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 difficult-to-control
- 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

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

February 2nd is CHD2 Awareness Day

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

Coalition to Cure CHD2 is a patient advocacy organization whose mission is to improve the lives of those affected by CHD2-related disorders by increasing education, building community, and accelerating research to uncover a cure.

Learn more about CHD2 by visiting: www.curechd2.org

CCC COALITION TO CURE CHD2