopmental disorders, as well as the associated effects on patient quality-of-life.

What is involved in the study?

If you agree to take part in this study, we will ask you to answer questions about your demographics as well as your child’s medical history and quality-of-life. If you agree, the questionnaire will take about 20 minutes.

What are the risks and benefits of this study?

As with any study involving collection of data, there is the possibility your confidentiality information will be shared with others. Every precaution will be taken to secure your personal information to ensure confidentiality.

There are no physical risks with the questionnaire, but you might experience momentary embarrassment or discomfort. You do not have to answer any questions that make you uncomfortable.

The knowledge gained from this study may help determine prevalence of non-seizure features in epileptic encephalopathies as well as understanding of quality-of-life concerns for patients and their families.

Do you need to give your consent in order to participate?

By completing the questionnaire, you are indicating that you have had your questions answered, and you agree to take part in this research study.

Participation in this study is voluntary. You do not have to take part in order to receive care through the University of Pennsylvania health system or the Children's Hospital of Philadelphia. If you decide not to take part or if you change your mind later there will be no penalties or loss of any benefits to which you are otherwise entitled. You can stop the questionnaire at any time.

What about privacy and confidentiality?

We will do our best to keep your personal information private and confidential. However, we cannot guarantee absolute confidentiality. Your personal information may be disclosed if required by law. People from oversight agencies and organizations such as the Department of Health and Human Services, Office for Human Research Protections may also look at your study records.

The results of this study may be shown at meetings or published in journals to inform other doctors and health professionals. We will keep your identity private in any publication or presentation about the study.

By law, the University of Pennsylvania is required to protect your private information. The investigator and staff involved with the study will keep your private information collected for the study strictly confidential.

Financial Information

There are no costs to participate in the study. We are offering $20 gift cards through a random drawing for study participants. At the end of the survey, there will be a link to a separate survey where you can enter your email address to be entered into the lottery. This ensures that your email address cannot be tied to your survey responses.

Where are these questions from?

Questions from this survey were derived from multiple validated tools. Please visit the following references for more information:

Downs J, Jacoby P, Leonard H, et al. Psychometric properties of the Quality of Life Inventory-Disability (QI-Disability) measure. Qual Life Res. 2019;28(3):783-794. doi:10.1007/s11136-018-2057-3

Demarest S, Pestana-Knight EM, Olson HE, et al. Severity Assessment in CDKL5 Deficiency Disorder. Pediatr Neurol. 2019;97:38-42. doi:10.1016/j.pediatrneurol.2019.03.017

https://www.epilepsy.com/about-us/epilepsy-learning-healthcare-system-elhs

What if you have questions about the study?

If you have questions about the study, please email the study investigators Stacey Cohen at stacey.cohen@pennmedicine.upenn.edu, Laura Conway at laura.conway@pennmedicine.upenn.edu, or Katherine Helbig at helbigk@email.chop.edu.

Part 1. Demographics

Questions using "they/them" are referring to the individual affected with epilepsy and/or a neurodevelopmental disorder. Questions using "you" are referring to the caregiver.

Are you currently the primary caregiver of someone diagnosed with epilepsy or a genetic neurodevelopmental disorder?

◯ Yes
◯ No

Have you completed this survey before?

◯ Yes
◯ No

What is their current age?

_____
(Please enter in years. If they are less than 1-year-old, please enter in months. For example, enter their age as "11 years" or "8 months".)

What is your relationship to them? I am their _____.

◯ Parent
◯ Other family member
◯ Other

Please specify "other":

_____

Which organization(s) are you a member of? Please select all that apply.

☐ FamilieSCN2A Foundation
☐ Cute Syndrome Foundation (SCN8A)
☐ Dravet Syndrome Foundation
☐ LGS Foundation
☐ KCNQ2 Cure Alliance
☐ STXBP1 Foundation
☐ SLC6A1 Connect
☐ CureGRIN Foundation
☐ PCDH19 Alliance
☐ Wishes for Elliott
☐ Other

Please specify "other":

_____

Do you feel well supported and informed by this organization?

◯ I feel well supported and informed.
◯ I feel well supported but not well informed.
◯ I feel well informed but not well supported.
◯ I do not feel well informed nor well supported.
(If you are a member of multiple organizations, please answer for the organization you interact most frequently with.)

What is their biological sex?	◯ Male ◯ Female ◯ Intersex ◯ Prefer not to say
How would you describe the area you live in?	◯ Rural ◯ Suburban ◯ Urban
How far do you typically travel to attend medical appointments?	◯ 0-15 miles ◯ 16-50 miles ◯ 51-100 miles ◯ More than 100 miles (If there are multiple answers, choose the distance to the provider they see most frequently.)
What is your current employment status?	◯ Employed part time (< 30 hours a week) ◯ Employed full time (30 or more hours/week) ◯ Self employed ◯ Not employed and seeking work ◯ Not employed - retired ◯ Not employed - unable to work ◯ Student ◯ Prefer not to say
What is your approximate annual household income before taxes?	◯ Less than $10,000 ◯ $10,000-$14,999 ◯ $15,000-$24,999 ◯ $25,000-$34,999 ◯ $35,000-$49,999 ◯ $50,000-$74,999 ◯ $75,000-$99,999 ◯ $100,000-$149,999 ◯ $150,000-$199,999 ◯ $200,000+ ◯ Prefer not to say
What is the highest level of education you have received?	◯ Some high school, no diploma ◯ High school graduate ◯ Technical/trade school ◯ Some college, no degree ◯ Associates degree ◯ Bachelor's degree ◯ Master's degree or more ◯ Prefer not to say
Which of the following describes their health insurance status?	◯ No health insurance ◯ Private health insurance ◯ Medicaid health insurance ◯ Both private and Medicaid health insurance ◯ Don't know ◯ Prefer not to say
Are they registered in any of the following genetic registries? Please select all that apply.	☐ Simons Searchlight/Geisinger ☐ TIGER ☐ They are not in any genetic registries ☐ Don't know ☐ Other

Please specify "other":	_____
How would you describe their race?	◯ American Indian or Alaska Native ◯ Asian ◯ Black or African American ◯ Native Hawaiian or Other Pacific Islander ◯ White ◯ Two or more races ◯ Other ◯ Prefer not to say
How would you describe their ethnicity?	◯ Hispanic or Latino ◯ Not Hispanic or Latino ◯ Prefer not to say

Section 2. Medical History

Questions using "they/them" are referring to the individual affected with epilepsy and/or a neurodevelopmental disorder. Questions using "you" are referring to the caregiver.

For questions asking about age of diagnosis in years, if the individual was diagnosed at younger than 1-year-old, please fill in "0".

Which of the following behavioral conditions have they been diagnosed with by a medical professional? Please select all that apply.	☐ ADD ☐ ADHD ☐ Self-injurious behavior ☐ Bruxism (Teeth grinding) ☐ Aggression ☐ Stereotypies ☐ Other ☐ None of the above
Please specify "other":	_____
At what age (in years) were they diagnosed with the behavioral condition you indicated as "other"?	_____
At what age (in years) were they diagnosed with ADD?	_____
At what age (in years) were they diagnosed with ADHD?	_____
At what age (in years) were they diagnosed with self-injurious behavior?	_____
At what age (in years) were they diagnosed with bruxism?	_____
At what age (in years) were they diagnosed with aggression?	_____

At what age (in years) were they diagnosed with stereotypies?	_____
Which of the following speech conditions were they diagnosed with by a medical professional? Please select all that apply.	☐ Speech delay ☐ Motor speech disorder. This may include speech apraxia, dyspraxia, or dysphasia. ☐ Other speech condition, including lack of speech ☐ None of the above
Please specify "other":	_____
At what age were they diagnosed with the speech condition you indicated as "other"?	_____
At what age (in years) were they diagnosed with speech delay?	_____
At what age (in years) were they diagnosed with a motor speech disorder?	_____
Which of the following forms of developmental delay and/or regression have they been diagnosed with by a medical professional? Please select all that apply.	☐ Motor delay (hand-eye coordination, holding a pencil, crawling, walking, etc) ☐ Speech/language delay ☐ Social and emotional delay ☐ Cognitive delay (Intellectual function lower than expected for their age) ☐ Global developmental delay (delayed development across 2 or more domains) ☐ Motor regression (motor milestones that were achieved and subsequently lost) ☐ Speech/language regression (speech/language milestones that were achieved and subsequently lost) ☐ Other delay/regression ☐ None of the above (In this question, we wanted to ask about all dimensions of development. Please note that some of the features may have been assessed in some of the other questions and are listed for completion.)
Please specify "other":	_____
At what age (in years) were they diagnosed with "other" delay or regression?	_____
At what age (in years) were they diagnosed with motor delay?	_____
At what age (in years) were they diagnosed with speech/language delay?	_____
At what age (in years) were they diagnosed with social and emotional delay?	_____
At what age (in years) were they diagnosed with cognitive delay?	_____

At what age (in years) were they diagnosed with global developmental delay?	_____
At what age (in years) were they diagnosed with motor regression?	_____
At what age (in years) were they diagnosed with speech/language regression?	_____
Which of the following conditions affecting vision or eye movement have they been diagnosed with by a medical professional? Please select all that apply.	☐ Cortical visual impairment ☐ Strabismus and/or esotropia (eye or eyes turning inward) ☐ Oculomotor apraxia (difficulty producing controlled, voluntary, and purposeful eye movement) ☐ Nystagmus (repetitive, uncontrolled movements of the eye) ☐ Blindness ☐ Myopia (nearsightedness) ☐ Hyperopia (farsightedness) ☐ Other visual condition ☐ None of the above
Please specify "other":	_____
At what age (in years) were they diagnosed with the visual condition you indicated as "other"?	_____
At what age (in years) were they diagnosed with cortical visual impairment?	_____
At what age (in years) were they diagnosed with strabismus and/or esotropia?	_____
At what age (in years) were they diagnosed with oculomotor apraxia?	_____
At what age (in years) were they diagnosed with nystagmus?	_____
At what age (in years) were they diagnosed with blindness?	_____
At what age (in years) were they diagnosed with myopia?	_____
At what age (in years) were they diagnosed with hyperopia?	_____

Which of the following neurological conditions were they diagnosed with by a medical professional? Please select all that apply.	☐ Intellectual disability ☐ Hypotonia (low muscle tone) ☐ Hypertonia (stiffness of the muscles because of increased muscle tone) ☐ Spasticity ☐ Autism spectrum disorder ☐ Epilepsy/seizures ☐ Movement disorder ☐ Autonomic dysfunction ☐ Other neurological condition ☐ None of the above
Please specify "other":	_____
At what age (in years) were they diagnosed with the neurological condition you indicated as "other"?	_____
At what age (in years) were they diagnosed with intellectual disability?	_____
Which level of intellectual disability best describes their level of functioning?	◯ Mild (Can live independently with minimum levels of support.) ◯ Moderate (Independent living may be achieved with moderate levels of support, such as those available in group homes.) ◯ Severe (Requires daily assistance with self-care activities and safety supervision.) ◯ Profound (Requires 24-hour care.)
At what age (in years) were they diagnosed with hypotonia?	_____
At what age (in years) were they diagnosed with hypertonia?	_____
At what age (in years) were they diagnosed with spasticity?	_____
At what age (in years) were they diagnosed with autism spectrum disorder?	_____ (Please consider any diagnosis in this spectrum including Asperger's syndrome and Pervasive Developmental Delay- Not Otherwise Specified (PDD-NOS).)
At what age (in years) did they experience their first seizure?	_____

What type of seizures have they experienced? Please select all that apply.	☐ Tonic-clonic ☐ Tonic ☐ Atonic ☐ Myoclonic ☐ Myoclonic-atonic ☐ Epileptic spasms ☐ Typical absence ☐ Atypical absence ☐ Myoclonic absence ☐ Absence with eyelid myoclonia ☐ Aware focal ☐ Impaired awareness focal ☐ Focal motor ☐ Sensory focal ☐ Cognitive focal ☐ Autonomic focal ☐ Emotional focal ☐ Behavior arrest focal ☐ Focal to bilateral tonic-clonic ☐ Don't know ☐ Other
Please specify "other":	_____
Which of the following classification of epilepsy have they been diagnosed with?	◯ Generalized epilepsy ◯ Focal epilepsy ◯ Generalized and focal epilepsy ◯ Unknown

Which of the following epilepsy syndromes have they been diagnosed with? Please select all that apply.	☐ Atypical childhood epilepsy with centrotemporal spikes ☐ Autosomal dominant epilepsy with auditory features ☐ Autosomal dominant nocturnal frontal lobe epilepsy ☐ Childhood epilepsy with centrotemporal spikes ☐ Childhood occipital epilepsy (Gastaut type) ☐ Dravet syndrome ☐ Early myoclonic encephalopathy ☐ Epilepsy of infancy with migrating focal seizures ☐ Epilepsy with generalized-tonic clonic seizures alone ☐ Epilepsy with myoclonic-atonic seizures ☐ Epileptic encephalopathy with continuous spike-and-wave during sleep ☐ Familial focal epilepsy with variable foci ☐ Febrile seizures plus ☐ Juvenile absence epilepsy ☐ Juvenile myoclonic epilepsy ☐ Landau-Kleffner syndrome ☐ Lennox-Gastaut syndrome ☐ Myoclonic epilepsy in infancy ☐ Ohtahara syndrome ☐ Panayiotopoulos syndrome ☐ Photosensitive occipital lobe epilepsy ☐ Progressive myoclonus epilepsy ☐ Reflex epilepsy ☐ Self-limited familial and non-familial infantile epilepsy ☐ Self-limited neonatal seizures and self-limited familial neonatal epilepsy ☐ West Syndrome ☐ No syndrome diagnosis ☐ Other syndrome diagnosis ☐ Don't know
Please specify "other":	_____
At what age (in years) were they diagnosed with the seizure disorder you indicated as "other"?	_____
At what age (in years) were they diagnosed with atypical childhood epilepsy with centrotemporal spikes?	_____
At what age (in years) were they diagnosed with autosomal dominant epilepsy with auditory features?	_____
At what age (in years) were they diagnosed with autosomal dominant nocturnal frontal lobe epilepsy?	_____
At what age (in years) were they diagnosed with childhood epilepsy with centrotemporal spikes?	_____
At what age (in years) were they diagnosed with childhood occipital epilepsy (Gastaut type)	_____
At what age (in months) were they diagnosed with Dravet syndrome?	_____

At what age (in months) were they diagnosed with early myoclonic encephalopathy?	_____
At what age (in months) were they diagnosed with epilepsy of infancy with migrating focal seizures?	_____
At what age (in years) were they diagnosed with epilepsy with generalized-tonic clonic seizures alone?	_____
At what age (in years) were they diagnosed with epilepsy with myoclonic-atonic seizures?	_____
At what age (in years) were they diagnosed with epileptic encephalopathy with continuous spike-and-wave during sleep?	_____
At what age (in years) were they diagnosed with familial focal epilepsy with variable foci?	_____
At what age (in months) were they diagnosed with febrile seizures plus?	_____
At what age (in years) were they diagnosed with juvenile absence epilepsy?	_____
At what age (in years) were they diagnosed with juvenile myoclonic epilepsy?	_____
At what age (in years) were they diagnosed with Landau-Kleffner syndrome?	_____
At what age (in years) were they diagnosed with Lennox-Gastaut syndrome?	_____
At what age (in months) were they diagnosed with myoclonic epilepsy in infancy?	_____
At what age (in months) were they diagnosed with Ohtahara syndrome?	_____
At what age (in years) were they diagnosed with Panayiotopoulos syndrome?	_____
At what age (in years) were they diagnosed with photosensitive occipital lobe epilepsy?	_____
At what age (in years) were they diagnosed with progressive myoclonus epilepsy?	_____
At what age (in years) were they diagnosed with reflex epilepsy?	_____
At what age (in months) were they diagnosed with self-limited familial and non-familial infantile epilepsy?	_____

At what age (in months) were they diagnosed with self-limited neonatal seizures and self-limited familial neonatal epilepsy	_____
At what age (in months) were they diagnosed with West Syndrome?	_____
How often do they have seizures?	◯ more than one per day ◯ more than one per week ◯ one per month ◯ one per six months ◯ one per one year ◯ one to two years ago ◯ more than two years ago
How would you describe the frequency of their seizures over the last 6 months as compared to their past history?	◯ Stable ◯ Improved ◯ Worsened ◯ Unknown
How often have their seizures disrupted their routines in the past 2 weeks?	◯ Every day ◯ Most days ◯ Some days ◯ Once or twice ◯ Never ◯ Unknown
How often have they had non-convulsive (no associated movements) seizures in the last 6 months?	◯ Never had any non-convulsive seizure ◯ None in more than 6 months ◯ Has had at least one in the last 6 months, but not monthly ◯ Monthly (on average no more than 1 per month) ◯ Weekly (on average, 2-4 per month) ◯ Daily (on average, 5-30 per month) ◯ More than daily (more than 30 per month) ◯ Don't know
How often have they had convulsive seizures (has associated movements) in the last 6 months?	◯ Never had any convulsive seizure ◯ None in more than 6 months ◯ Has had at least one in the last 6 months, but not monthly ◯ Monthly (on average no more than 1 per month) ◯ Weekly (on average, 2-4 per month) ◯ Daily (on average, 5-30 per month) ◯ More than daily (more than 30 per month) ◯ Don't know
How often have they had epileptic spasms and/or myoclonic jerks (sudden bending forward of the body with stiffening of the arms and legs lasting for 1-2 seconds) in the last 6 months?	◯ Never had an epileptic spasm and/or myoclonic jerk ◯ None in more than 6 months ◯ Has had at least one in the last 6 months, but not monthly ◯ Monthly (on average no more than 1 per month) ◯ Weekly (on average, 2-4 per month) ◯ Daily (on average, 5-30 per month) ◯ More than daily (more than 30 per month) ◯ Don't know

How many distinct seizure types have they experienced in the last 30 days?	◯ No seizures in the last 30 days ◯ One seizure type ◯ Two seizure types ◯ Three seizure types ◯ Four seizure types ◯ Five or more seizure types ◯ Don't know
How many times have they had a prolonged (5 minutes or more) seizure in the last 6 months?	◯ None ever ◯ None in more than 6 months ◯ Has had at least one in the last 6 months, but not monthly ◯ Monthly (on average no more than 1 per month) ◯ Weekly (on average, 2-4 per month) ◯ Daily (on average, 5-30 per month) ◯ More than daily (more than 30 per month) ◯ Don't know
How many times have they had to go to the emergency department as a result of having a prolonged seizure in the last 30 days? Please select all that apply.	☐ No emergency department visits in last 30 days ☐ One emergency department visit in last 30 days ☐ Two emergency department visits in last 30 days ☐ Three or more emergency department visits in last 30 days ☐ Admitted once to hospital more than 24 hours in last 30 days ☐ Admitted to hospital and required ICU in last 30 days
How many anticonvulsant medications have they used during over their lifetime, not including rescue medications, vagus nerve stimulation or diet?	◯ None ◯ One ◯ Two ◯ Three ◯ Four ◯ Five or more
How many different daily anticonvulsant medications have they taken in the last 30 days, not including rescue medications, vagus nerve stimulation, or diet?	◯ None ◯ One ◯ Two ◯ Three ◯ Four ◯ Five or more
How would you describe their adherence to anticonvulsant medications?	◯ Adherent ◯ Not adherent ◯ Unknown
Which of the following best describes their use of the ketogenic diet to manage their seizures over the past 30 days?	◯ Never ◯ Past use but discontinued ◯ Currently used
Which of the following best describes their use of vagus nerve stimulation as seizure treatment over the past 30 days?	◯ Never ◯ Past and shut off ◯ Current
What is the longest period of time they have been seizure-free, with particular focus on the last 30 days?	◯ No seizures ever ◯ Greater than 6 months ◯ Greater than 1 month ◯ Greater than 1 week ◯ Greater than 1 day ◯ Always with daily seizures

How often have epilepsy treatment side effects impacted their daily routines over the past 2 weeks?	◯ None ◯ Tolerable ◯ Intolerable ◯ Unknown
Which of the following movement disorders were they diagnosed with by a medical professional? Please select all that apply.	☐ Dystonia (abnormal fixed position) ☐ Ataxia (muscle coordination issues) ☐ Chorea (jerky involuntary movements affecting especially the shoulders, hips, and face) ☐ Athetosis (involuntary writhing movements) ☐ Tremor ☐ Tics ☐ Parkinsonism ☐ Myoclonus (quick, involuntary muscle jerk) ☐ Other movement disorder ☐ None of the above
Please specify "other":	_____
At what age (in years) were they diagnosed with the movement condition you indicated as "other"?	_____
At what age (in years) were they diagnosed with dystonia?	_____
At what age (in years) were they diagnosed with ataxia?	_____
At what age (in years) were they diagnosed with chorea?	_____
At what age (in years) were they diagnosed with athetosis?	_____
At what age (in years) were they diagnosed with tremor?	_____
At what age (in years) were they diagnosed with tics?	_____
At what age (in years) were they diagnosed with parkinsonism?	_____
At what (in years) were they diagnosed with myoclonus?	_____

Which of the following symptoms, if any, are you aware of them experiencing repeatedly without a clearly explained cause such as pain, infection, or injury? Please select all that apply.

- ☐ Abnormal pupil dilation
- ☐ Excessive sweating
- ☐ Inability to sweat
- ☐ Flushing/splotchy skin
- ☐ Unexplained pain or over-reactivity to normally non-painful things
- ☐ Unusually high pain tolerance
- ☐ Vomiting (cyclic/episodic)
- ☐ Inability to regulate temperature
- ☐ Digestive difficulties
- ☐ Hand/feet bluish in color, clammy, or cold
- ☐ Heart rate irregularity
- ☐ Blood pressure irregularity
- ☐ Dizziness/fainting
- ☐ Urinary retention
- ☐ Sleep disturbances
- ☐ Unexplained irritability/agitation
- ☐ None of the above

Which of the following symptoms, related to their autonomic dysfunction, do they typically experience? Please select all that apply.

- ☐ Abnormal pupil dilation
- ☐ Excessive sweating
- ☐ Inability to sweat
- ☐ Flushing/splotchy skin
- ☐ Unexplained pain or over-reactivity to normally non-painful things
- ☐ Unusually high pain tolerance
- ☐ Vomiting (cyclic/episodic)
- ☐ Inability to regulate temperature
- ☐ Digestive difficulties
- ☐ Hand/feet bluish in color, clammy, or cold
- ☐ Heart rate irregularity
- ☐ Blood pressure irregularity
- ☐ Dizziness / fainting
- ☐ Urinary retention
- ☐ Sleep disturbances
- ☐ Unexplained Irritability/agitation
- ☐ Other (please specify)
- ☐ None of the above

Please specify "other":

_____

Which of the following best explains how they are most recently experiencing these symptoms?

- ◯ They typically experience multiple symptoms simultaneously
- ◯ Sometimes they experience multiple symptoms, but sometimes they experience only a single symptom
- ◯ Typically they only experience a single symptom at a time
- ◯ They only experience one of these symptoms

Have you ever discussed autonomic dysfunction with their doctor(s)?

- ◯ Yes
- ◯ No
- ◯ Not sure

What type of clinician did you discuss autonomic dysfunction with?

_____

What was the outcome of this discussion?

- ◯ Autonomic Dysfunction was ruled out
- ◯ Received diagnosis of Autonomic Dysfunction
- ◯ Inconclusive discussion

At what age (in years) were they diagnosed with autonomic dysfunction?	_____
What type of medical provider diagnosed them with autonomic dysfunction?	_____
At the time of diagnosis with autonomic dysfunction, how was treatment management altered? Please select all that apply.	☐ New medication(s) was/were prescribed ☐ Referral to new specialist ☐ Treatment was not altered
What is their genetic diagnosis?	◯ GRIN1 ◯ GRIN2A ◯ GRIN2B ◯ GRIN2C ◯ GRIN2D ◯ GRIN3A ◯ GRIN3B ◯ KCNQ2 ◯ PCDH19 ◯ SCN1A ◯ SCN2A ◯ SCN8A ◯ SLC6A1 ◯ STXBP1 ◯ They do not have a genetic diagnosis ◯ Don't know ◯ Other
Please specify gene:	_____
What is the name of their genetic variant, if known?	_____ (Please enter as (c. , p.), for example: c.4943G>A, p.Arg1648His. This information may be found in their doctor's letters.)
At what age (in years) did they receive a genetic diagnosis?	_____ (If they received a diagnosis before 1-year-old, please enter "0.")
Which of the following describes the inheritance of their genetic mutation?	◯ De novo (new in the them) ◯ Inherited from a parent ◯ Don't know
Have they been diagnosed with any other pathogenic (disease-causing) genetic changes?	◯ Yes ◯ No ◯ Don't know ◯ Prefer not to say
Please specify gene:	_____

Which of the following types of medical providers do they visit at least once per year? Please select all that apply.

- ☐ Cardiologist
- ☐ Complex care physician
- ☐ Developmental pediatrician
- ☐ Endocrinologist
- ☐ Epileptologist
- ☐ Gastroenterologist
- ☐ Geneticist/Neurogeneticist/Genetic Counselor
- ☐ Neurologist
- ☐ Occupational therapist
- ☐ Ophthalmologist/Neuro-opthalmologist
- ☐ Orthopedics
- ☐ Palliative care
- ☐ Pediatrician
- ☐ Physical therapist
- ☐ Psychiatrist/Neuropsychologist
- ☐ Pulmonologist
- ☐ Speech language pathologist
- ☐ Urologist
- ☐ Other

Please specify "other":

_____

Which of the following medical providers are a source of information about their genetic diagnosis?

- ☐ Cardiologist
- ☐ Complex care physician
- ☐ Developmental pediatrician
- ☐ Endocrinologist
- ☐ Epileptologist
- ☐ Gastroenterologist
- ☐ Geneticist/Neurogeneticist/Genetic Counselor
- ☐ Neurologist
- ☐ Occupational therapist
- ☐ Ophthalmologist/Neuro-opthalmologist
- ☐ Orthopedics
- ☐ Palliative care
- ☐ Pediatrician
- ☐ Physical therapist
- ☐ Psychiatrist/Neuropsychologist
- ☐ Pulmonologist
- ☐ Speech language pathologist
- ☐ Urologist
- ☐ Other

Please specify "other":

_____

Please indicate your level of agreement with the following statement. Use a 100-point scale where 0 = "Do not agree at all" and 100 = "Strongly agree.

I feel I have sufficient resources to learn about their diagnoses.

Do not agree at all — Strongly agree

(Place a mark on the scale above)

Please indicate your level of agreement with the following statement. Use a 100-point scale where 0 = "Do not agree at all" and 100 = "Strongly agree.

I feel we have sufficient access to doctors that are knowledgeable/experienced in treating their diagnoses.

Do not agree at all — Strongly agree

(Place a mark on the scale above)

Please indicate your level of agreement with the following statement. Use a 100-point scale where 0 = "Do not agree at all" and 100 = "Strongly agree.

I feel that we currently have sufficient access to specific treatments for their diagnoses.

Do not agree at all — Strongly agree

(Place a mark on the scale above)

Section 3. Quality-of-life

These questions are about the individual with epilepsy and/or neurodevelopmental disorder's life over the past month. We would like to know how you observe them respond to a range of life experiences.

There are no right or wrong answers - please provide your best answers for them. For each question, please reflect on your observations of their well-being and enjoyment of life over the past month.

Health and well-being

Over the past month, how often have they...

	Never	Rarely	Sometimes	Often	Very Often
Had enough energy to participate in daily routines and activities	○	○	○	○	○
Kept in good general health (e.g. avoided coughs, colds, fever)	○	○	○	○	○
Slept well during the night	○	○	○	○	○
Been alert and aware during the day	○	○	○	○	○

Feelings and Emotions

Over the past month, how often have they...

	Never	Rarely	Sometimes	Often	Very often
Been in a good mood	○	○	○	○	○
Smiled or brightened their facial expression	○	○	○	○	○
Showed happiness through body language (e.g. making eye contact, body facing others)	○	○	○	○	○
Showed cheeky or comical mannerisms (e.g. laughed, giggled)	○	○	○	○	○
Been unsettled without an apparent reason	○	○	○	○	○

Showed aggression (e.g. hitting, kicking, using offensive language, being destructive)	○	○	○	○	○
Appeared upset or angry (e.g. crying, screaming, moving or stiffening the body)	○	○	○	○	○
Become withdrawn with a low mood	○	○	○	○	○
Deliberately hurt themselves	○	○	○	○	○
Expressed discomfort with changes in routine (e.g. carers, school, respite, out-of-home care)	○	○	○	○	○
Showed signs of being anxious or agitated (e.g. teeth grinding, fast breathing, avoidance)	○	○	○	○	○

Family and friends

Over the past month, how often have they...

	Never	Rarely	Sometimes	Often	Very Often
Expressed happiness when they were understood	○	○	○	○	○
Appeared relaxed when making eye contact	○	○	○	○	○
Initiated greetings with people verbally or nonverbally (e.g. eye contact)	○	○	○	○	○
Enjoyed being included	○	○	○	○	○
Enjoyed the social experiences of meal times	○	○	○	○	○
Responded positively when others paid attention to them (e.g. smiled, showed interest)	○	○	○	○	○
Showed pleasure or excitement when looking forward to activities (e.g. going to school, outings, events)	○	○	○	○	○

Activities and the outdoors

Over the past month, how often have they...

	Never	Rarely	Sometimes	Often	Very Often
Enjoyed moving their body (e.g. crawling, walking, swinging, swimming)	○	○	○	○	○

Enjoyed feeling steady or stable during physical activities (e.g. sitting, standing, bike riding)	◯	◯	◯	◯	◯
Enjoyed physical activities (e.g. going out for a walk, swimming, swinging, dancing)	◯	◯	◯	◯	◯
Enjoyed going on outings in the community (e.g. shopping, party, sports, theatre)	◯	◯	◯	◯	◯
Enjoyed spending time outdoors (e.g. contact with water, grass, wind, sunshine)	◯	◯	◯	◯	◯

Daily life

Over the past month, how often have they...

	Never	Rarely	Sometimes	Often	Very Often
Expressed their needs ((e.g. hunger, thirst, toileting)	◯	◯	◯	◯	◯
Made their own choices for activities or things they enjoy (e.g. DVDs, toys)	◯	◯	◯	◯	◯
Helped to complete routine activities (e.g. dressing, feeding)	◯	◯	◯	◯	◯
Enjoyed making things with their hands - can be with help (e.g. building blocks, painting, cooking)	◯	◯	◯	◯	◯
Enjoyed using technology (e.g. computer, tablet, applications on phones)	◯	◯	◯	◯	◯

Please indicate your level of agreement with the following statement. Use a 100-point scale where 0 = "Really worse," 50 = "Same," and 100 = "Really better."

In the last month, OVERALL ISSUES with them are better, worse or the same?

Really worse — Same — Really better

(Place a mark on the scale above)

On average, over the last 30 days, how many good days per week do they have?

A "good" day may be defined as: minimally disrupted by seizures or engaged, interactive, able to finish therapies throughout the day.

◯ Almost always, this has been a good month
◯ Only a few days this past month
◯ More than half of the days per week are good
◯ Always at least 2 or 3 days per week
◯ Maybe 1 or 2 good days per week
◯ Never has any good days per week

Please indicate your level of agreement with the following statement. Use a 100-point scale where 0 = "Really worse," 50 = "Same," and 100 = "Really better."

In the last month, the ADAPTABILITY OR RESILIENCE (coping) of caregiver(s) at home is better, worse or the same?

Really worse Same Really better

(Place a mark on the scale above)