Cortical Dysplasia-Focal Epilepsy Syndrome

National Cancer Institute

Source

National Cancer Institute. Cortical Dysplasia-Focal Epilepsy Syndrome. NCI Thesaurus. Code C133743.

An autosomal recessive condition caused by mutation(s) in the CNTNAP2 gene, encoding contactin-associated protein-like 2. It is characterized by normal development until the onset of intractable focal seizures at age 1-9. After the onset of seizures, language regression, intellectual disability, hyperactivity, and impulsive behaviors begin to occur. The majority of children eventually fulfill the criteria for autism spectrum disorder.