



COMPLEX LEARNING DIFFICULTIES AND DISABILITIES RESEARCH PROJECT (CLDD)

FRAGILE X

Fragile X syndrome is the most common inherited form of learning disability. It is a chromosomal disorder caused by a 'fragile' site on the end of the X chromosome, which appears to be breaking but not quite separated. The gene which causes fragile X syndrome has been identified as the FMR1 gene. It is a gene present in everybody, but mutation, or an increase in the size of part of the gene can prevent it from working properly and thus causes learning disability.

Those affected with fragile X syndrome have a full mutation. Those with a small change or premutation of the FMR1 gene are carriers of fragile X syndrome, but are not necessarily affected by it. Premutation can make the gene 'unstable' and the size can change when passed between generations, but this can only be passed from a woman to her children.

Both men and women can be carriers of a premutation gene and the syndrome can occur in both sexes in all populations. However, despite the link to the X chromosome, it is more prevalent in males than females; this is because females have another X chromosome to compensate to a varying degree for the one damaged or 'fragile'. Current research estimates the number of males carrying the full mutation is 1 in 3800, and around 1 in 800 males carry the premutation (Beckett and Long, 2005). For females, the full mutation rate is currently at 1 in 8000, with the number carrying the premutation is at 1 in 250 (Turner et al, 1996).

Fragile X syndrome can cause mild to severe learning disability. It can also cause a range of additional difficulties, including social, speech and language, attentional, emotional and behavioural problems. However, it affects people in a variety of ways, and not all people with fragile X syndrome experience these difficulties. Some girls with fragile X syndrome may be affected by learning disability to a lesser extent, as their other X chromosome protects them some of the effects (Fragile X Society, 2007). A lot of typical profiles of fragile X syndrome are similar to those of autistic spectrum disorders (ASD) and, indeed, it is estimated that 25–35 percent of young children have a dual diagnosis of fragile X syndrome and ASD (Hagerman, 2004). For most, there will just be a cross-over in characteristics.

Physical features

Facial features associated with fragile X syndrome often become more pronounced with age and can affect some female carriers. These include a large head with long face, large jaw, prominent ears, nasal bridge long and flattened, and a high arched palate, often with dental overcrowding, as well as connective tissue problems such as flat feet, double jointedness, soft skin and a curved spine (Turk, 2008). Connective tissue looseness also predisposes students with fragile X syndrome to eye and vision problems, such as strabismus or 'wobbly eye', and ear infections such as 'glue ear' (Hagerman, 2004). Squints and poor focus on work may be common if tactile defensiveness (see below) prevents them from wearing glasses comfortably (Saunders, 2000). It is important these are monitored, as if undetected there could be permanent damage to sight and hearing, and therefore language development.

The onset of puberty (often premature) can bring enlarged testicles in males and menstrual problems in females; the potential physical and social discomfort of this should be considered, especially within the everyday pressures of school. Seizures also affect around 20 per cent of students with fragile X syndrome, but do not tend to persist into adulthood (Hagerman, 2004). Despite all these potential characteristics, students often do not present an unusual appearance and the indicators may not be

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present at all (Fragile X Society, 2007).

Motor skills

Floppy muscle tone, or hypotonia, can occur in people with fragile X syndrome due to poor connective tissue, particularly in babies and younger boys (Fragile X Society, 2007). This often leads to delay in reaching motor milestones, such as walking. Poor balance will usually persist into adulthood, as will difficulties with moving limbs in a controlled or precise way (Saunders, 2000). Fine motor skills are usually impaired with poor control of finger and hand muscles in boys. This can make handwriting, dressing and manipulating tools or objects, such as cutlery, very difficult. This combined with poor oral-motor co-ordination (Sudhalter and Belser, 2004) means students with fragile X syndrome can be messy eaters.

Cognitive development

Generally speaking, students with fragile X syndrome are characterised by a profile of cognitive strengths and difficulties. These strengths can be: good expressive and receptive vocabularies, as well as good verbal labelling; short and long-term memory for meaningful information, including good visual memory for the environment; and recognising and understanding emotional expression in others (Cornish, 2004). Difficulties that can be experienced, particularly by boys, are: a delay in language development (often persisting into adulthood); excessive repetition of a word or phrase; impulsive speech and poor pragmatic skills; poor short and long term memory for abstract, non-meaningful information (related to poor function of working memory); attention and concentration problems (discussed in more detail below); and arithmetic, in particular processing and recalling sequential and abstract information. It should be noted that studies into the cognitive development of very young children with fragile X syndrome report a very varied result of comprehension and word production, with some children understanding and producing many words, and others very few (see Scerif and Cornish, 2004), highlighting that not all students with fragile X will present in the same way. Clearly these strengths and difficulties have big implications for teaching and learning, but staff should remain mindful of the individual differences the student with fragile X may present.

Speech in students with fragile X syndrome may be hard to understand, particularly when speaking for a longer period of time. It is often delivered in short bursts followed by long pauses, with repetitions of sounds, words or phrases (Saunders, 2000). Whilst boys with fragile X syndrome might be good in a conversation, in terms of their knowledge and vocabulary their sequencing of ideas might get muddled, and they may struggle with appropriate turn-taking. Speech perseveration is the repetition of a single word or phrase. It is often bought on in states of anxiety, mostly from social situations, or from an inability to locate the next word or idea in a conversation.

Sensory integration

People with fragile X syndrome can have difficulties with integrating two or more sensory inputs at one time. Without being able to recognise, understand and filter the sensory inputs around them, they can become 'sensory defensive' – a mechanism akin to the 'fight or flight' process experienced by people when in a hostile situation. This defensiveness due to sensory overstimulation naturally impacts on behaviour (Wolstencroft, 2004).

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The sensory profile of an individual with fragile X syndrome is very similar to that of someone with ASD.

- Tactile defensiveness is very common in people with fragile X syndrome. Tactile defensiveness is when, to a varying extent, even harmless sensations are perceived as potentially dangerous and therefore unpleasant (Saunders, 2000). They are often sensitive to light touch to the point that it may be painful for them. However, it might be that deep pressure is sought after for its calming effect. They may also dislike exploratory art activities such as painting or use of clay, or carrying out daily care or hygiene tasks such as washing hands or shampooing hair. Some students find wearing gloves can reduce their sensitivity.
- Many people with fragile X syndrome may have difficulty in filtering auditory input and struggle to focus on tasks as they cannot shut out peripheral noises (eg the noise of traffic outside, or others talking, even if quietly). With too much going on at one time, the situation can get confusing, and as a consequence can cause anxiety and maybe challenging behaviour. This will have implications for engagement and learning if students cannot concentrate easily.
- Oral sensitivity can result in an individual with fragile X syndrome experiencing food with an unusual texture or taste, or particular cutlery when eating, as unpleasant. They may adopt certain behaviours at mealtimes to avoid these sensations.
- Students with fragile X syndrome are often hypersensitive to smells. This can provoke heightened positive or negative responses ranging from extreme preoccupation with them or aversion to them.

Students with fragile X syndrome might engage in certain behaviours to avoid being in a situation where they experience these sensory stimuli. For an example, in the classroom a student may particularly dislike the tactile sensation of holding a pencil, so might get anxious or display challenging behaviours to prevent having to hold the pencil (Saunders, 2000).

Vestibular and proprioceptive systems

Whilst they may not sound familiar, these systems play an important role for students with special educational needs, and an appreciation of their function is essential to understanding some of the postural and movement difficulties which students with fragile X syndrome and also those with ASD may have. Understanding these systems might explain behaviours such as jumping up and down, hand flapping and rocking, and perhaps other behaviours that appear to be without reason.

The vestibular system gives us our awareness of body position, movement position and movement in a space, balance, and stabilisation of the eyes during head movements (Saunders, 2000), the receptors for which are in the inner ear. An under functioning system will lead to poor coordination of movements and posture, an inability to maintain a calm yet alert state, and fidgety behaviour, perhaps walking around the room in order to give the brain the stimulation it requires to respond with improved posture and movement information. Conversely, an over functioning system will leave a person with too much information about posture and movement. This will prompt movement and posture insecurity, perhaps fear that particular movements may lead to falling or loss of control of their body. Students with an over functioning vestibular system may be reluctant to try new activities, preferring to stick to familiar activities requiring well-rehearsed movements.

The proprioceptive system provides us with an awareness of our body in space, giving us the ability to

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move intentionally and easily. This sensory information is generated by stretching and contracting of muscles, so occurs when we are moving. It enables us to be aware of our movements within a space and how our body parts are moving, without needing to look. Students with poor proprioceptive systems are slower and more clumsy and have to rely on visual and cognitive information to move appropriately.

Often people with fragile X syndrome will not only avoid stimuli, but also seek sensory experiences to calm or to heighten their levels of arousal. If this is the case, try: massage; use of weighted blankets or jackets; wearing backpacks; activities such as hiking, rolling or gardening; or games involving smelling. Consultation with an occupational therapist will provide a full sensory profile and recommendations specific for each student.

Behavioural development and social interaction

Males with fragile X syndrome are often friendly, helpful and curious, with a good sense of humour (Sudhalter and Belser, 2004), but may also demonstrate overactivity, inattentiveness, distractibility and poor inhibitory control. It is estimated that 70–90 per cent of boys and 30–50 per cent of girls with fragile X syndrome have attention deficit hyperactivity disorder (ADHD) (Hagerman, 2004). The unpredictability of everyday life for people with fragile X syndrome can cause anxiety, especially if struggling to communicate or experiencing unpredictability. Students may display hand-flapping or self-injurious behaviour, such as biting hands as a result of anxiety, frustration or excitement. This anxiety can often lead to challenging behaviours. Scerif (2004) reports that younger children seem to have more difficulty with inhibiting unwanted behaviours than older children.

Despite often enjoying others' company, social avoidance is a behaviour also commonly demonstrated by boys and girls with fragile X syndrome (Powell, 2004). This can include gaze aversion, displayed by over 90 per cent with the condition and even overt body turns away from a social situation or person (Sudhalter and Belser, 2004). It has been suggested that this behaviour is engaged in as a diversion tactic due to the awareness that social interaction will require language comprehension, auditory processing and motor planning, usually areas of weakness, despite often wanting to be involved in social interaction (Sudhalter and Belser, 2004).

Girls with fragile X syndrome are likely to show: particular difficulties related to organising their thoughts, planning ahead and shifting between topics; extreme shyness and anxiety in social situations; oversensitivity to perceived criticism and rejection; difficulty in picking up signals in social situations; and difficulty in seeing the consequences of their actions (Fragile X Society, 2003). This will naturally impact on their self-esteem and their ability to make friends, however much they want to.

People with fragile X syndrome also experience a pervasive impairment in their ability to regulate states of arousal, leading to hyperarousal. This hyperarousal is prone to manifest in anxious states, particularly, for example, in social interactions, with hyperarousal taking longer to subside than in people without the syndrome. Hyperarousal is frequently combined with states of anxiety, which can often lead to outbursts, tantrums and even aggression in boys; girls tend to be less hyperactive and have more mood stability than boys (Hagerman, 2004). Hyperarousal in conjunction with a reduced inhibitory control can lead to repetitive use of language, or favourite or associated topics being discussed regardless of appropriateness or conversational demands (Sudhalter and Belser, 2004).

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Implications / suggestions for learning

It appears that for students with fragile X syndrome, many of their characteristics will compound in the classroom making learning very difficult. For example, Cornish (2004) states that it is likely that the 'social demands of the language environment for children (eye-contact, co-ordination of syntax, semantics and conversational pragmatics) promote hyperarousal and anxiety, which in turn may result in an inability to inhibit verbal responses' (Cornish, 2004, p 22).

- When communicating abstract information, give small chunks at a time clearly, focusing upon the student's strength of verbal skills and good memory in a meaningful context.
- Breaking everything down into small steps may not always be best practice as students may need to see a more complete picture of what they are learning.
- When the student is learning to read, try a sight alphabet and whole words within a meaningful context, as phonics require abstract thinking and memory, sequential processing to put them together, and a dependency on sight and sound integration – areas of difficulty for some boys with fragile X syndrome.
- Much like strategies in place for students with ASD, visual aids in teaching and more practical-based approaches may make tasks more concrete and easier to engage with, rather than detailed spoken instruction.
- Tasks need to be made clear, attainable and given in a reasonable time span.
- Again, similarly to ASD practice, students will benefit from a focused, structured and predictable routine with change being kept to a minimum.
- IT based approaches might be beneficial as material can be delivered visually, with an instant response to action; this would also avoid direct teacher interaction, which some students may find hard to cope with.
- Give lots of praise; maybe have a reward system in place to boost self-esteem, and focus on the positives and strengths, rather than let the student focus on their difficulties.
- It is recommended that any direct instruction should be given whilst sitting or standing alongside the student, rather than directly in front of them, to avoid expectation of eye contact.
- Frequent states of anxiety and/or hyperarousal will require calming strategies, particularly for use when out in the community.
- When considering any point to action, it is important to always consider the student's individual needs, interests and preferences to make the intervention personalised to them. This could be achieved by consulting the student, family and all staff who know the student well.

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