WHAT IS FOXP1 SYNDROME?

FOXP1 syndrome is an autosomal dominant neurodevelopmental disorder caused by defects in FOXP1, part of the Forkhead box (FOX) group of proteins. FOXP1 is widely expressed in the developing and adult brain, and is considered a key regulatory gene during neural development. Based on current data, lifespan is not limited for individuals with FOXP1 Syndrome.

WHAT CAUSES FOXP1 SYNDROME?

Chromosomes carry our genetic information in units called genes, made up of four "letters" (A, G, T, C). FOXP1 Syndrome occurs when there's a change or deletion in the FOXP1 gene. These changes can involve swapping a single letter, losing or adding a few letters, or even deleting part or all of one copy of the gene. These mutations usually happen by chance (not inherited) and having only one faulty copy of the gene while one copy is fully functioning can still cause the condition.

PREVALENCE

FOXP1 Syndrome is a rare disorder, with fewer than 1,000 individuals officially diagnosed. It is associated with autism spectrum disorder (ASD), contributing to the diagnosis of ASD in many affected individuals.

CHARACTERISTICS

FOXP1 Syndrome is characterized by a range of symptoms that vary in severity. Key characteristics include mild-to-severe intellectual disability and speech and language impairments that affect all individuals, regardless of cognitive level. Individuals often experience behavioral abnormalities, such as autism spectrum disorder (ASD) traits, ADHD, anxiety, and repetitive behaviors. Other common findings are dysmorphic features, oromotor dysfunction (contributing to speech and feeding difficulties), refractive errors, strabismus, cardiac abnormalities, renal abnormalities, cryptorchidism, hypertonia, hearing loss, and epilepsy.

DIAGNOSIS

FOXP1 syndrome is diagnosed in a proband with a heterozygous pathogenic variant in FOXP1, confirmed by molecular genetic testing and supported by clinical findings. A large majority of cases are diagnosed through whole exome sequencing, as FOXP1 is not typically included in standard neurodevelopmental panels. Most individuals with FOXP1 syndrome have the condition due to an autosomal dominant disorder caused by a de novo pathogenic variant, though in rare cases, a parent may have somatic or germline mosaicism, or a complex chromosomal rearrangement involving FOXP1. Each child of an affected individual with FOXP1 syndrome has a 50% chance of inheriting the variant.

CURRENT RESEARCH EFFORTS

The International FOXP1 Foundation is dedicated to advancing treatments for FOXP1 syndrome by funding innovative research. Key projects include the development of a three-dimensional organoid model to better understand how FOXP1 mutations affect brain development and function. The Foundation is also supporting efforts to restore FOXP1 haploinsufficiency through AAV-mediated gene rescue, a technique that uses viral vectors to deliver the missing or mutated gene to brain cells. These initiatives aim to uncover new treatment strategies and improve the lives of individuals with FOXP1 syndrome.

INTERNATIONAL FOXP1 FOUNDATION™

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foxp1foundation

The International FOXP1 Foundation is a 501(c)3 dedicated to supporting those impacted by FOXP1 syndrome and finding treatments and a cure.

MANAGING CARE

Supportive care is crucial to improving quality of life, maximizing function, and minimizing complications in individuals with FOXP1 syndrome. A comprehensive, multidisciplinary approach is often recommended, involving specialists in pediatrics, developmental medicine, neurology, physiatry, occupational and physical therapy (oftentimes including orthotics), speech-language pathology, psychiatry, psychology, otolaryngology, ophthalmology, cardiology, urology, and medical genetics.

In addition to traditional therapies, other interventions such as equine therapy, water therapy, and sensory integration therapy may be beneficial for improving motor skills, communication, and social engagement. These therapies can offer benefits, particularly in enhancing physical coordination, balance, and emotional well-being.

Individualized Education Programs (IEPs) may be important for children with FOXP1 Syndrome, as they provide specialized educational support to address cognitive, speech, and behavioral challenges.

WHAT IS THE INTERNATIONAL FOXP1 FOUNDATION (IFF)?

The IFF was created to build a global community that empowers and supports families and individuals with FOXP1 syndrome by sharing knowledge, inspiring hope, encouraging research, and raising awareness. The IFF encourages anyone affected by FOXP1 Syndrome to join IFF (https://www.foxp1.org/join-us). IFF provides a host of resources:

- Families have access to a wide range of support, including monthly Zoom chats, support from National Ambassadors, and access to social media support groups. In addition, fundraising resources are available to help families raise awareness and funds for research and care.
- A variety of resources are also provided on our website such as monthly newsletters, webinars, and easy-to-print research materials. These materials can be shared with medical providers and therapists to help them better understand and support the needs of individuals with FOXP1 Syndrome.
- Families can participate in the data collection program by sharing valuable information that will aid in future studies. To enroll, please visit https://www.foxp1.org/data-collection.

For a more detailed overview of FOXP1 Syndrome, consult: Rappold G, Siper P, Kostic A, et al. FOXP1 Syndrome. 2023 Sep 21. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews®

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