

Fragile X Syndrome

What is Fragile X Syndrome (FXS)?

Fragile X Syndrome (FXS) is a genetic condition that causes intellectual disability, behavioural and learning challenges and various physical characteristics. It is also the most common single gene cause of Autism Spectrum Disorder worldwide. It appears in people of all ethnic, racial and socio-economic backgrounds and can affect both genders (however, males are generally affected with greater severity). It is estimated that 5 per cent of people with a diagnosis of an Autism Spectrum Disorder (ASD) also have Fragile X Syndrome.

What are the common features of Fragile X Syndrome (FXS)?

Fragile X Syndrome can affect people in various ways and a person may be fully affected, but not show all of the following signs. Females are usually less affected than males.

Behavioural and Emotional Features:

- Anxiety and shyness
- Attention Deficit Hyperactivity Disorder (ADHD)
- Autistic type behaviours
- Difficulty with eye contact
- Tactile defensiveness
- Repetitive speech

- Aggression

Developmental Features:

- Intellectual disability
- Learning difficulties
- Developmental delay
- Speech and communication difficulties.
- Difficulty with coordination
- Difficulty with fine and gross motor skills.

Physical Features: (may be subtle or not present)

- Low muscle tone and loose joints.
- Long narrow face, prominent ears (in older males).
- High palate
- Large testicles (in post pubescent males).
- Mitral valve prolapse - heart murmur.

Carriers of the Fragile X gene may show the following features:

- Social interaction and communication difficulties.
- Learning difficulties
- Attention Deficit Hyperactivity Disorder (ADHD)
- Anxiety
- Depression
- Females may experience early menopause called Fragile X Primary Ovarian Insufficiency (FXPOI).
- Parkinson's-type disease symptoms in later life called Fragile X Tremor Ataxia Syndrome (FXTAS) in 20% of male carriers and fewer females.

Common difficulties often (but not always) experienced by the child with Fragile X Syndrome (FXS):

- Poor understanding of the conventions of social interaction.
- High levels of anxiety particularly in social settings.
- Resistant to change and very rigid in routine.
- Can be impulsive or aggressive.
- Poor non-verbal communication.

- Poor understanding of instructions, questions or jokes.
- Difficulty with fine and gross motor skills.
- Difficulties accessing the school curriculum.

Management strategies that support the child with Fragile X Syndrome (FXS) (at pre-school, school and/or home):

- Set up an individualised plan with parents/carers that have small achievable goals to help develop the child's skills.
- Develop social stories to improve appropriate behaviour in social situations.
- Positive encouragement
- Provide opportunities to succeed by simplifying activities.
- Teach new skills in a step by step manner and keep the environment as predictable as possible during teaching.
- Introduce new skills or environments on an individual basis before introducing peers.
- Use simple language and instructions.
- Provide visual as well as verbal cues.
- Provide extra time to complete tasks.
- Recognise and reinforce the child's strengths.
- Provide appropriate set up for the school desk.
- Set realistic and achievable goals for all task performance and completion.
- Make participation, not competition, the goal.
- Liaise between health professionals and educational staff to provide information to be incorporated for an education plan and/or implementing ideas/suggestions/activities to help improve the child's ability to access the curriculum.

Occupational Therapy approaches and activities that can support the child with Fragile x Syndrome and/or their carers include:

- **Expanding abilities:** Developing a broad range of skill areas.

- **Social stories:** Providing ideas and education around social story development.
- **School transition:** Advocating and professionally supporting the transition to school and liaising with teachers, as required.
- **Visual cues** can be used to support routine and to introduce new, or a change in tasks.
- **Gross and fine motor skills:** Determining the current age level of a child's gross and fine motor abilities.
- **Devise goals:** Setting functional and achievable goals in collaboration with the child, parents and teachers so that therapy has a common focus beneficial to everyone involved.
- **Educating** parents, carers, teachers and others involved in the child's care about Fragile X syndrome and the age appropriate skills a child should be demonstrating.
- **Management strategies:** Providing management strategies/ideas to assist the child in the home, at school, and in the community.
- **Teaching cognitive planning** and problem solving strategies.
- **Task engagement:** Providing alternative ways to encourage task engagement.
- **Direct skill teaching** through a task based approach.
- **Developing Underlying skills:** Developing the underlying skills necessary to support whole body (gross motor) and hand dexterity (fine motor) skills, such as providing activities to support:
 - balance and coordination
 - strength and endurance
 - attention and alertness
 - body awareness
 - movement planning

Speech Therapy approaches and activities that can support the child with Fragile X Syndrome and/or their carers include:

- **Speech and language assessment** to help the family to understand how the child is processing, understanding, learning and using language and

communication.

- **Communication strategies:** Providing the family with strategies and techniques to increase and enhance communication with the child.
- **Daily activities:** Helping the child to understand the environment, routines and language.
- **Developing language:** Helping the child to understand and use richer language and to use language more spontaneously.
- **Conversation skills:** Developing conversation skills (e.g. back and forth exchange, turn taking).
- **Concept skills:** Developing concept skills, especially abstract concepts, such as time (e.g. yesterday, before, after).
- **Visuals** can be used to help with understanding and the child's ability to express their needs, wants and thoughts.
- **Social skills:** Development of social skills (i.e. knowing when, how to use language in social situations).
- **Enhancing verbal and non-verbal communication** including natural gestures, speech, signs, pictures and written words.
- **Visual strategies:** Using visual information to help understand, organise and plan the routine for the day.
- **Liaising with educational staff** regarding the nature of the difficulties and ways to help the child to access the curriculum.

Why should I seek therapy for my child with Fragile X Syndrome?

Diagnosis alone is NOT the solution. It simply opens the door to getting the help that is needed by arming all involved with the relevant information.

The 'help' still needs to be provided. The help that is provided (at least from a therapy perspective) will reflect:

- First and foremost what medical intervention is needed.
- What the parents/teachers/carers biggest concerns are for the child (i.e. what are the most significant functional challenges).
- The specific areas that are problematic to the child (which will vary even within children with the same diagnosis).
- The capacity of the child's environments to meet the child's needs.

If left untreated, the child with Fragile X Syndrome (FXS) may have difficulties with:

- Following instructions within the home, kindergarten or school environment.
- Vocabulary whereby a child cannot clearly get their message across due to limited word knowledge.
- Understanding jokes and figurative language during interactions with others, and when watching TV shows and movies and reading books.
- Learning to talk, speech intelligibility and clarity.
- Managing a full school day due to poor strength and endurance.
- Participating in sporting activities leading to an inactive lifestyle, increasing the risks of other health related issues such as obesity, diabetes, cardiovascular disease or similar conditions.
- Self esteem and confidence when they realise their skills do not match their peers.
- Bullying when others become more aware of the child's difficulties.
- Fine motor skills (e.g. writing, drawing and cutting) due to poor core stability, meaning they do not have a strong base to support the use of their arms and hands.
- Completing self-care tasks (e.g. doing up shoelaces, buttons, zips, using cutlery).
- Self regulation and behaviour as the child is unable to regulate themselves appropriately to settle and attend to a task for extended periods of time.
- Accessing the curriculum because they are unable to attend to tasks long enough to complete assessment criteria.
- Sleep habits, impacting upon skill development due to fatigue.
- Social isolation because they are unable to cope in group situations or busy environments, impacting on their ability to form and maintain friendships.
- Anxiety and stress in a variety of situations leading to difficulty reaching their academic potential.
- Reading/understanding social situations and being perceived as 'rude' by others.
- Social communication, such as eye contact, appropriate distance when talking to someone, turn-taking within a conversation.

- Academic performance: Developing literacy skills such as reading and writing and coping in the academic environment.
- Academic assessment: Completing tests, exams and academic tasks in higher education.

More specific implications of not seeking treatment will be influenced by the common difficulties that are most influencing your individual child.

For more information see the relevant fact sheets under areas of concern or refer to the other relevant resources section below.

What does the diagnosis of Fragile X Syndrome (FXS) really mean for the child?

Diagnoses are used to label a specific set of symptoms that are being experienced by a child.

This label then helps to narrow down and specifically tailor what:

- Other issues commonly occur simultaneously.
- Medication might be appropriate.
- Therapies might help the child (e.g. Medical, Occupational Therapy, Speech Therapy, Psychology).
- Course of intervention (medical and/or allied health) might be and what outcome might be expected (prognosis).
- Can be done to help the child.

A diagnosis helps the child and their carers (parents, teachers, health professionals, carers) to:

- Access information about the relevant cluster of symptoms.
- Communicate the salient features of the child's challenges to all people involved in the child's care.
- Possibly interpret certain behaviours differently in light of the diagnosis.
- Obtain information about what can be done to help the child.
- Determine specifically where and how to help the child.

- Access funding or services that might not otherwise be accessible.

Contact us today to make an initial enquiry or book an assessment for your child
on 1800 KID SENSE (1800 543 736)

