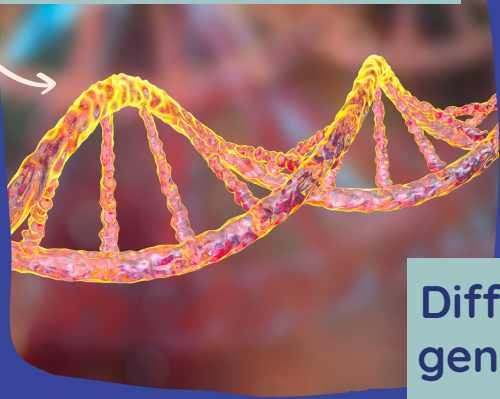


THE CNKSR2 GENE

A gene is an instruction set that is the basic unit of heredity



Differences in the CNKSR2 gene that result in disease lead to...



Intellectual Disability



Developmental Delay



Epilepsy (Seizures)



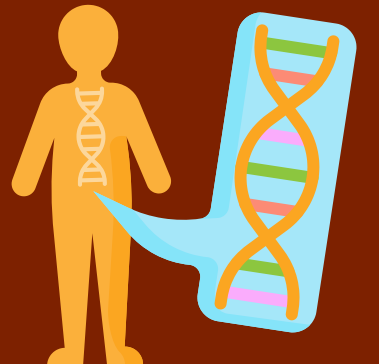
Sleep Disturbances



Feeding Issues



- Study motivated by an active and engaged family support group
- Focused on 13 males, ages 3-20 years, that had CNKSR2-related disorder
- All of the affected males shared symptoms of seizures and neurodevelopmental concerns



WHAT WAS OBSERVED?

46%

Had Seizures During Sleep

61%

Were Diagnosed with Autism

23%

Were Diagnosed with ADHD

- Degree of developmental delay can be severe and affect language skills
- Severity of seizures appears to decrease over time
 - Almost all cases resolved during pre-teen years
- 3 of the mothers were carriers of CNKSR2-related disorder, but none of them have had a history of seizures or neurodevelopmental concerns

62%

Had Sleep Disturbances

46%

Had Significant Hyperactivity

31%

Had Significant Feeding Issues

WHY IS THIS IMPORTANT?

Provide a better understanding of the clinical effects of differences in the CNKSR2 gene

Improve ability to care for and manage those affected by CNKSR2-related disorder

Guide medical research into treatments for CNKSR2-related disorder

