

## **FEMALES AFFECTED BY THE FRAGILE X SYNDROME**

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The affectedness of females varies from case to case. To date there is no clear-cut method of predicting the level of affectedness based on fragility or historical pedigree or number of CGG repeats. There does, however, appear to be a cognitive profile, which exemplifies the affected female.

Research has determined that mental impairment with a retarded or borderline IQ occurs in approximately 35% of carriers (Sherman et al, 1985; Turner et al, 1980). Learning disabilities are often seen in carriers with normal IQ's (Kemper et al, 1986). These females present a cluster of cognitive features, which affect Arithmetic, Digit Span and Block Design performance on the Weschler Intelligence Scales.

Mazzocco studied the neuropsychological, physical and emotional functioning of females with fragile X syndrome ages 18-45 years who carry the gene and score low average or higher on cognitive (IQ) testings. She compared these women's results to several different controls, including 57 women who do not carry the gene but are related to either a fragile X individual or an individual who does not have fragile X but is developmentally disabled. The results indicated that the clinically expressing group (low-average or higher cognitive functioning), have more difficulty than the controls in:

- 1) Attention and/or visual-spatial abilities
- 2) Executive functioning (the ability to sum up intention, form a plan, and execute it)
- 3) More difficulty with novel or unfamiliar tasks

Hinton studied the difference in cognitive functioning among fragile X females who had inherited the gene from their mothers versus their fathers. She found that the females in the paternal inheritance group were no different in their cognitive skills than the control group. She noted that the group who inherited the gene from their mothers was split with a subgroup that scored at one SD from the norm, 85 IQ or lower. This was not evident in the paternal inherited group.

Hagerman has also noted a significant manifestation of shyness in both normal and impaired females with fragile X. The cognitive deficits are often masked and compensated for so as to preclude placement in Special Education programs. In addition, with the push for inclusion, the trend to remediate learning disabilities in a "pull-out" special education system is now considered restrictive and inappropriate. Therefore, many of these females go undetected by the school systems in the cognitive, as well as social arenas.

The female with fragile X will often exhibit a social learning disability, which results in interpersonal difficulties, shyness, inadequate communication, lack of eye contact and no reference to specific referents (he told me) when speaking. Additionally, clinicians have further noted a non-verbal learning disability (NLD) in many of the females with fragile X. Rourke (1988, 1987) has investigated the non-verbal learning disability extensively. Mizejeski and Hinton presented information at the 1992 International Conference which compared females with Fragile-X to those traits that defined NLD and noted the following:

- 1) Tactile-perceptual on left and right side; more on the left than on the right. Not evaluated.
- 2) Psychomotor coordination deficits; more on left side than right side. Without reference to laterality. Grigsby et al. (1990); Hagerman et al. (1985).

- 3) Visual-spatial-organizational deficits. Cianchetti et al. (1991); Grigsby et al. (1990); Hinton et al. (1992b); Mizejeski et al. (1984, 1986).
- 4) Deficits in nonverbal problem solving, including using information feedback in novel or complex settings. Mazzocco et al. (1992b).
- 5) Very well developed rote verbal skills, including extremely well developed rote verbal memory. Borghraef et al. (1990).
- 6) Extreme difficulty adapting to novel or complex situations, with over-reliance on mechanical, rote behaviors. Poor learning of novel task. Mazzocco et al. (1992b).
- 7) Relative to reading and spelling, very poor arithmetic. Mazzocco et al. (1992b); Mizejeski et al. (1984, 1986).
- 8) Reliance on language for social relating, learning and relief from anxiety. Not evaluated.
- 9) Interpersonal difficulties, which increase with age. Cronister et al. (1991); Lachiewicz (1992); Reiss et al. (1989).

The handouts give a graphic overview of the recent clinical findings, a triad of cognitive deficit areas, and what cognitive attributes contribute to the deficit areas. The suggested strategies have been developed over time in my practice with affected females. The suggestions can be applied at any level and can be modified to meet the particular academic levels of functioning.

In my clinical practice with females affected with fragile X, I have noted a number of difficulties with interpersonal relationships:

- 1) Many females have expressed an extreme fear of conflict and/or confrontation. When conflict is inevitable, they will choose to resolve it by becoming overly compliant, impulsive and hostile, or talking incessantly to avoid direct communication.
- 2) Emotional experiences are also difficult. Many times the experience involves a child also affected by fragile X. These conflicts create a two edged emotional sword. On the one hand, it is difficult to deal with their own feelings, but in addition, they are dealing with the behavioral difficulties or developmental delays for which they feel responsible.
- 3) As the interview process progresses, it is not uncommon to witness redness of the facial or chest area akin to blushing. This happens most often when embarrassment or emotional flooding occurs. Additionally, the language can become slow, with word-finding deficits.
- 4) Eye contact can be fleeting and diverted, especially during an intense discussion. Some females engage in laughing or giggling during these moments. Humor is also used to dilute the intensity of the emotional content.

Research indicates deficits in the area of executive function based on standardized measures. Females affected with fragile X frequently describe how this impacts their self-esteem and ability to accomplish goals. Often, I am struck by the creative ideas and interesting sense of vision, delayed by the ability to complete a project and truly bring the idea to fruition. This phenomenon, if repeated enough times, erodes initiative, incentive and personal self-worth. Many females affected with fragile X are excellent writers and create wonderful stories. One woman explained that she was much more able to express emotion through a fictional character than through her real life experiences. Analogies and abstract literary techniques are easily elicited to provide wonderful imagery.

Others nurture their creative tendencies through art forms, crafts and home decorating. Younger girls often enjoy paint by number, free form drawing and pattern drawing. It appears that the structure is welcomed, giving to them a sense of closure when completed. Interestingly, as young girls they are either described as extremely shy or outgoing and gregarious. As adults, they learn to compensate for the shyness in order to survive, but continue to struggle with social learning deficits throughout their lives.

### **Affected Heterozygous Females: Clinical Findings Thus Far**

- Retarded or borderline IQ in approximately 35% of carrier females (Sherman et al 1985, Turner et al 1980).
- Learning disabilities common in normal IQ carriers (Kemper et al, 1986).
- Reported weaknesses in Arithmetic, Digit Span and Block Design of Weschler Scales (Kemper et al 1986).
- Language deficits and dyslexia in 50% of 15 normal IQ carriers (Wolff et al 1988).
- Strengths in inductive reasoning and semantic memory function. Weaknesses in short-term memory, quantitative skills and visual motor tasks. Freund and Reiss (1989)
- There is a significant difference in cognitive ability between fragile X negative and fragile X positive carriers (Brainard et al, 1991).

### **Suggested Intervention Strategies for Math Deficits**

- Use concrete manipulative materials to teach concepts and mathematical operations.
- Use visual cues whenever possible to reinforce mathematical operations.
- Allow additional time to reduce the possibility of provoking performance anxiety.
- Minimize auditory distractions during time periods when concentration is required (computation, problem solving).
- Use diagrams, illustrations and visual patterns whenever teaching a new concept.
- Use repetition and patterning whenever rote memory tasks are required.

### **Suggested Intervention Strategies for Auditory Memory**

- Give specific instruction in a slow, simple and concrete manner.
- Place the student in close proximity of the instruction to insure attention and concentration.
- Structure the environment to be void of auditory distractions - example - earphones, carrels, or seating arrangements.
- Vary presentation to include frequent breaks to avoid attentional difficulties and lack of concentration.

### **Suggested Intervention Strategies for Visual Disorganization**

- Limit amount to be copied from printed or written materials.
- Simplify visually presented materials to eliminate a cluttered or excessively stimulating format.
- Provide visual cues - such as color coding, numbering, or using arrows to organize written tasks.
- Give specific concrete cues when giving oral directions that require an organized format.
- Additional time may be required for written assignments.

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