NEWS

# Close look at rare syndrome bolsters autism-epilepsy link

BY **ALISON BRUZEK**

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The first systematic look at adults with the rare seizure disorder Dravet syndrome suggests that nearly two-thirds of them also have autism. The findings, published 23 May in *Epilepsy and Behavior*, add to mounting evidence that **epilepsy** and autism share biological roots[1].

Dravet syndrome is a **severe form of epilepsy** that appears shortly after birth. Children with the disorder have frequent, fever-related seizures that usually subside by age 5. The more frequent the seizures, the more likely a child is to have cognitive impairments. These impairments sometimes subside in adolescence or adulthood as seizures become less frequent and severe.

Many children with Dravet syndrome show signs of autism, such as mild social deficits and **repetitive behaviors**. In a 2011 study, researchers found that **roughly one in four children** with Dravet syndrome have autism. But it has been unclear whether autism symptoms — such as restricted interests and abnormal eye contact — change over time in these individuals as cognition sometimes does.

“[We were] trying to confirm our clinical impression that autism occurs frequently in [adults] with Dravet syndrome,” says study leader **Bert Aldenkamp**, clinical neuropsychologist at the **Epilepsy Center Kempenhaeghe** in Maastrict, the Netherlands.

Aldenkamp and his team assessed nine men and four women with Dravet syndrome, who ranged in age from 18 to 60 years, for autism symptoms, using a developmental scale that is standard in the Netherlands. All of the individuals were institutionalized at the Epilepsy Center Kempenhaeghe for severe epilepsy and intellectual disability.

Only two of the adults had previously been diagnosed with autism, but all of them had a history of autism symptoms, according to medical records, suggesting that they'd had the disorder since childhood. The evaluations by Aldenkamp's team revealed that 8 of the 13 study participants — 61 percent — meet the criteria for autism as adults.

The finding suggests that autism is common among adults with Dravet syndrome. By contrast, behavioral problems, such as hyperactivity, that are not related to autism but are associated with the syndrome seemed to fade in these adults.

"A lot of times, we concentrate on children because that's when they're diagnosed, but we need to think of how they end up as adults," says **Scott Perry**, medical director of the Epilepsy Monitoring Unit at Cook Children's Hospital in Fort Worth, Texas, who was not involved in the study. "It's an important thing that hasn't really been looked at [until now]."

The persistent overlap between Dravet syndrome and autism reinforces the notion **that a common molecular pathway is altered in both disorders.**

Dravet syndrome usually stems from a mutation in **SCN1A**, a gene that encodes a sodium channel necessary for nerve and muscle function. Mutations in this gene can suppress inhibitory neurons, which may result in too much neuronal activity and epileptic seizures.

Disruptions in the balance of **inhibitory and excitatory signals may also underlie autism**. In fact, rare mutations in SCN1A have been **linked to autism** as well. And a related gene, **SCN2A**, is rising to prominence as a **top autism candidate**.

The number of adults with Dravet syndrome who also have autism is high in this small sample compared with the proportion of children thought to have both conditions. Because the individuals in this study have symptoms severe enough to require institutional care, however, they may not be representative of affected people more generally.

## References:

**1. Aldenkamp A.P.** *et al. Epilepsy Behav.* **47**, 11-16 (2015) **PubMed**