



Timothy Syndrome Alliance

CACNA1C

CACNA1C Consensus is Live:

Preprint Now Available

Hi everyone!

We are really excited to let you know about the recent changes that have been announced regarding naming and classification for individuals with CACNA1C variants.

Working with a panel of global experts, we have laid out the key symptoms/disease presentations of patients with Timothy Syndrome and the **newly defined** umbrella syndrome: CACNA1C-Related Disorders (CRDs).

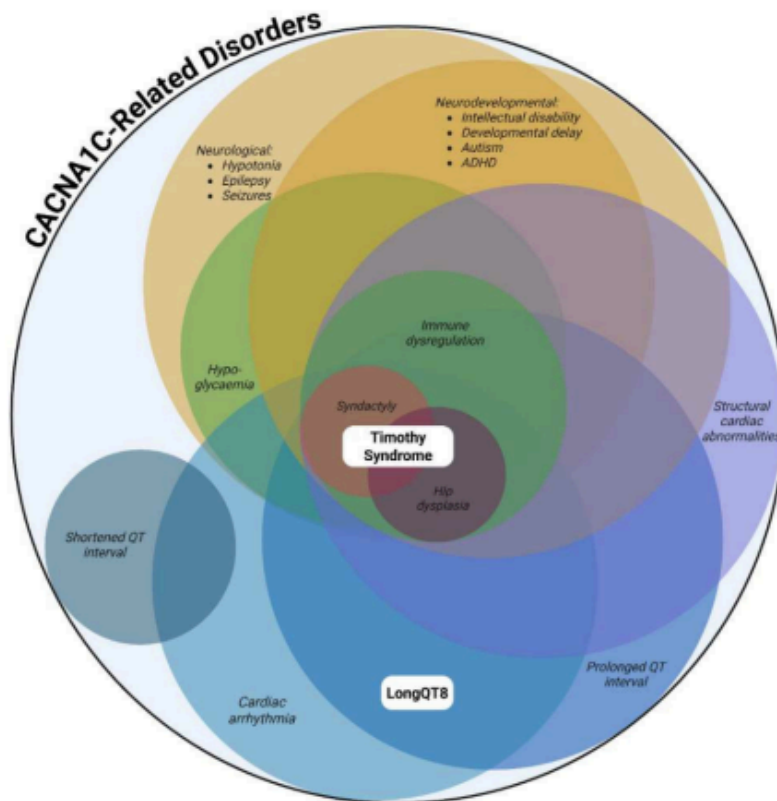


Figure from "Timothy Syndrome and CACNA1C-Related Disorder: First International Language and Management Guidelines Consensus Statement," Underwood JFG et al., Research Square (2024), licensed under CC BY 4.0.

Over the last 5 years, there has been rapid growth in the identification of individuals with rare CACNA1C variants. Alongside growing numbers, confusion surrounding the language of Timothy Syndrome and conditions caused by CACNA1C needed clarification.

With these new guidelines, we hope to simplify the process of diagnosis in patients with pathogenic CACNA1C variants, whether that be for Timothy Syndrome or CRDs. We have also provided guidelines for the minimum standards of expected clinical care for patients with these disorders.

Although this work still needs to be peer reviewed before it is officially published, we are making it available to read and share now:

**[Access the preprint of the
Consensus here](#)**

Feel free to contact me for more information!

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