

So I've been treating patients with SCN8A-related epilepsy for as probably as long as I've been taking care of patients with epilepsy. We've actually only known about it fairly recently. It's it was a first found in association with epilepsy a little less than 10 years ago.

So we've been treating patients with this for quite a long time without actually knowing exactly what it is. SCN8A is a gene. And this gene can cause different kinds of neurological problems. Sometimes it just causes issues like developmental delay, intellectual disability. A lot of people who have a mutation, a change in this SCN8A gene though have epilepsy, and there's different degrees of epilepsy that people can have with SCN8A.

Some cases are mild. They have what are known as benign neonatal or infantile seizures. In other cases are much more severe with known as a developmental and epileptic encephalopathy, where children have severe intractable seizures that are very difficult to treat and have a whole host of other problems again, including intellectual disability, problems breathing, and a higher risk of death.

So there are no strikingly distinctive features to SCN8A related epilepsy. It is one of the few genetic epilepsies that can present very early on in the first month of life. Actually the average age of onset of seizures in SCN8A related epilepsy is four months old, so it affects kids that are very young oftentimes. So I do think that SCN8A is mistaken for other forms of epilepsy and not necessarily because they have given a child another genetic diagnosis, but perhaps just because they haven't done the test to look for what's the underlying cause. And children who have and otherwise unnamed focal epilepsy or other form of generalized epilepsy may actually have SCN8A and that's the more specific diagnosis.

I think giving this disorder a name and knowing why it's why their child has seizures or has these other issues can really be helpful.

It also prevents the need for other unnecessary testing, you know, you go down this long diagnostic odyssey trying to find out why your child has seizures or you know, whatever else you're dealing with. And you finally at the end of this you have an answer. That odyssey thankfully has been much shorter, thanks to earlier use of genetic testing for a lot of our kids. But it still is it's a challenging one to make for families.

The current treatments are effective for some but they're not effective for a lot who have the moderate or more severe forms of SCN8A related epilepsy, the kind we call developmental and epileptic encephalopathy. We have to modify our goals for therapy. In a lot of these patients because you know, we aim for no seizures, no side effects with these anti-seizure therapies. But for a lot of our patients with SCN8A related epilepsy, we can't achieve that so we try to reduce seizures. So they're not putting a child that at such a high risk. So they're not so frequent that they're impacting these child's daily lives. They're not so prolonged that their risk for going into these prolonged seizures we call status epilepticus.

So for current treatments under development for SCN8A, I'd like to see better seizure control and not just number of seizures, but also duration, severity of seizures, less hospitalizations related to seizures, less prolonged seizures and then improvements in a child's development and the family's life and lower risk of death and mortality.