DLG4-Related Synaptopathy (SHINE syndrome) – Understanding the Clinical and Genetic Features

You/your child are invited to participate in a study aiming to improve our understanding of the clinical and genetic features of *DLG4*-related synaptopathy, the disorder caused by genetic variants in the *DLG4* gene.

The clinical and genetic information will be collected through a survey, which is to be completed by a clinician. If you are interested in participating in this study, please contact your/your child's clinician who, in your opinion, best knows your/your child's medical history and ask them to contact us.

Data protection

Data will be treated confidentially and stored in a secure REDCap database housed and administered by Copenhagen University Hospital, Rigshospitalet, Denmark in compliance with the data protection law in Europe, General Data Protection Regulation (GDPR, https://gdpr.eu/what-is-gdpr/). The project is registered at the Capital Region of Denmark's record of processing activities (p-2023-15100) in accordance with GDPR article 30 and approved by the legal department of Rigshospitalet. The results will be anonymized and published in a peer-reviewed scientific journal upon completion.

About us

Our group is employed at the Department of Clinical Genetics, Copenhagen University Hospital, Rigshospitalet, which is a public Danish hospital. Our group has been working with *DLG4*-related synaptopathy since 2018 and has several publications including the clinical and genetic descriptions of 53 individuals (publication), epilepsy features of 35 individuals (publication), and a chapter in GeneReviews (chapter). In this study we aim to deepen our understanding of the disorder, which will benefit both families, clinicians, and researchers working on treatment possibilities.

You are welcome to contact us if you have any questions or concerns regarding the survey or handling of data. Moreover, you can at any time request to withdraw your data.

Kind regards

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