



What is SCN8A?

SCN8A is a gene that affects how brain cells function. Mutations on this gene can cause neurological problems including epilepsy and learning difficulties. ***The role of SCN8A in the human brain was discovered in 2012.*** Since then, ***~550 patients*** have been diagnosed with SCN8A mutations. Due to advances in genetic testing, more patients are being discovered all the time. It is now believed that SCN8A mutations may cause up to 1% of all epilepsies.

What is The Cute Syndrome?

The Cute Syndrome was a term used by the family of Esmé Savoie to describe the suite of symptoms that she had since birth that are now presumed to be caused by as many as four separate genetic mutations. However, the Cute Syndrome has evolved into something more than just Esmé's condition—it is now the name of the foundation created by Esmé's mom, Hillary Savoie, that represents an international community affected by SCN8A.

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