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THE BOGGS CENTER ON DEVELOPMENTAL DISABILITIES
New Jersey's University Center for Excellence in Developmental Disabilities Education, Research, and Service
New Jersey's Leadership Education in Neurodevelopmental and Related Disabilities Program

Disease Conceptual Model of KCNT1-Related Epilepsy: Caregiver Impacts

Jasmine Lafferty, NJLEND Genetic Counseling Fellow
Dr. Gary Heiman, Rutgers University Department of Genetics

Background

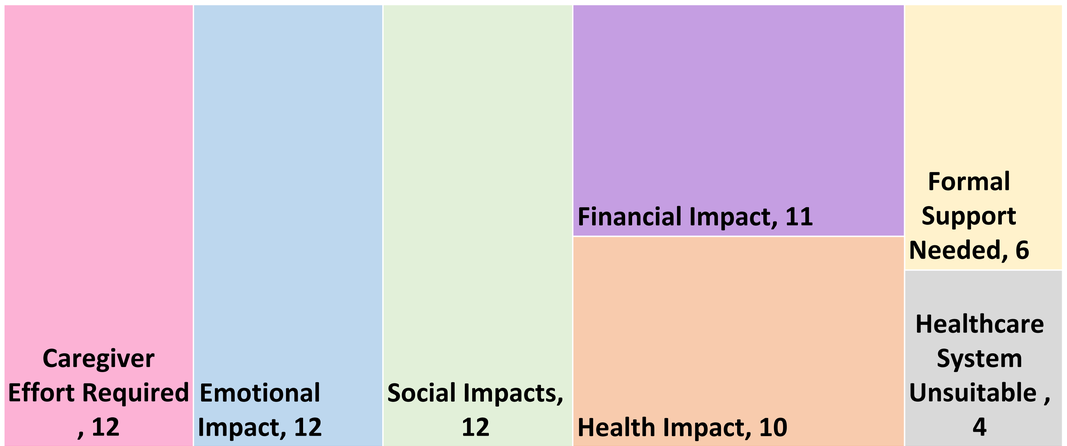
- KCNT1-related epilepsy is a neurodevelopmental condition caused by missense variants in the *KCNT1* gene.
- This condition has two distinct phenotypes, sleep-related hypermotor epilepsy (SHE) and epilepsy of infancy with migrating focal seizures (EIMFS)
- Seizures usually occur before age 6 months in EIMFS patients and after age 6 months in SHE patients
- EIMFS patients are usually nonverbal, nonambulatory, and have many other physical symptoms associated
- SHE patients typically have some verbal and/or ambulatory ability and have a high incidence of intellectual and developmental disabilities

Methods

- 12 caregivers of patients with KCNT1-related epilepsy were recruited through the KCNT1 Epilepsy Foundation
- Interviews were conducted via Zoom using a semi-formal nondirective framework
- .Interview transcripts were then coded through Dedoose using an adapted coding tree.

"…….there's a lot of grief and I think at some point it really kind of smacks you in the face and you just kind of zoom out and think is this really happening? Is this my life now? And is this why, you know, like is this real, why did it happen to us?" ~Caregiver 9

Domains of Caregiver and Family Burdens (n=12)



"Um and this was awful like I had no idea who our neighbors were because I never was there. Like I never even got to go to the park." ~Caregiver 8

Conclusions

- Every caregiver reported at least one emotional impact, social impact, and effort required suggesting these are the areas of the largest concern for this community
- Overall, it is important to understand the full effect this condition has on everyone involved, including the caregiver, which could influence available support systems and protocols

References

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