



24 May 2023

Dear Parents of children with a *KCNQ2* disorder,

I am a paediatric neurologist who specialises in epilepsy and a professor at the University of Melbourne in Australia. I also work as a paediatric neurologist at the Royal Children's Hospital in Melbourne. Our 30 year research program aims to understand the genetic causes of epilepsy; we discovered the first gene for epilepsy in 1995. One of my major research interest is the severe epilepsies of infancy or childhood, known as the developmental and epileptic encephalopathies, and we study how patients' symptoms change through life.

We were part of the team that discovered that *KCNQ2* can cause severe epilepsy. As you know, *KCNQ2* disorders affect many aspects of health, including sleep. We wish to better understand the type of sleep problems in individuals with *KCNQ2* encephalopathy and their relationship with epilepsy.

We would like to invite families who have a child or adult family member with a *KCNQ2* disorder to participate. We would like you to complete the attached questionnaire about how your child or adult family member with a *KCNQ2* disorder is sleeping. It consists of 30 short questions and will take approximately **10 minutes** to complete. **We will also need you to sign consent forms for participation in the study please.** This will allow us to access clinical records and test results, such as EEG, MRI and the exact *KCNQ2* mutation results. All of the information we collect is kept strictly confidential.

We hope this research will assist us to understand the nature of sleep problems in *KCNQ2* disorders. Our work will help families and their doctors to recognise when sleep is a problem in children or adults with a *KCNQ2* mutation and help to initiate steps to assist with sleep.

If you are interested in finding out more about this study, please contact us through the email address below.

This study has been approved by the Austin Health Human Research Ethics Committee Reference number H2007/02961.

**Contact email** – epilepsy-austin@unimelb.edu.au

**If you would like to learn more about what we do, you can find more information at our website for the Epilepsy Research Centre** – <http://www.epilepsyresearch.org.au> and <https://www.genes4epilepsy.org>

Many thanks for working with us to improve the lives of people with *KCNQ2* disorders. We look forward to working with you soon.

With best wishes

Prof Ingrid Scheffer

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