Clinical and Genetic Features in Patients With Reflex Bathing Epilepsy

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Study Question
What are the clinical and genetic features of patients with bathing epilepsy?

What Is Known and What This Paper Adds
Hot-water epilepsy and bathing epilepsy are among the most common forms of reflex epilepsy in children and are considered as part of a spectrum, but recent evidence suggests that they may be genetically different entities. However, this investigation’s results suggest that bathing epilepsy triggered by pouring water is a genetically distinct type of reflex epilepsy caused by SYN1 gene mutations.

Methods
For this cross-sectional study, the investigators recruited 12 individuals from 10 unrelated families (10 male patients; age range, 2.5–47 years) who presented with seizures primarily triggered by bathing or showering. Confirmation of seizures came in the form of either video-EEG recordings or home-video recordings by the patients’ parents. Recruitment occurred through the Network for Therapy in Rare Epilepsies. The investigators isolated genomic DNA from peripheral blood leukocytes, and they used epilepsy gene panels or whole-exome sequencing to identify genetic variants of interest in the patients from 4 families. Sanger sequencing was used for targeted analyses of the SYN1 gene in patients from all families, following guidelines from the American College of Medical Genetics for pathogenicity assessments. Participants in a previously reported cohort of patients with hot-water epilepsy were also screened for SYN1 mutations to determine if the identified mutations were specific to bathing epilepsy.

Results and Study Limitations
In all families affected by bathing epilepsy, the investigators identified SYN1 variants. This set of variants included 8 distinct pathogenic or likely pathogenic variants and 2 variants of unknown clinical relevance. Nine of these variants were novel. Conversely, none of the patients with hot-water epilepsy carried SYN1 mutations. The nonprimary seizure triggers included fingernail-clipping, hair-cutting, or watching someone else take a shower. The present study’s limitations include a small sample of patients with bathing epilepsy.

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A draft of the short-form article was written by M. Dalefield, a writer with Editage, a division of Cactus Communications. The corresponding author(s) of the full-length article and the journal editors edited and approved the final version.
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