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

Genetic Testing

1. Has the participant ever had genetic testing

☐ Yes
☐ No
☐ Unknown

If “other”, please specify:

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

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

If “other”, please specify:

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

☐ KCNMA1 Variant(s)
☐ Other Genetic Variant(s)
☐ No genetic defect
If KCNMA1 variant was selected, please write out the complete specific variant 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.Asnn999Ser

Enter here: ____________________________________________________

Is the variant listed as heterozygous or homozygous?  
☐ Heterozygous  ☐ Homozygous  ☐ Unknown

If “Other Genetic Variant(s)” was selected in question 3, please indicate the name of the variant(s), writing out the variant description as in the examples above:

Enter here: ____________________________________________________

Is the variant listed as heterozygous or homozygous?  
☐ Heterozygous  ☐ Homozygous  ☐ Unknown

4. Is the participant’s genetic variant 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? If yes, please upload the report upon completion of the CORDs survey.

☐ Yes  ☐ No

6. 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:

---

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

7. 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

8. How was the participant delivered at birth (vaginal or caesarian section)?

☐ Vaginal  ☐ Caesarian

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

If “other”, please specify:

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

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

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

13. 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

Seizures

14. Does the participant have epileptic/seizure episodes currently or in the past?
   
   ☐ Yes  ☐ No  ☐ Unknown

15. At what age was the participant’s first epileptic/seizure episode?
   
   _______   years   _______ months

16. Does the participant currently display signs or symptoms immediately before epileptic/seizure activity begins?
   
   ☐ Yes, date (DD/MM/YYYY) of diagnosis?  ☐ No  ☐ Unknown

17. Has the participant ever been formally diagnosed with an epileptic/seizure disorder?
   
   ☐ Yes, date (DD/MM/YYYY) of diagnosis?  ☐ No  ☐ Unknown

18. If yes, what type of epileptic/seizure disorder(s) has the participant been diagnosed with? (Check all that apply)
   
   ☐ Unknown
| ☐ Absence/petit mal | ☐ Febrile |
| ☐ Myoclonic       | ☐ Tonic-clonic |
| ☐ Atonic          | ☐ Grand mal/Generalized |
| ☐ Clonic          | ☐ Myoclonic-atonic |
| ☐ Partial Complex | ☐ Unknown |
| ☐ Tonic           | ☐ Other |

If “other” please specify:

19. Has the participant ever had an EEG epileptic/seizure disorder analyses? Year?

20. Which of the following best describes the participant’s epileptic/seizures?

| ☐ loss of consciousness or responsiveness | ☐ Evolving to a bilateral convulsive seizure |
| ☐ motor or autonomic components            | ☐ Unknown                                    |
| ☐ with sensory or psychic phenomena only   |                                            |

21. Which of the following best describes the duration of the participant’s epileptic/seizure episodes?

| ☐ Several seconds (brief) | ☐ Evolving to a bilateral convulsive seizure |
| ☐ Less than 3 minutes (short) | ☐ Unknown |
| ☐ Less than 15 minutes (prolonged) |                                            |
| ☐ Greater than 30 minutes |                                            |
| ☐ Status epilepticus, how many times? (#) |                                            |

22. How frequently does the participant experience epileptic/seizure episodes?

Example 1: 1 time a day, 30 times a month, 12 times a year (participant experiences one seizure every day)

Example 2: 3 times a day, 1 time a month, 5 times a year (participant experiences three seizures in a day, once a month for five months out of the year)

__________ time(s) a day ______________ time(s) a month _____________ time(s) a year

Comments:
23. Does the participant have a family history of epilepsy/seizure disorder?

<table>
<thead>
<tr>
<th>☐ Yes</th>
<th>☐ No</th>
<th>☐ Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

If yes, please describe (Example: Family member – diagnosis)

24. Does the participant receive medication for epilepsy/seizure disorder?

<table>
<thead>
<tr>
<th>☐ Yes</th>
<th>☐ No</th>
<th>☐ Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

If yes, what is the name of the medication(s)?

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

25. In the past, has the participant tried medications for an epilepsy/seizure diagnosis that did not help?

<table>
<thead>
<tr>
<th>☐ Yes</th>
<th>☐ No</th>
<th>☐ Unknown</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

If yes, what is the name of the medication(s)?

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________

Name: ___________________________
Dose: __________________________
Frequency: ____________________
### Movement Disorder

29. Does the patient have movement disorder episodes currently or have they in the past?
- [ ] Yes
- [ ] No
- [ ] Unknown

30. At what age was the participant’s first movement disorder ‘attack’ first noticed?
   - [ ] [________ years ________ months]

31. Does the participant currently display sensory or physiological symptoms immediately before a movement disorder activity begins?
- [ ] Yes
- [ ] No
- [ ] Unknown

32. Has the participant received a formal movement disorder diagnosis?
- [ ] Yes,
- [ ] No
- [ ] Unknown

33. If yes, please list movement disorder diagnose(s)
   - Diagnosis: __________________________________________ Date received: _______________________
   - Diagnosis: __________________________________________ Date received: _______________________
   - Diagnosis: __________________________________________ Date received: _______________________
   - Diagnosis: __________________________________________ Date received: _______________________

34. Has the participant ever received any of the following movement disorder analyses? (Check all that apply)
- [ ] Gait analysis Year? ______________
- [ ] Electromyography (EMG) Year? ______________
- [ ] Muscle biopsy Year? ______________
- [ ] Nerve conduction studies Year? ______________

35. What type of movement disorder symptoms has the participant been diagnosed with? (Check all that apply)
- [ ] Spasticity
- [ ] Dystonia
- [ ] Hypotonia
- [ ] Myoclonus
- [ ] Chorea/choreoathetosis
- [ ] Dyskinesias
- [ ] Unknown
- [ ] Other
If “other” please specify:

36. Which of the following best describes the participant’s movement disorder episodes?

| ☐ | affecting the whole body |
| ☐ | affecting walking only |
| ☐ | affecting isolated parts of the body |

Please list the affected parts:
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________

☐ triggered by specific stimuli

Please list the specific stimuli:
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________
____________________________________________________________________

☐ present all the time

☐ spontaneous onset without triggers

☐ Unknown

37. Which of the following best describes the duration of the participants movement disorder episodes?

| ☐ | Several seconds (brief) |
| ☐ | Less than 3 minutes (short) |
| ☐ | Less than 15 minutes (prolonged) |
| ☐ | Greater than 30 minutes |
| ☐ | Tetanic muscle stiffness |

38. How frequently does the participant experience movement disorder episodes?

Example 1: 1 time a day, 30 times a month, 12 times a year (participant experiences one episode every day)
Example 2: 3 times a day, 1 time a month, 5 times a year (participant experiences three episodes in a day once a month for five months out of the year)

_________ time(s) a day ____________ time(s) a month _____________ time(s) a year

Comments:

39. Does the participant have a family history of movement disorders?

☐ Yes  ☐ No  ☐ Unknown

If yes, please describe (Example: Family member – diagnosis)

40. Does the participant receive medication for a movement disorder?

☐ Yes  ☐ No  ☐ Unknown
41. In the past, has the participant tried medications for a movement disorder diagnosis that did not help?

☐ Yes
☐ No
☐ Unknown

If yes, what is the name of the medication(s)?
Name:___________________________________ Dose:__________________________ Frequency:________________
Name:___________________________________ Dose:__________________________ Frequency:________________
Name:___________________________________ Dose:__________________________ Frequency:________________
Name:___________________________________ Dose:__________________________ Frequency:________________

42. Does the participant require assistive devices for a movement disorder diagnosis? (Check all that apply)

☐ None
☐ Braces/Crutches
☐ Walker
☐ Wheelchair
☐ Helmet
☐ Other

If “other” please specify:

43. Is there anything that makes the movement disorder worse? ________________________________

44. Is there anything that makes the movement disorder better? ________________________________

45. When do the movement disorder episodes most commonly occur? ___________________________
46. Is there anything else we should know about the participants movement disorder history?

---

### Sleep

47. Has the participant been formally diagnosed with a sleep disorder?

- [ ] Yes
- [ ] No
- [ ] Unknown

48. Has the participant undergone sleep studies with a sleep specialist?

- [ ] Yes
- [ ] No
- [ ] Unknown

49. If yes, please list sleep disorder diagnose(s)

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Date received</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>

50. How frequently does the participant sleep problems?

- Example 1: 1 time a night, 30 times a month, 12 times a year (participant experiences one episode every night)
- Example 2: 3 times a night, 1 time a month, 5 times a year (participant experiences three episodes a night once a month for five months out of the year)

- ________ time(s) a night__________ time(s) a month ___________ time(s) a year

If “other” please specify:
51. Is there anything else we should know about the participants sleep history?

---

### Development

52. What is the participants current living situation?

<table>
<thead>
<tr>
<th>Option</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐</td>
<td>Dependent with parent(s) or relative(s)</td>
</tr>
<tr>
<td>☐</td>
<td>Independent (alone)</td>
</tr>
<tr>
<td>☐</td>
<td>Semi-independent with limited assistance from parent(s), relative(s) or friend(s)</td>
</tr>
<tr>
<td>☐</td>
<td>Independent (with housemate)</td>
</tr>
</tbody>
</table>

Comments:

---

53. What is the current or highest level of education that the participant has completed?

<table>
<thead>
<tr>
<th>Option</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐</td>
<td>Too young to attend school</td>
</tr>
<tr>
<td>☐</td>
<td>Unable to attend school</td>
</tr>
<tr>
<td>☐</td>
<td>Unknown</td>
</tr>
<tr>
<td>☐ Elementary School (K-5(^{th}) grade)</td>
<td></td>
</tr>
<tr>
<td>☐ Middle School (6(^{th})-8(^{th}) grade)</td>
<td></td>
</tr>
<tr>
<td>☐ High School (9th – 12(^{th}) grade)</td>
<td></td>
</tr>
<tr>
<td>☐ College/University</td>
<td></td>
</tr>
</tbody>
</table>

54. Has the participant ever been diagnosed, or is in the process of being diagnosed with intellectual disability? (Check all that apply)

<table>
<thead>
<tr>
<th>Option</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐</td>
<td>Speech Delay</td>
</tr>
<tr>
<td>☐</td>
<td>Autism Spectrum Disorder (ASD)</td>
</tr>
<tr>
<td>☐</td>
<td>Attention Deficit Disorder (ADD/ADHD)</td>
</tr>
<tr>
<td>☐</td>
<td>Dyslexia</td>
</tr>
<tr>
<td>☐ Visual Processing Disorders</td>
<td></td>
</tr>
<tr>
<td>☐ Auditory Processing Disorders</td>
<td></td>
</tr>
<tr>
<td>☐ Unknown</td>
<td></td>
</tr>
<tr>
<td>☐ Other</td>
<td></td>
</tr>
</tbody>
</table>

If “other” please specify:

---

55. Does the participant use an individualized development plan (IDP)?

<table>
<thead>
<tr>
<th>Option</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐</td>
<td>Yes</td>
</tr>
<tr>
<td>☐ No – but planning to begin this year</td>
<td></td>
</tr>
<tr>
<td>☐ No – participant is not school aged</td>
<td></td>
</tr>
<tr>
<td>☐ No – participant is beyond school aged</td>
<td></td>
</tr>
</tbody>
</table>

56. Does the participant take medications for a diagnosis of intellectual disability?
**Structural Changes**

<table>
<thead>
<tr>
<th>58. Has the participant been formally diagnosed with any structural malformations? (brain, spinal or skeletal)</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ Yes</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>59. If yes, please list structural malformation diagnose(s)</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Diagnosis: _____________________________________________</td>
<td>Date received: ______________________</td>
</tr>
<tr>
<td>Diagnosis: _____________________________________________</td>
<td>Date received: ______________________</td>
</tr>
<tr>
<td>Diagnosis: _____________________________________________</td>
<td>Date received: ______________________</td>
</tr>
<tr>
<td>Diagnosis: _____________________________________________</td>
<td>Date received: ______________________</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>60. Has the participant received any of the following tests to diagnose structural malformation analyses?</th>
<th>Year?</th>
</tr>
</thead>
<tbody>
<tr>
<td>☐ Computed Tomography (CT or CAT) of the brain and/or spinal chord</td>
<td>___________</td>
</tr>
<tr>
<td>☐ Magnetic Resonance Imaging (MRI) of the brain and/or spinal chord</td>
<td>___________</td>
</tr>
<tr>
<td>☐ Functional Magnetic Resonance Imaging (fMRI) of the brain and/or spinal chord</td>
<td>___________</td>
</tr>
<tr>
<td>☐ Head Ultrasound (US)</td>
<td>___________</td>
</tr>
<tr>
<td>☐ Positron Emission Tomography (PET) scan</td>
<td>___________</td>
</tr>
<tr>
<td>☐ Echocardiogram (ultrasound of the heart)</td>
<td>___________</td>
</tr>
<tr>
<td>☐ Electrocardiogram (ECG or EKG)</td>
<td>___________</td>
</tr>
<tr>
<td>☐ Spinal Tap/Lumbar Puncture</td>
<td>___________</td>
</tr>
</tbody>
</table>

57. Is there anything else we should know about the participants intellectual disability/developmental delay history?  

If yes, what is the name of the medication(s)?

<table>
<thead>
<tr>
<th>Name: _____________________________________________</th>
<th>Dose: ___________________________</th>
<th>Frequency: __________________</th>
</tr>
</thead>
<tbody>
<tr>
<td>Name: _____________________________________________</td>
<td>Dose: ___________________________</td>
<td>Frequency: __________________</td>
</tr>
<tr>
<td>Name: _____________________________________________</td>
<td>Dose: ___________________________</td>
<td>Frequency: __________________</td>
</tr>
<tr>
<td>Name: _____________________________________________</td>
<td>Dose: ___________________________</td>
<td>Frequency: __________________</td>
</tr>
</tbody>
</table>

☐ Yes  ☐ No  ☐ Unknown
### Other Symptoms

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

- [ ] Bladder dysfunction
- [ ] Gastrointestinal abnormalities
- [ ] Endocrine abnormalities
- [ ] Breathing difficulties
- [ ] Behavioral/conduct disorders/psychiatric
- [ ] Depression
- [ ] Anxiety or anxiety disorder
- [ ] Failure to thrive
- [ ] Reproductive problems/complications
- [ ] Prolonged hospitalization
- [ ] Other
- [ ] None of the above

For each selected please specify the diagnoses:

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

- [ ] Sudden unexplained increases in heart rate
- [ ] Sudden unexplained decreases in heart rate
- [ ] Heart rate is too high, most of the time
- [ ] Heart rate is too low, most of the time
- [ ] None of the above (heart rate is typically ok)

- [ ] Sudden unexplained increases in blood pressure
- [ ] Sudden unexplained decreases in blood pressure
- [ ] Blood pressure is too high, most of the time
- [ ] Blood pressure is too low, most of the time
- [ ] None of the above (blood pressure is typically ok)

63. Please describe any other persistent disability not covered in the previous questions.