FOXP1 Syndrome
Care Guidelines

What can we expect for our child, and how can we best help them? We ask this question at every stage of development of our children as we search for the best treatments and therapies to enhance their quality of life. The GeneReviews® [Internet] article on FOXP1 Syndrome has published the following treatments for the manifestations. The first point they make is "there is no cure for FOXP1 syndrome." They then state, "Supportive care to improve quality of life, maximize function, and reduce complications is recommended. This can include multidisciplinary care by specialists in pediatrics, developmental medicine or neurodevelopment, neurology, physiatry, occupational and physical therapy, speech-language pathology, psychiatry, psychology, ophthalmology, and medical genetics, see Table 4 in Europe PMC FOXP1 Syndrome: Treatment of Manifestations." Ref GeneReviews® [Internet]

Developmental Delay / Intellectual Disability Management Issues. "The following information represents typical management recommendations for individuals with developmental delay / intellectual disability in the United States; standard recommendations may vary from country to country.

Ages 0-3 years. Referral to an early intervention program is recommended for access to occupational, physical, speech-language, and feeding therapy as well as infant mental health services, special educators, and sensory impairment specialists. In the US, early intervention is a federally funded program available in all states that provides in-home services to target individual therapy needs.

Ages 3-5 years. In the US, developmental preschool through the local public school district is recommended and results from referral to Child Find programs. Before placement, an evaluation is made to determine needed services and therapies and an individualized education plan (IEP) is developed for those who qualify based on established motor, speech, language, social, or cognitive delay(s). The early intervention program typically assists with this transition. Developmental preschool is center based; for children too medically unstable to attend, home-based services are provided.

All ages. Consultation with a developmental pediatrician is recommended to ensure the involvement of appropriate community, state, and educational agencies (US) and to support parents in maximizing quality of life. Some issues to consider:

- IEP services:
  - An IEP provides specially designed instruction and related services to children who qualify.
  - IEP services will be reviewed annually to determine whether any changes are needed.
  - Special education law requires that children participating in an IEP be in the least restrictive environment feasible at school and included in general education as much as possible, when and where appropriate.
  - Vision and hearing consultants should be a part of the child's IEP team to support access to academic material.
  - PT, OT, and speech services will be provided in the IEP to the extent that the need affects the child's access to academic material. Beyond that, private supportive therapies based on the affected individual's needs may be considered. Specific recommendations regarding type of therapy can be made by a developmental pediatrician.
  - As a child enters the teen years, a transition plan should be discussed and incorporated in the IEP. For those receiving IEP services, the public school district is required to provide services until age 21.
  - Vocational opportunities and programming including vocational rehabilitation should be considered early with a focus on achievement of meaningful employment
- A 504 plan (Section 504: a US federal statute that prohibits discrimination based on disability) can be considered for those who require accommodations or modifications such as front-of-class seating, assistive technology devices, classroom scribes, extra time between classes, modified assignments, and enlarged text.
- Developmental Disabilities Administration (DDA) enrollment is recommended. DDA is a US public agency that provides services and support to qualified individuals. Eligibility differs by state but is typically determined by diagnosis and/or associated cognitive/adaptive disabilities.
- Families with limited income and resources may also qualify for supplemental security income (SSI) for their child with a disability.

Fine motor dysfunction. Occupational therapy is recommended for difficulty with fine motor skills that affect adaptive function such as self-feeding, grooming, dressing, and writing.

Oral motor dysfunction. Feeding therapy (typically from a speech-language pathologist or occupational therapist) is recommended to help improve coordination of oral movement skills for feeding or sensory-related feeding issues using relevant approaches including postural modification and altering the consistency of food and fluid [Morgan et al 2012]. Lactating caregivers may need support from a breastfeeding or lactation consultant in the early weeks or months of life.

Gross motor dysfunction. Physical therapy may be recommended for difficulty with crawling, walking, running, and building strength resulting from hypotonia.

Speech and language disorder. Consider evaluation for nonverbal support or alternative means of communication (e.g., augmentative and alternative communication [AAC]) for individuals with severe speech and expressive language difficulties. An AAC evaluation can be completed by a speech-language pathologist who has expertise in the area. The evaluation will consider cognitive abilities and sensory impairments to determine the most appropriate form of communication. AAC devices can range from low-tech, such as picture exchange communication, to high-tech, such as voice-generating devices. Contrary to popular belief, AAC devices do not hinder verbal development of speech, but rather support optimal speech and language development.

In terms of verbal development, difficulties with motor planning (apraxia) and execution (dysarthria) is severe in the early years of life, and intensive evidence-based motor speech therapies should be applied [Morgan et al 2018]. Early phonologic awareness tasks should be implemented to support speech and later literacy development. Therapies addressing both receptive and expressive semantics and grammar are also recommended. The optimal intervention will be tailored to the child's specific profile as it changes during development." Ref GeneReviews® [Internet]

Social/Behavioral Concerns. Children may qualify for and benefit from interventions used in treatment of autism spectrum disorder, including applied behavior analysis (ABA). ABA therapy is targeted to the individual child's behavioral, social, and adaptive strengths and weaknesses and typically performed one on one with a board-certified behavior analyst. Consultation with a developmental pediatrician, neurologist, or psychiatrist may be helpful in guiding parents through appropriate behavior management strategies or providing prescription medications, such as medication used to treat attention-deficit/hyperactivity disorder, when necessary. Concerns about serious aggressive or destructive behavior can be addressed by a neurologist, developmental specialist, psychologist, or psychiatrist.

Surveillance. To monitor existing manifestations, the individual's response to supportive care, and the emergence of new manifestations, the evaluations summarized in Table 5 are recommended. [Table 5 is published in the Europe PMC.]" Ref GeneReviews® [Internet]

Parent Observations. Following are summaries of parent observations of their child's development. These summaries were written between 2017 and 2023, and are inline with the FOXP1 Syndrome article published in the Ref GeneReviews® [Internet].

FOXP1 Infants. Infants and children with FOXP1 syndrome should be followed by a clinician with expertise in pediatric neurodevelopmental disorders. These include but are not limited to developmental pediatricians, child and adolescent psychiatrists, and neurologists. A primary clinician can follow the child as they develop and recommend follow-up with other specialties as needed. Early intervention is important to help infants diagnosed with FOXP1 syndrome to maximize their potential. As many infants with FOXP1 syndrome have low muscle tone, physical therapy can help with meeting motor milestones like sitting, crawling and walking. Occupational therapy can also help infants learn to develop fine motor skills, which are often delayed, like learning to pick up pieces of food for feeding. Some children with FOXP1 syndrome will require an occupational therapist with expertise in feeding difficulties. Since FOXP1 syndrome is associated with language delays, children should begin speech therapy with a certified speech and language pathologist (SLP) by 12 months of age. If a child is not making sounds of pleasure (coos, laughs) and vowel sounds by 6 months of age, a consultation with a SLP is warranted.

FOXP1 Children. Children may benefit from a variety of therapies to target language, motor, and cognitive development. Speech therapy should focus on functional communication (e.g., ability to express basic wants/desires). As a child's functional language develops, speech therapy should focus on pragmatic language, or the social use of language. Pragmatic language interventions may focus on initiating, maintaining, and terminating back and forth exchanges. Occupational therapy should focus on fine motor skills, activities of daily living (e.g., feeding, dressing, toileting) and sensory reactivity. "Sensory diets" may be helpful for children who are sensory-seeking or sensory-averse to specific stimuli. Sensory diets can range from fine-motor manipulatives and activities for tactile stimulation to gross-motor activities such as jumping on a trampoline. Physical therapy may also be necessary when gross motor delays are present.

FOXP1 Teenagers. Several families of FOXP1 teens have reported worsening behavior issues after puberty, especially aggressive behaviors. An important area for future research is to understand how prevalent these issues are and what behavior management practices can help teenagers with FOXP1 syndrome and their families deal with these behavioral issues.

Families may benefit from working with a Board Certified Behavior Analyst (BCBA) to address challenging behavior. A Functional Behavioral Assessment (FBA) can be performed in the home or at school to develop appropriate treatment plans. Some adolescents may benefit from cognitive behavioral therapy (CBT) with a licensed psychologist to target internalizing symptoms such as anxiety. A child and adolescent psychiatrist with expertise in treating individuals with neurodevelopmental disorders will be critical in addressing clinically significant externalizing (i.e., hyperactivity, aggression) and internalizing (i.e., anxiety, depression) symptoms.

Academic curricula should continue to focus on functional academics and activities of daily living. Vocational training may begin during this time, as well as plans for transition to adulthood.

FOXP1 Adults. There are a fewer adult FOXP1s identified, as most individuals were identified with intellectual disability (ID) before the comprehensive genomic testing was available. The low number of identified adults does not imply an increased number in the population, it is just that many ID adults have not been tested.

There may be significant differences in individuals, dependent upon the severity of the genetic mutation. As FOXP1 adults mature, the extreme mood swings of puberty mellow, language and speech continue to improve, bladder control is often mastered, the dentist is no longer scary, and independence grows. FOXP1 adults tend to be regimented in their routines, enjoy collecting things, and often show autistic behaviors; however, autism is not always officially diagnosed. Communication is one of the biggest challenges for an individual with the FOXP1 diagnosis. As an adult it can be challenging for others to understand what they are saying. However, with today's technology it has opened a whole new world for our FOXP1 adults to communicate successfully. Although they may not take an active part in the conversation, their receptive language is excellent.

Advocating for your adult child becomes a priority, as the support and services they receive in school does not transfer to their adult life. In many countries there are limited services and multi-year wait lists. Families may benefit from working with a facilitator to develop their child's Person-Directed Plan. A Person-Directed Plan assists in identifying life goals and finding community connections, services and/or supports with the help of the family members and/or significant others of their choice.

A few years before individuals with FOXP1 graduate from the school system, parents should become familiar with local community service for adults with Intellectual and Developmental Disabilities (IDD). You may want to visit the services on site, ask questions, and determine which is best suited for your child. Services that support IDD adults may include:

- Day programs
- Independent-living, group homes, or family homes
- Paid work opportunities
- Volunteering within the community
- Transportation services
- Summer camps
- Recreation activities
- Respite services

"Based on current data, life span is not limited in FOXP1 syndrome [Palumbo et al 2013, Song et al 2015]. Progression of neurologic findings in adulthood has not been described. Since many adults with disabilities have not undergone advanced genetic testing, it is likely that adults with FOXP1 syndrome are under-recognized and underreported." Ref GeneReviews® [Internet].

The supportive care provided throughout their lives will help to improve their quality of life, maximize their function and contributions to their lives, and reduce possible complications in how they live.

This is what we strive for.