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)	☐ 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.Asn999Ser

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)

☐ 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:	

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)		

☐ Absence/petit mal ☐ Myoclonic ☐ Atonic ☐ Clonic ☐ Partial Complex ☐ Tonic	☐ Febrile ☐ Tonic-clonic ☐Grand mal/ Generalized ☐ Myoclonic-atonic ☐ Unknown ☐ Other
If "other" please specify:	

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

20. Which of the following best describes the participant's epileptic/seizures?	
☐ loss of consciousness or responsiveness ☐ motor or autonomic components ☐ with sensory or psychic phenomena only	☐ Evolving to a bilateral convulsive seizure ☐ Unknown

21. Which of the following best describes the duration of the participants epileptic/seizure episodes?
☐ Several seconds (brief) ☐ Less than 3 minutes (short) ☐ 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?		
☐ Yes	☐ No	☐ Unknown
If yes, please describe (Example: Family member – diagnosis)		

24. Does the participant receive medication for epilepsy/seizure disorder?
☐ 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:__________________
25. In the past, has the participant tried medications for an epilepsy/seizure 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:__________________

26. What factors are associated with the participants' seizures? 27. When are they most likely to occur?
28. Is there anything else we should know about the participants epilepsy/seizure history?

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

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

Name:______________________________________ Dose:____________________________ Frequency:_________________

Name:______________________________________ Dose:____________________________ Frequency:_________________

Name:______________________________________ Dose:____________________________ Frequency:_________________

Name:______________________________________ Dose:____________________________ Frequency:_________________

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		☐ **Unknown**

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

☐ Yes	☐ No	☐ Unknown

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

Diagnosis:____ Date received: _________________________

Diagnosis:____ Date received: _________________________

Diagnosis:____ Date received: _________________________

Diagnosis:____ Date received: _________________________

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?

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

Comments:

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

☐ Too young to attend school ☐ Unable to attend school ☐ Unknown ☐ Elementary School (K-5th grade)	☐ Middle School (6th-8th grade) ☐ High School (9th – 12th grade) ☐ College/University

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

☐ Speech Delay ☐ Autism Spectrum Disorder (ASD) ☐ Attention Deficit Disorder (ADD/ADHD) ☐ Dyslexia	☐ Visual Processing Disorders ☐ Auditory Processing Disorders ☐ Unknown ☐ Other

If "other" please specify:

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

☐ Yes ☐ No – but planning to begin this year	☐ No – participant is not school aged ☐ No – participant is beyond school aged

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

☐ 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:_____________

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

Structural Changes

58. Has the participant been formally diagnosed with any structural malformations? (brain, spinal or skeletal)

☐ Yes	☐ No	☐ Unknown

59. If yes, please list structural malformation diagnose(s)

Diagnosis:_____________________________________ Date received: ____________________

Diagnosis:_____________________________________ Date received: ____________________

Diagnosis:_____________________________________ Date received: ____________________

Diagnosis:_____________________________________ Date received: ____________________

60. Has the participant received any of the following tests to diagnose structural malformation analyses?

Test	Year
☐ Computed Tomography (CT or CAT) of the brain and/or spinal chord	Year? ____________
☐ Magnetic Resonance Imaging (MRI) of the brain and/or spinal chord	Year? ____________
☐ Functional Magnetic Resonance Imaging (fMRI) of the brain and/or spinal chord	Year? ____________
☐ Head Ultrasound (US)	Year? ____________
☐ Positron Emission Tomography (PET) scan	Year? ____________
☐ Echocardiogram (ultrasound of the heart)	Year? ____________
☐ Electrocardiogram (ECG or EKG)	Year? ____________
☐ Spinal Tap/Lumbar Puncture	Year? ____________

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 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	☐ blood pressure is too high, most of the time
☐ 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)

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