KCNMA1 Channelopathy International Advocacy Foundation
(KCIAF.org)

Instructions

Thank you for taking the time to enroll with the CoRDS Registry. This module will ask you questions specific to your diagnosis. The questions below were developed in partnership with the KCNMA1 Channelopathy International Advocacy Foundation (KCIAF). Please note, this module:

- Takes approximately 30-45 minutes to complete
- Will refer to the person with the diagnosis as "**the participant**"
- Survey responses can be updated at any time by logging in to the CoRDS online portal or by contacting CoRDS personnel

If you have any questions while completing this form, please contact CoRDS at (877) 658-9192 during business hours, 8:30am-5:00pm (CST) Monday through Friday. If you need assistance after business hours, please leave a message or email cords@sanfordhealth.org.

Permissions & Data Sharing

I give permission to CoRDS to provide my information that may or may not be identifiable to the following Patient Advocacy Group (PAG) for non-research purposes.

☐ KCNMA1 Channelopathy International Advocacy Foundation	☐ I do not give my permission

Diagnosis/Assessment

1. Has the participant ever had genetic testing

☐ Yes
☐ No – Genetic testing was offered but the participant has not been tested due to costs
☐ No – Genetic testing was offered but the participant/guardian declined
☐ No – Genetic testing was offered but the participant has not done it yet for other reasons
☐ No – The healthcare provider has not talked about genetic testing for the participant
☐ No – The healthcare provider said that genetic testing is not required
☐ No – Other
☐ Unknown

If "other", please specify:

2. Which types of DNA (molecular) genetic tests have been performed? (Select all that apply)

☐ Epilepsy gene panel	☐ Exon/gene deletion/duplication panel (aka microarray)
☐ Paroxysmal dyskinesia or movement disorder gene panel	☐ Other
☐ Whole genome sequencing (WGS)	☐ Unknown
☐ Whole exome sequencing (WES)	☐ None
☐ Mitochondrial analysis	

If "other", please specify:

3. Please select the genetic defect that applies to the participant (Select all that apply)

☐ KCNMA1 Mutation(s)	☐ Other Genetic Mutation(s)	☐ No genetic defect

If KCNMA1 mutation was selected, please write out the complete specific mutation name(s) from the genetics report. For example, it may look something like:

Example 1 -- NM_001014797.2 (KCNMA1):c.1301A>G (p.Asp434Gly)
Example 2 -- NM001014797.2 chr10:g.78651467T>C(GRCh37)c.2996A>Gp.Asn999Ser

Enter here: _____
Is the mutation listed as heterozygous or homozygous? ☐ Heterozygous ☐ Homozygous ☐ Unknown

If "Other Genetic Mutation(s)" was selected in question 3, please indicate the name of the mutation(s), writing out the mutation description as in the examples above:

Enter here: _____
Is the mutation listed as heterozygous or homozygous? ☐ Heterozygous ☐ Homozygous ☐ Unknown

4. Is the participant's genetic mutation inherited from a parent, or considered "*de novo*"

☐ *De novo*/sporadic (meaning, both birth parents had genetic testing and neither have the participants genetic problem)
☐ Inherited from Father
☐ Inherited from Mother
☐ Inherited from both birth parents
☐ Unsure (one or both parents did not have genetic testing performed)

5. Would the participant be able to provide a copy of the genetic report upon request?

☐ Yes	☐ No	☐ Unsure

6. Select whether the participant had any of the following tests performed. Enter the results and age at the time the test was performed. Select all that apply. If the same type of test has been performed several times (for example, if there have been multiple EEGs) please select the MOST RECENT results and age at which the test was performed."

Neurological Test	Had test, normal results	Had test, abnormal	Had test, results	Test not performed	Unknown	Age at time of test
Computed Tomography (CT or CAT scan) of the brain and/or spinal cord	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Magnetic Resonance Imaging (MRI) of the brain and/or spinal cord	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Electroencephalogram (EEG)	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+

Electromyogram (EMG)	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Functional Magnetic Resonance Imaging (fMRI)	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Head Ultrasound	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Nerve Conduction Studies (NCS)	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Magnetoencephalography (MEG)	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Muscle Biopsy	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Positron Emission Tomography (PET)	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Spinal Tap / Lumbar Puncture	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+

Echocardiogram (ultrasound of the heart)	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+
Electrocardiogram (ECG or EKG)	☐	☐	☐	☐	☐	☐ <1 (yr) ☐ 1-2 (yrs) ☐ 3-5 (yrs) ☐ 6-10 (yrs) ☐ 11-17 (yrs) ☐ 18+

Comments (describe any abnormal results if possible):

7. Would the participant be able to provide a copy of the above test results upon request?

☐ Yes	☐ No	☐ Unsure

8. Which specialist(s) has the participant seen? (Select all that apply)

☐ Neurologist ☐ Geneticist ☐ Primary Care Pediatrician ☐ Psychologist/Psychiatrist ☐ Orthopedic (bone/joint/muscle doctor) ☐ Cardiologist (heart doctor) ☐ Gastroenterologist (stomach/gut doctor)	☐ Endocrinologist ☐ Ophthalmologist ☐ Otolaryngologist (ENT) ☐ Pulmonologist (lung doctor) ☐ Nephrologist (kidney doctor) ☐ Physiatrist (aka physical medicine and rehabilitation doctor) ☐ Other

If "other", please specify:

Demographics

9. What is the height of the participant?

_____ inches (in)	_____ centimeters (cm)

10. What is the weight of the participant?

_____ pounds (lbs.)	_____ kilograms (kg)

11. What is the participant's most recent head circumference?

_____ inches (in)	_____ centimeters (cm)	☐ Unknown

12. What is the participant's current living situation?

☐ Dependent with parent(s) or relative(s)	☐ Semi-independent with limited assistance from parent(s), relative(s), or friend(s)
☐ Independent (alone)	☐ Independent (with housemate)

Comments:

13. What is the current or highest level of education that the participant completed?		
☐ No schooling completed	☐ Elementary School	☐ Middle School
☐ High School	☐ College/University	☐ Unknown

Birth History (Please provide information about the birth of the participant in this section)

14. What was the participant's gestational age? (Gestational age refers to how far along the pregnancy was at the time of the participant's birth)	
_____ weeks	☐ Unknown

15. How was the participant delivered at birth (vaginal or caesarian section)?	
☐ **Vaginal**	☐ **Caesarian**

16. If a C-section was performed, was it due to any of the following reasons? (Select all that apply)	
☐ Planned C-section ☐ Baby presented breech or transverse or upside down ☐ Cephalopelvic disproportion (meaning the size of the baby's head was too large for the mother's pelvis) ☐ Concerns about the mother's ability to deliver vaginally ☐ Emergency (fetal/baby distress or problem)	☐ Emergency (maternal distress or problem) ☐ Failure to progress (the baby was not coming down the birth canal) ☐ Problems with the umbilical cord (knotted, collapsed, wrapped around the baby, other) ☐ Other ☐ Unknown

If "other", please specify:

17. What was the participant's birth weight?		
_____ (lbs.) _____ ounces	_____._____ (kg)	☐ Unknown

18. What was the participant's birth length?		
_____ (in)	_____ (cm)	☐ Unknown

19. What was the participant's head circumference at birth? (Please round to the nearest half inch or centimeter)		
_____ (in)	_____ (cm)	☐ Unknown

20. How many days did the participant spend in the neonatal intensive care unit (NICU; not the nursery) after the participant's birth?		
_____ (months) _____(days)	☐ Did not stay in the NICU	☐ Unknown

Development

21. **At what age were the following communication milestones achieved? (Please answer in terms of "months" if the participant was 24 months or younger. Please answer in "years" if the participant was 2 or older).**

Milestone	Age	Has not yet reached	Unknown

Social smile (smiled at other faces)	_____ (years) **OR** _____ (months)	☐	☐
Single syllable utterances (i.e. ma)	_____ (years) **OR** _____ (months)	☐	☐
First verbal word (i.e. go)	_____ (years) **OR** _____ (months)	☐	☐
First two-word verbal sentence (i.e. car go)	_____ (years) **OR** _____ (months)	☐	☐
Recognizes written letters/numbers	_____ (years) **OR** _____ (months)	☐	☐
Phonics-style reading (sounds out words)	_____ (years) **OR** _____ (months)	☐	☐
Read whole words	_____ (years) **OR** _____ (months)	☐	☐
Write/type using a keyboard	_____ (years) **OR** _____ (months)	☐	☐

22. At what age were the following self-help skills achieved? (Please answer in terms of "months" if the participant was 24 months or younger. Please answer in "years" if the participant was 2 or older).

Milestone	Age	Has not yet reached	Unknown
Able to undress without assistance	_____ (years) **OR** _____ (months)	☐	☐

Able to dress without assistance	_____ (years) **OR** _____ (months)	☐	☐
Toilet-trained (during the day)	_____ (years) **OR** _____ (months)	☐	☐
Toilet-trained (during the night)	_____ (years) **OR** _____ (months)	☐	☐
Able to drink independently	_____ (years) **OR** _____ (months)	☐	☐

23. At what age were the following motor milestones achieved? (Please answer in terms of "months" if the participant was 24 months or younger. Please answer in "years" if the participant was 2 or older).

Milestone		**Has not yet reached**	**Unknown**
Hold head up on his/her own	_____ (years) **OR** _____ (months)	☐	☐
Roll over back to stomach	_____ (years) **OR** _____ (months)	☐	☐
Sit when placed	_____ (years) **OR** _____ (months)	☐	☐
Crawl on hands and knees	_____ (years) **OR** _____ (months)	☐	☐
Walk unassisted	_____ (years) **OR** _____ (months)	☐	☐

Climb stairs standing up without help	_____ (years) **OR** _____ (months)	☐	☐
Descend stairs without help	_____ (years) **OR** _____ (months)	☐	☐
Jump with both feet	_____ (years) **OR** _____ (months)	☐	☐
Pedal a tricycle	_____ (years) **OR** _____ (months)	☐	☐

Puberty

For FEMALES

24. How old was the participant when she got her first menstrual period?

_____ (yrs)	☐ Has not occurred yet	☐ Unknown

For MALES

25. How old was the participant when he first developed pubic hair?

_____ (yrs)	☐ Has not occurred yet	☐ Unknown

Symptoms

26. Has the participant ever experienced epileptic seizures?

☐ Yes	☐ No	☐ Unknown

27. If yes, at what age did the epileptic seizures start? (Please answer in terms of "months" if the participant was 24 months or younger. Please answer in "years" if the participant was 2 or older).

_____ (years) **OR** _____ (months)	☐ Unknown

28. If there are seizures, which of the following seizure diagnosis was given that best describes the seizures that the participant experiences or has ever experienced? (Select all that apply)

☐ Absence seizure/staring spells (petit mal seizures)
☐ Myoclonic seizure
☐ Generalized tonic-clonic seizure (GTCS or grand mal seizures)
☐ Atonic seizure
☐ Infantile spasms
☐ Other
☐ Unknown

If "other", please specify:

29. Which of the following best describes how often the participant currently experiences or experienced their seizures?

☐ No seizures in > 6 months	☐ No seizures in the last 6 months	☐ 1 to 3 seizures per month
☐ 4 to 24 seizures per month	☐ 25 to 100 seizures per month	☐ > 100 seizures per month
☐ at least one seizure per day	☐ Other	☐ Unknown

If "other", please specify:

30. If the participant is no longer experiencing seizures, at what age did the seizures stop? (Please answer in terms of "months" if the participant was 24 months or younger. Please answer in "years" if the participant was 2 or older).

_____ (years) **OR** _____ (months)	☐ Unknown

31. Which of the following medications is the participant currently taking, or has ever taken, for <u>epileptic seizures</u>? List daily medication only, and not rescue or "as needed" medication. (Please answer in terms of "months" if the participant was 24 months or younger. Please answer in "years" if the participant was 2 or older).

Medication	Age at the time the medication was started	Currently taking, helps with symptoms	Currently taking, not sure if it helps	Tried in the past, but did not work	Tried in the past, had side effects so was stopped	Tried in the past, unsure why it was stopped	Tried in the past, made symptoms worse	Unknown ☐
Phenobarbital	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Phenytoin / Dilantin	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐

Valproic Acid / Depakote / Depakene	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Levetiracetam / Keppra	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Lamotrigine / Lamictal	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Carbamazepine / Tegretol	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Oxcarbazepine / Trileptal	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Clobazam / Onfi	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Zonisamide / Zonegran	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Ethosuximide / Zarontin	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Topiramate / Topamax	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Clonazepam / Klonapin	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐

If not listed above (please write-in the name below)								
	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐

Comments:

32. Has the participant ever experienced episodes of abnormal movements of muscle function?

☐ Yes	☐ No	☐ Unknown

33. If yes, at what age did the movement problems start? (Please answer in terms of "months" if the participant was 24 months or younger. Please answer in "years" if the participant was 2 or older).

_____ (years) **OR** _____ (months)	☐ Unknown

34. Which of the following best describes the abnormal muscle movements or other symptoms the participant experiences or has ever experienced? (Select all that apply)

☐ Freezes or falls while going **limp** ☐ Freezes or falls while going **stiff** ☐ Freezes or falls (unsure if stiff, or limp, or can depend) ☐ Abnormal eye movements ☐ Abnormal mouth movements ☐ "Drop Attacks" ☐ Muscles too loose (in general) ☐ Muscles too tight (in general)	☐ Drooling ☐ Muscle weakness ☐ Tremor ☐ Hyperkinetic (e.g. overactive) body movements ☐ Spasms ☐ Other ☐ Unsure or no abnormal movements

If "other", please specify:

35. Which of the following, if any, has the participant ever received as a diagnosis by a medical professional?

☐ Spasticity
☐ Dystonia
☐ Hypotonia
☐ Myoclonus
☐ Chorea/choreoathetosis
☐ Dyskinesias
☐ None of the above
☐ Unsure

36. Which of the following best describes how often the participant currently experiences or experienced their movement attacks?

☐ None in > 6 months	☐ None in the last 6 months	☐ 1 to 3 per month
☐ 4 to 24 per month	☐ 25 to 100 per month	☐ 100-150 per month
☐ More than 5 per day on average	☐ Other	☐ Unknown

If "other", please specify:

37. If the participant is no longer experiencing movement attacks, at what age did they stop? (Please answer in terms of "months" if the participant was 24 months or younger. Please answer in "years" if the participant was 2 or older).

_____ (years) **OR** _____ (months)	☐ Unknown

38. Which of the following medications is the participant currently taking or has ever taken for <u>movement attacks?</u> (Please answer in terms of "months" if the participant is 24 months or younger. Please answer in "years" if the participant is 2 or older).

Medication	Age at the time the medication was started	Currently taking, helps with symptoms	Currently taking, not sure if it helps	Tried in the past, but did not work	Tried in the past, had side effects so was stopped	Tried in the past, unsure why it was stopped	Tried in the past, made symptoms worse ☐	Unknown
Acetazolamide / Diamox	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Lamotrigine / Lamictal	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐

Carbamazepine / Tegretol	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
Clonazepam / Klonapin	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
If not listed above (please write-in the name below)								
	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐
	_____ (years) **OR** _____ (months)	☐	☐	☐	☐	☐	☐	☐

Comments:

39. Does the participant have any of the following problems with heart rate or blood pressure?

☐ sudden unexplained increases in heart rate	☐ sudden unexplained increases in blood pressure
☐ sudden unexplained decreases in heart rate	☐ sudden unexplained decreases in blood pressure
☐ heart rate is too high, most of the time (for example, does not come and go but is usually high every day)	☐ blood pressure is too high, most of the time (for example, does not come and go but is usually high every day
☐ heart rate is too low, most of the time	☐ blood pressure is too low, most of the time
☐ none of the above (heart rate is typically ok)	☐ none of the above (blood pressure is typically ok)

Comments:

40. What was the participant's last recorded heart and blood pressure?

Heart Rate _____ (BPM)	Blood Pressure _____ /_____ (mmHg)	☐ Unknown

41. Does the participant have any of the following skin conditions? (Select all that apply)

☐ episodes of turning blue or “mottling” of the skin
☐ episodes of redness or flushing
☐ sudden increases in sweating
☐ too much sweating, all the time
☐ too little sweating, or inability to sweat
☐ none of the above
☐ other skin conditions

If “other”, please specify:

42. Which of the following "neuropsychiatric" or ‘neurocognitive’ conditions does the participant have or has ever been diagnosed with had? (Select all that apply)

☐ Depression
☐ Anxiety or anxiety disorder
☐ Autism
☐ ADD / ADHD
☐ Learning or intellectual disability
☐ Aggressive outbursts
☐ Crying for no reason (even though he/she is not sad)
☐ Laughing for no reason (even when he/she is not happy or responding to something funny)
☐ Failure to thrive
☐ Other
☐ Unsure

If “other”, please specify:

43. Does the participant have or has ever had any other symptoms described below? (Select all that apply)

☐ Vision or eyesight problems
☐ Hearing problems
☐ Speech delay
☐ Sleep disturbances
☐ Bladder dysfunction (infections, incontinence, etc.)
☐ Gastrointestinal abnormalities (for example vomiting, diarrhea, constipation, fecal incontinence, etc.)
☐ Breathing difficulties
☐ Other
☐ Unsure
☐ None of the above

If “other”, please specify: