KCNT1 Demographics and Genetics Survey 1.0

Participant Name:_____ Doctor Name:_____

Parent Name: _____

For Study Personnel Use, Only:
Coordinator Code: _____ Date:_____

1. Person filling out this form and your relation to the participant

- ☐ Primary caregiver of someone with KCNT1 (living or deceased)
- ☐ Self (adult) living with KCNT1
- ☐ Clinical provider of someone with KCNT1

If you are answering for yourself, for the remainder of this survey, questions will be asked about "your child", but since you are an adult with a KCNT1 gene mutation, please answer the questions about YOURSELF. If you have questions, please contact us directly at info@kcnt1epilepsy.org.

2. Please select your child's sex at birth. ☐ **Male** ☐ **Female** ☐ **Prefer not to answer**

3. Was your child born prematurely? ☐ **Male** ☐ **Female** ☐ **I'm not sure**

4. If yes, how many weeks early was your child born? _____ **Weeks**

5. Describe any prenatal issues or birth complications, or write "none"

_____ _____ _____ _____

7. 7a. What is your child's age today? If under the age of 2 years, indicate how many weeks or months old. _____

8. In what country was your child born?_____

9. Does your child currently live in the United States?

☐ Yes ☐ No

9a. In which U.S. state or territory does your child currently live?

_____

10. Does your child have a CRID number?

☐ Yes ☐ No

11. In addition to KCNT1, has your child ever experienced or been diagnosed with any of the following conditions?

___None
___Aicardi Syndrome
___Autism Spectrum Disorder
___Autonomic Disorder or Dysautonomia
___Brugada Syndrome
___Coppula-Dulac Syndrome
___EIEE (Early infantile epileptic encephalopathy)
___EIMFS / MMPSI / MPSI (Early-onset epileptic encephalopathy with migrating focal seizures)
___SHE / ADNFLE (Autosomal dominant Nocturnal frontal lobe epilepsy Sleep/Hypermotor Epilepsy)
___Hypsarrhythmia
___Lennox-Gastaut Syndrome
___Major Aortopulmonary Collateral Arteries (MAPCAs)
___Microcephaly (small head size)
___Ohtahara Syndrome
___West Syndrome
___ 11a. Other Diagnosis (please specify)._____

12. Was the diagnosis of KCNT1 confirmed with Genetic Laboratory Analysis?

☐ Yes ☐ No ☐ I'm not sure

13. Do you have a copy of your child's genetic report?

☐ Yes ☐ No ☐ I'm not sure

14. What KCNT1 variant does your child have?

___c.841C>T (p.Leu281Phe)
___c.862G>A (p.Gly288Ser)
___c.1038C>C (p.Phe364Leu)
___c.1066C>T (p.Arg356Trp)
___c.1192C>T (p.Arg398Trp)
___c.1193G>A (p.Arg398Gln)
___c.1238G>A (p.Arg428Gln)
___c.1420 >T (p.Arg474Cys)
___c.1421G>A (p.Arg474His)
___c.1975C>T (p.Arg659Cys)
___c.2782C>T (p.Arg928Cys)
___c.2687T>G (p.Met896Lys)
___c.2800G>A (p.Ala934Thr)
___c.2849G>A (p.Arg950Gln)
___c.2849G>T (p.Arg950Leu)
___c.2882G>A (p.Arg961His)
___c.3689G>A (p.Arg1230His)

14a. Other _____

____Report not available

15. Please select how the KCNT1 variant/mutation is described on the genetic report.

___Not sure/No report available
___Benign
___Pathogenic (PATH)
___Likely Pathogenic (LPATH)
___Conflicting Interpretations
___Variant of Uncertain Significance (VOUS or VUS)
Other _____

16. What type of KCNT1 mutation, if any, was found in your child's genetic testing? (Select all that may apply)

- ☐ Not sure/No report available
- ☐ No mutations were found
- ☐ Missense
- ☐ Nonsense
- ☐ Insertion
- ☐ Deletion
- ☐ Duplication
- ☐ Frameshift
- ☐ Repeat expansion
- ☐ Other

17. What does the report specify about KCNT1 inheritance? (Select all that may apply)

___Not sure/No report available
___Autosomal dominant
___Autosomal recessive
___De Novo (random)
___Multifactorial
___Mitochondrial inheritance
___ Other (specify). _____17a.

18. What kind of genetic testing was done? (Select all that apply)?

- ☐ Single Gene Sequencing (sgs)
- ☐ Whole Exome Sequencing (wes)
- ☐ Whole Genome Sequencing (wgs)
- ☐ Commercially available gene panel
- ☐ Customized gene panel
- ☐ Not sure/None

19. Does the report include any additional gene mutations? (Check any/all that apply)?

- ☐ No report available
- ☐ None
- ☐ BRAT1
- ☐ CDKL5
- ☐ CHRNA4
- ☐ CHRNB2
- ☐ DEPDC5
- ☐ CRH
- ☐ CABP4
- ☐ SCN2A
- ☐ Xp11.3
- ☐ Other (specify)_____

20. Approximately how long ago was the most recent genetic test confirming KCNT1 completed?

___Less than 1 year ago
___1-2 years ago
___More than 3 years ago
___Not sure/No report available

21. What is the name of the genetic testing company on your report?

- ☐ Not sure/no report available
- ☐ Ambrygen
- ☐ Athen
- ☐ Baylor
- ☐ CleanPlex
- ☐ Concert Genetics
- ☐ Dante Labs
- ☐ NeviCore
- ☐ Fulgent
- ☐ Gene DX
- ☐ Helix
- ☐ Hospital system lab (e.g., private hospital lab at your hospital)
- ☐ Illumina
- ☐ Impact
- ☐ Invitae
- ☐ Knight
- ☐ Myriad
- ☐ Labcorp
- ☐ Sema4
- ☐ Other (Not listed here)

22. What were the first KCNT1 symptoms that your child experienced (or symptoms that you or a doctor/nurse noticed)?

- ☐ Seizures
- ☐ Infantile spasms
- ☐ Feeding difficulties
- ☐ Learning disorder
- ☐ Inconsolable crying
- ☐ Muscle twitches or tremors
- ☐ Reflux and/or vomiting
- ☐ Delayed Development
- ☐ Lack of Neck Control
- ☐ Motor Delay
- ☐ Sleep-related issues
- ☐ 22a. Other (Please specify)

_____

23. Select the approximate age of your child when signs or symptoms were first experienced and believed to be due to KCNT1.

- ☐ Within the first 30 days of life
- ☐ 1 month - 3 months
- ☐ 4 months - 6 months
- ☐ 7 months - 12 months
- ☐ 1 year - 3 years
- ☐ 4 years - 6 years
- ☐ 7 years - 10 years
- ☐ 11 years - 14 years
- ☐ 15 years - 18 years
- ☐ After age 18

24. Select the approximate age of your child at time of diagnosis of KCNT1

- ☐ Within the first 30 days of life
- ☐ 1 month - 3 months
- ☐ 4 months - 6 months
- ☐ 7 months - 12 months
- ☐ 1 year - 3 years
- ☐ 4 years - 6 years
- ☐ 7 years - 10 years
- ☐ 11 years - 14 years
- ☐ 15 years - 18 years
- ☐ After age 18

25. Please indicate which type of healthcare provider first diagnosed the participant with KCNT1.

☐ Geneticist ☐ Neurologist
☐ Pediatrician ☐ Other

26. Has your child ever experienced a seizure without a fever?

☐ Yes ☐ No ☐ I don't know

27. What is the approximate age today of the biological FATHER (in years)? _____

28. Does this child's biological father have a KCNT1 mutation?

☐ Father has no known KCNT1 mutation, after genetic testing
☐ Father has KCNT1 mutation
☐ Unknown if father has KCNT1 mutation

29. If the father has a KCNT1 variant/mutation, is it the same variant/mutation as the child?

____ Yes ____ No _____ Unsure

30. What is the approximate age today of the biological MOTHER (in years)? _____

31. Does this child's biological mother have a KCNT1 Mutation?

☐ Mother has no known KCNT1 mutation, after genetic testing
☐ Mother has KCNT1 mutation
☐ Unknown if mother has KCNT1 Mutation

32. If the mother has a KCNT1 variant/mutation, Is it the same variant as the child?

____ Yes ____ No _____ Unsure

33. Does your child have any other relatives or family members diagnosed with a KCNT1-related epilepsy or a confirmed KCNT1 mutation?

____ Yes ____ No _____ Unsure

34. Does your child have any family members or relatives with a history of any of the following? (Check ALL that apply)

- ☐ Sibling ever had a seizure
- ☐ Relative ever had a seizure
- ☐ Sibling ever have a diagnosis of intellectual impairment
- ☐ Relative ever have a diagnosis of intellectual impairment
- ☐ Sibling had/has behavioral or psychiatric issues
- ☐ Relative had/has behavioral or psychiatric issues
- ☐ Sibling was born with a motor or muscle disorder
- ☐ Relative was born with a motor or muscle disorder
- ☐ Sibling with a diagnosis of epilepsy
- ☐ Relative with a diagnosis of epilepsy
- ☐ Sibling with a diagnosis or suspicion of Autism Spectrum Disorder
- ☐ Relative with a diagnosis or suspicion of Autism Spectrum Disorder
- ☐ Sibling died of heart-related issue
- ☐ Relative died of heart-related issue
- ☐ Sibling died of another sudden, unexplained cause
- ☐ Relative died of another sudden, unexplained cause
- ☐ Sibling died of sudden infant death syndrome
- ☐ Relative died of sudden infant death syndrome
- ☐ I don't know

35. Does your child have any siblings or half-siblings?

___ Yes ___ No

36. Categories that Describe Your Affected Minor or Ward

- ☐ American Indian
- ☐ Alaska Native
- ☐ Asian
- ☐ Black, African American, or African
- ☐ Central or South American Indian
- ☐ Hispanic, Latino, or Spanish
- ☐ Middle Eastern or North African
- ☐ Native Hawaiian or other Pacific Islander
- ☐ White
- ☐ Other
- ☐ Prefer not to respond

37. Additional categories that Describe Your Affected Minor or Ward

- ☐ Afghan
- ☐ African American
- ☐ Algerian
- ☐ Asian Indian
- ☐ Barbadian
- ☐ Cambodian
- ☐ Caribbean
- ☐ Chamorro
- ☐ Chinese
- ☐ Chuukese
- ☐ Colombian
- ☐ Cuban
- ☐ Dominican
- ☐ Dutch
- ☐ Ecuadorian
- ☐ Egyptian
- ☐ English
- ☐ Ethiopian
- ☐ European
- ☐ Fijian
- ☐ Filipino
- ☐ French
- ☐ German
- ☐ Ghanaian
- ☐ Haitian
- ☐ HmongIranian
- ☐ Honduran
- ☐ Iranian
- ☐ Iraqi
- ☐ Irish
- ☐ Israeli
- ☐ Italian
- ☐ Jamaican
- ☐ Japanese
- ☐ Korean
- ☐ Lebanese
- ☐ Liberian
- ☐ Marshallese
- ☐ Mexican
- ☐ Mexican American
- ☐ Moroccan
- ☐ Native Hawaiian
- ☐ Nigerian
- ☐ Norwegian
- ☐ Pacific Islander
- ☐ Pakistani
- ☐ Palauan
- ☐ Polish
- ☐ Puerto Rican
- ☐ Russian
- ☐ Salvadoran
- ☐ Samoan
- ☐ Scottish
- ☐ Somali
- ☐ South African
- ☐ Spanish
- ☐ Swedish
- ☐ Syrian
- ☐ Tahitian
- ☐ Tongan
- ☐ Tunisian
- ☐ Vietnamese
- ☐ Other
- ☐ Prefer not to respond