

Pennsylvania Becomes One Of The First States to Proclaim Feb 24th as SCN2A Awareness Day

The FamilieSCN2A Foundation Announces
Pennsylvania State House of Representatives
Citation proclaiming February 24 as Pennsylvania's SCN2A Awareness Day.

HARRISBURG, PA, UNITED STATES, February 24, 2022 /EINPresswire.com/ -- The FamilieSCN2A

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We hope to utilize the resources in Pennsylvania to educate the medical community about genetic testing for children with autism and epilepsy. Many children, like my son, go undiagnosed."

Victoria Opthof-Cordaro, Lawrence's mom Foundation, a nonprofit organization established to improve the lives of those affected by SCN2A related disorders, today announced state proclamations and citations declaring February 24 as SCN2A Awareness Day.

The significance of the February 24th (2/24) date comes from the location of the SCN2A gene on the long (q) arm of chromosome "2" at position "24.3." SCN2A encodes voltage-gated sodium ion channel Nav1.2. Sodium ion channels play a key role in a cell's ability to generate and transmit electrical signals. Mutations or deletions of this gene are associated with autism, epilepsy, and other neurological issues such as movement disorders, cortical

visual impairment and dysautonomia.

"SCN2A related disorders affect patients in a wide spectrum ranging from severe, life threatening conditions to intellectual disability, and almost all patients will live a life completely dependent on others for their care and safety," said SCN2A Executive Director Leah Myers. "The recognition of February 24th as SCN2A Awareness Day will continue to help our efforts for early diagnosis, treatment and ultimately a cure for those suffering from this devastating disorder and to protect future lives."

The FamilieSCN2A Foundation and its mission was first brought to the attention of the State House by 4-year-old Lawrence Opthof-Cordaro and his family. Lawrence suffers from the SCN2A related disorders of autism, global developmental delay, feeding difficulties, and hypotonia. His family, in conjunction with the Foundation, sought the support of Pennsylvania's Representatives in bringing about awareness for the disorder in the pursuit of a cure. The citation is sponsored

by State Representatives Steve Samuelson (D-135th District), Robert Freeman (D – 136th District), and Joe Ciresi (D – 146th District).

Victoria Opthof-Cordaro, Lawrence's mom, said: "Our hope is that through advocacy we can utilize the resources available in Pennsylvania to educate the medical community about recognizing and obtaining genetic testing for undiagnosed children with SCN2A. Many Children like my son go undiagnosed, which is a significant barrier to available treatments and a future cure."

SCN2A related disorders have recently been identified as the leading single gene cause of autism and epilepsy. Do you know someone with autism or epilepsy? If so, encourage them to talk to their clinicians about genetic testing and visit our website for additional resources.



4 year old Lawrence suffers from SCN2A related autism

You can help us amplify SCN2A voices by wearing the Foundation's colors on February 24th: purple (epilepsy,) blue (autism) and/or green (movement disorders) and sharing information about SCN2A related disorders with others.

The FamilieSCN2A Foundation started in 2015 with fewer than 100 families and now represents more than 1000 families around the globe. The Foundation not only offers direct support to families affected by this devastating disorder, but is also the largest non-government funding source for SCN2A research, primarily from grassroots donors. For more information, please visit www.scn2a.org.

Leah Myers
FamilieSCN2A Foundation
leah.schust@scn2a.org
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