

# Understanding CHD2-related Epilepsy & Autism

## What is CHD2?

CHD2 is a gene located on the 15th chromosome at 15.q26.1

The CHD2 gene provides instructions for making a protein called Chromodomain DNA Helicase Protein 2. This protein is found in cells throughout the body and regulates gene activity through a process known as chromatin remodeling.

A mutation in the CHD2 gene can cause epilepsy and/or autism

CHD2 disorders were first identified in 2013

## Clinical Characteristics May Include:

- Epilepsy that is often refractory
- Global developmental delays
- Photosensitivity
- Autism Spectrum Disorder or autistic features
- ADHD, hyperactive behavior, or inattentive behavior
- Intellectual disability (mild to severe) or learning disabilities
- Behavioral problems including some with aggression
- Speech and motor delays
- Regression (loss of milestones)

seizure onset typically occurs between 6 months and 4 years of age

February 2nd is CHD2 Awareness Day

- The phenotype for CHD2 varies person to person. No two cases are exactly the same

CHD2 is a rare disorder; it likely affects only 1% of children with developmental & epileptic encephalopathies

Coalition to Cure CHD2 is a patient advocacy organization whose mission is to improve the lives of those affected by CHD2-related disorders by funding research necessary for uncovering a cure



[www.CURECHD2.org](http://www.CURECHD2.org)