

Fall 11-15-2021

Frequency of Each Sex Affected by Sudden Unexplained Deaths due to Developmental Epilepsy

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Recommended Citation

Garcia-Trujillo, Betsua; Hines, Dustin Ph.D.; Barker, Jeffrey; and Hines, Rochelle Ph.D., "Frequency of Each Sex Affected by Sudden Unexplained Deaths due to Developmental Epilepsy" (2021). *Undergraduate Research Symposium Posters*. 39.

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Frequency of Each Sex Affected by Sudden Unexplained Deaths due to Developmental Epilepsy

UNLV

by Betsua Garcia-Trujillo

Contributors: Dustin Hines, Ph.D. & Jeffrey Barker, Psychology Department

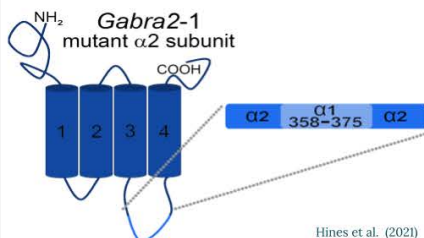
Mentor: Rochelle Hines, Ph.D., Psychology Department

Introduction

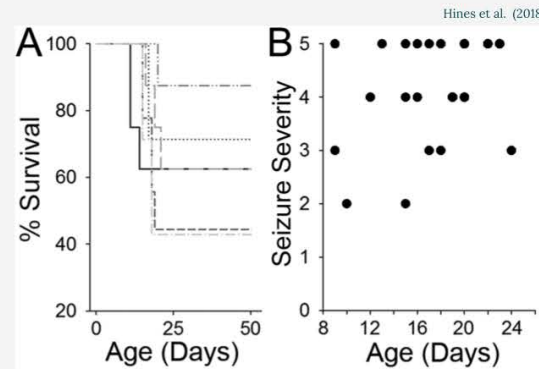
- Developmental epilepsy → group of neurodevelopmental disorders characterized by different types of seizures, developmental delays, and electroencephalogram (EEG) changes.
- Presence of symptoms and causes vary amongst groups and individuals.
- Previous research has examined sex differences of epilepsy within adult populations, but research is lacking for sex differences within younger populations with neurodevelopmental epilepsy.
- Objective of research is to determine the frequency of each sex within mice population that died prematurely due to GABA_A receptor $\alpha 2$ subunit mutations.

Methods

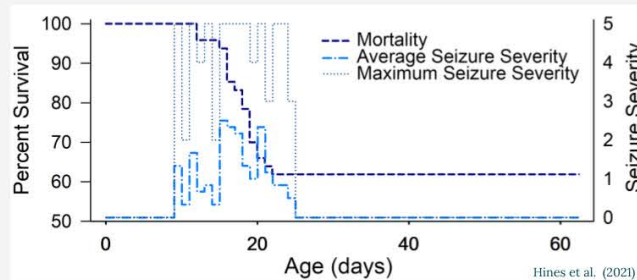
- DNA extracted from tails of mice that died prematurely due to developmental epilepsy.
- Sex determined by running DNA through PCR to amplify sex-determining region on the Y chromosome (SRY) and then separating PCR products by agarose gel electrophoresis.
- Males are identified by presence of two bands at 300 and 350 base pairs (bp) while females are identified by a single 350 bp band.



Schematic depicting the mutated GABA_A receptor $\alpha 2$ subunit. Thirteen amino acid sequences from the $\alpha 1$ subunit were used to substitute the collybitin binding domain within the $\alpha 2$ subunit. As a result, binding interactions between the $\alpha 2$ subunit and collybitin are reduced.

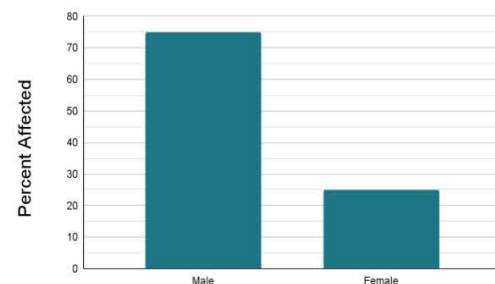


Graph A demonstrates survival rate of mice plotted against age in days, which also shows peak mortality at around day 20. Graph B plots seizure severity on a scale of one to five (Racine scoring) against age in days.



Graph demonstrating the relationship between observed spontaneous seizures and mortality, plotted against the age of mice in days.

Frequency of each sex affected by SUDEP

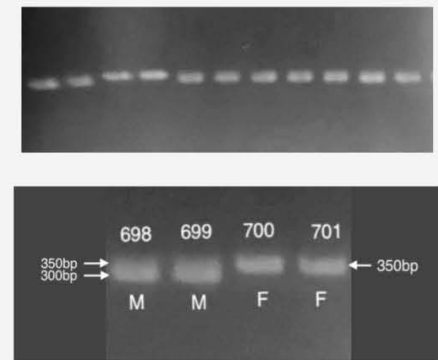


Graph that shows the percentage of each sex affected by SUDEP from samples examined so far. (Preliminary results)

Preliminary Results

- Other research has shown that males are at higher risk for sudden unexplained death in epilepsy (SUDEP).
- Current study aims to examine whether males are similarly at higher risk for SUDEP in our model of developmental epilepsy.
- Results may guide future research in providing additional insight into the mechanisms that lead to sex differences for SUDEP risk.

HINES Lab (2021)



Top image shows results of PCR products after agarose gel electrophoresis. Bottom image shows similar results after annotation that includes DNA sample numbers, base pair length, and observed sex.

Works Cited

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