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Global SCN8A Community Unites for Awareness: Recognizing the Unseen Struggle on International SCN8A Awareness Day

On February 9, 2024, the global SCN8A community will come together to observe International SCN8A Awareness Day, an event dedicated to shedding light on SCN8A, a rare neurological disorder. This year's theme, "If You Only Knew," aims to reveal the hidden challenges faced by individuals and families affected by SCN8A and to foster a deeper understanding and support network for them.

SCN8A mutations are linked to severe epilepsy and developmental issues, impacting numerous lives worldwide. Despite its significant impact, SCN8A remains under-recognized. This awareness day seeks to change that by educating the public, healthcare professionals, and policymakers about the disorder.

The SCN8A Awareness Day is more than an event; it's a vital platform for change. "If only more people knew about SCN8A, early diagnosis and effective treatment would be more attainable, and families would feel less isolated," says Kacie Craig, Executive Director of The Cute Syndrome Foundation. "This day is about turning empathy into action and ensuring that individuals living with SCN8A receive the recognition and support they deserve."

February 9th will mark what would have been the 28th birthday for Shay Hammer whose father Michael, at the time a population geneticist, was the first to isolate the SCN8A gene as the cause of his daughter's struggle with epilepsy. Sadly, the discovery came weeks after Shay's sudden and unexpected death in 2011 at 15 years old – the first documented case of SUDEP (Sudden Unexplained Death in Epilepsy) in a child with an SCN8A mutation.

We invite rare disease organizations, media outlets, healthcare professionals, educators, and the public to join us in this important cause. Together, we can shine a light on SCN8A and make a tangible difference in the lives of those affected.

For more information about International SCN8A Awareness Day and how to get involved, please visit **scn8aawarenessday.net** or contact Kacie Craig.

ABOUT SCN8A

SCN8A is a gene that affects how brain cells function. Mutations on this gene cause a range of symptoms that can include severe epilepsy, developmental delay, and other medical challenges. Every person with this condition is affected differently and with varying severity. The role of SCN8A in the human brain was discovered in 2012. Since then, ~550 patients have been diagnosed with SCN8A epilepsy and related disorders. Due to advances in genetic testing, more patients are being discovered all the time.

Little is known about SCN8A including an absence of established treatments. Like many rare disorders, families and their efforts to advance research offer the greatest hope for these beautiful children who deserve answers.

ABOUT THE CUTE SYNDROME FOUNDATION

The Cute Syndrome Foundation raises awareness of SCN8A mutations, funds the dedicated and talented scientists researching SCN8A, and supports families around the world who are affected by this disorder. Our commitment to raising awareness and supporting research for SCN8A is driven by our belief in a future where every individual with SCN8A has access to the best possible care and support.

The Cute Syndrome Foundation is the starting point for support for new families to reach as they receive a diagnosis. We help clinicians standardize treatment for individuals with SCN8A. We work to expand scientific knowledge of rare genetic mutations associated with pediatric epilepsy and are the mainstay foundation to increase public knowledge of SCN8A mutations.

KEYWORDS

SCN8A, Childhood epilepsy, Intractable epilepsy, Genetic epilepsy, Epilepsy research, Precision medicine, Rare epilepsies, Genetic disorders, SUDEP, The Cute Syndrome Foundation, Wishes for Elliott: Advancing SCN8A Research, Dr. Michael Hammer, International SCN8A Epilepsy, EIEE-13, Special needs parenting, SCN8A family foundation, Rare disease registry

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