

HADDS QUICK GUIDE

What is EBF3 HADDS?

Early **B-Cell Factor 3** (EBF3) **Hypotonia, Ataxia, and Delayed Development Syndrome** (HADDS) is a neurodevelopmental syndrome caused by a mutation in the gene EBF3, a master controller gene on Chromosome 10, which is important for neurologic function. The EBF3 gene controls how other genes are turned on and off in cells. A mutation of the EBF3 gene causes dysfunction in the cerebellum region of the brain, impacting development.

HADDS is often characterized by global developmental delay, moderate to severe intellectual disability, ataxia (balance issues) and hypotonia (low muscle tone) making it difficult for coordination and walking, speech delay, strabismus (crossed eyes), dysphagia (swallowing issues), chronic constipation, bladder dysfunction and high risk for urinary tract and kidney infections. Individuals with HADDS often are also at higher risk for having seizures.

EBF3 HADDS

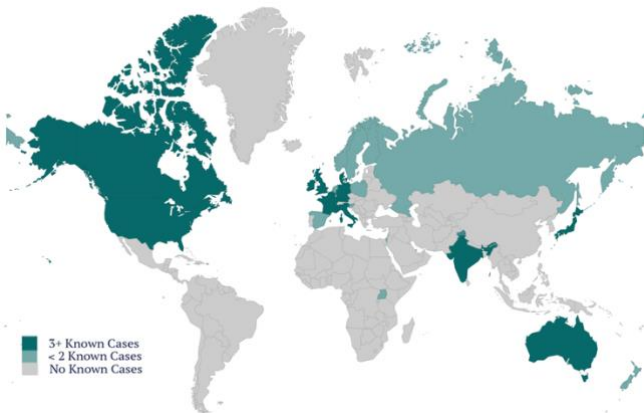
What are the symptoms?



The Growing HADDS Community

HADDS was discovered in 2016 at Texas Children's Hospital in Houston, TX, by Dr. Hsiao-Tuan Chao, Dr. Michael Francis Wangler and Dr. Hugo Bellen. As of 2020, there are approximately 150-200 people in over 13 countries worldwide with the HADDS diagnosis, but there may be upwards of a few thousand people undiagnosed globally. Many go undiagnosed because they are adults, unaware of the more recent genetic tests, such as the whole exome sequencing (WES) test or live in areas and/or countries where information and access to care isn't as readily available.

To support the growing community, the Foundation has facilitated international HADDS conferences and social events, raised awareness via social media campaigns, as well as provided travel grants to HADDS community members to ensure that they are able to participate in research and connect with the leading HADDS medical providers in Houston, TX.



HADDS Research – A Long-term Endeavor

Research is ongoing in Drs. Hsiao-Tuan Chao and Michael Wangler's HADDS clinic and lab, located at Texas Children's Hospital and the Jan and Dan Duncan Neurological Research Institute (Houston, TX, US). The goal is to study fruit fly and mice models to better understand the neurological nature of the syndrome. The long-term hope is the research can be applied towards developing gene therapies to better manage symptoms associated with HADDS syndrome.

Together, by raising awareness and funding, we can help enable those living with HADDS to thrive and receive the support they deserve!

LEARN, CONNECT and DONATE

www.HADDS.org