

# Fragile x syndrome



Fragile X Syndrome, also known as Martin-Bell Syndrome or Escalante Syndrome, is a genetic syndrome which manifests in a wide range of physical, intellectual, emotional, cognitive and behavioral features that can be either mild or severe. The syndrome is characterized by the expansion of a single tri-nucleotide gene sequence (CGG) on the X chromosome and results in a failure to express a certain protein (FMR1) which controls the length of the repeated CGG sequence. This failure causes damage to the neural development of the body, including the brain, something which results in cognitive disabilities among others.

Many of the people who suffer from the syndrome have some cognitive weaknesses in their cognitive development, including the processes of thinking, problem solving, concept understanding, information processing and overall intelligence. Their overall potential tends to be lower than that of their peers and siblings, but they can nonetheless have patterns of strength too in their development and do very well in certain aspects of learning and cognition.

As many as 80% of males with the syndrome are described as "cognitively delayed". Older studies suggested that nearly all the boys or men with the syndrome have a moderate or severe mental retardation. Today there is more accuracy in the testing and in recognizing all those with the syndrome (not only those who were institutionalized, on which these previous studies were carried out), and only 10-15% of the males tested have the IQ that fits a borderline or mild mental retardation.

There are a number of boys and girls with the full mutation of the syndrome. They are described as learning disabled, as their overall intelligence is at least at the low average range but they have specific strengths and

weaknesses in their cognitive learning (" Cognitive Development Characteristics", 2009).

Cognition is also affected by Attention Deficit Hyperactivity Disorder (ADHD), seizure disorders, anxiety, disorders in speech and language, sensory motor problems and other factors (" Cognitive Development Characteristics", 2009).

In boys and men, there are several points of strength in cognition. These include verbal labeling, single word vocabulary and receptive (listening) vocabulary. Also, the vocabulary for subjects of interest may be higher than expected. Furthermore, visual matching and visual perception tasks with meaningful information (a familiar object, for example) are often performed well. Memory for situations and for favorite TV shows, song and videos is also excellent. Boys and men who have the syndrome are often great mimics, mimicking not only the words spoken but also the tone in which they were spoken. However, weaknesses are apparent in higher thinking and reasoning skills. Abstract tasks such as cause and effect questions or complex problem solving are very difficult if not impossible to perform by boys or men. Quantitative skills and arithmetic functions are also a daunting task for them.

In girls and women, strengths are found in comprehension and vocabulary items in the IQ test. Short term memory of meaningful visual material is also very good. As a result, reading, spelling and writing can be achieved well and correctly by these girls. Weaknesses however are seen in " executive functioning", the ability to plan, attend, sustain effort, using feedback, monitor ones self, generating problem solving skills and shifting abilities. These are abilities that help a person to think abstractly and reasonably ("

Cognitive Development Characteristics", 2009).

To sum up, the cognitive aspect in sufferers of the Fragile X Syndrome is affected in many fields of the human cognition, including ones which are vital for proper learning, understanding and making sense of ones environment. Be that as it may, sufferers also have strong compensatory abilities which help them function properly.

#### References

" Cognitive Development Characteristics" (2009). The National Fragile X Foundation Website. Retrieved October 15, 2009 from <http://fragilex.org/html/cognitive.htm>