

Braden on Behavior



In The Beginning . . .

Marcia Braden, Ph.D.

Thinking back to our First National Fragile X Conference in 1987, I remember feeling excited and proud. We had finally pulled the experts together to provide a real conference. I was thrilled to have been invited and looked forward to meeting many of the fragile X experts that I had cited in my dissertation research. To think that I would meet Dr. Stephanie Sherman, Dr. Ted Brown and Dr. Optiz, just to name a few. It was such an incredible opportunity and I was truly humbled.

The conference was in Denver at The Clarion Hotel in December — not exactly a great time to travel to the Rocky Mountain state, but we were on a mission! It would be a two day conference — Thursday and Friday. My topic was entitled “*Techniques, Strategies and Identification of Learning Idiosyncrasies in the Fragile X/Autistic Patient*”—what a title! If I used that title today, I’d need several days to do it justice. But in 1987, I was just beginning to scratch the surface. Having seen less than fifty males with fragile X, I had begun to put a cognitive profile together, but with many unanswered questions.

The scientific presenters were exploring prenatal diagnosis, recombinant DNA technology and somatic cell hybrid studies which were new theories then concerning the etiology of fragile X syndrome. We were really at the infancy of our development.

My usual charge in this column is to write something about behavior, but nostalgia is taking over and I want to reflect on just how much more we now know and how far we’ve come. It is rewarding to recognize how much more we understand those affected by the elusive gene we have had a love-hate relationship with for twenty years.

In retrospect, perhaps the most important lesson I’ve learned is that this group of individuals is like none other. The people affected by fragile X syndrome have been relentless in their efforts

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to show me what they need. Their patience with me and my mistakes has been comforting. I have attempted to respond to their needs by applying a variety of educational strategies and behavioral principles to make them feel more productive and successful. The carriers have provided an added bonus because so often, they can verbalize those things that others with a full mutation have trouble articulating.

The educational outcome for those affected by fragile X syndrome has improved with the passage of I.D.E.A. (Special Education Law) and its subsequent amendments. This is a big change from where we were in 1987. We are now more able to provide a free and appropriate education to all children and we are compelled to include students with special needs in the least restrictive environment.

As I looked through my notes used to prepare for my lecture in 1987 (yes, there is a diagnosis in the fact that I still have those notes), I chuckled at some of my discoveries and thought the list would be useful to share:

- Great sense of humor— I love these guys!
- Easily excitable— I must be calm when interacting with them.
- Like to help others— love to have special jobs.
- Seem to get nervous or anxious when complimented (this was hard for me to explain).
- Phonics is a black hole; but I’ve seen them read— why is this?
- Their moods can change— delightful and suddenly anxious and resistant.
- Smarter than any IQ measure indicates.
- Able to imitate accents, intonation patterns, etc.
- Food likes are based on textures not taste.
- Problems with certain kinds of clothing, fabric, etc.
- Love music, cooking and sports.

Well, looking back on this list, I realized that a lot of the information gleaned through my early observations are still right on target, but we are now better able to meet those needs through technology and educational strategies.

Perhaps the most salient realization is that from the very beginning I saw these individuals as malleable and open to remediation when certain techniques were used. Their behavior could be modified, especially if we accounted for their fear of making a mistake, being embarrassed or frightened. The depth of understanding we now have in 2004, has enabled us to successfully craft behavioral

remedies that recognize and include antecedents which contribute to their behavioral difficulties. Instead of spending our time and energy on developing consequences that “teach a lesson,” we now focus on what causes the behavior to be present. Instead of waiting for a student to act out, we now intervene before the acting out is necessary. Instead of modifying a behavior using a consequence or punishment, we address the reason for the behavior and hope to eliminate the need for a consequence.

When I look back on the tremendous contributions so many gifted clinicians have made, I am in awe of all that has been accomplished. I find myself reflecting on one of Thomas Edison’s famous quotes and feel we have embraced it with a passion “If there’s a way to do it better...find it.”

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Genetic Counseling Then and Now

Liane Abrams

At the first fragile X Conference I was one of just a few genetic counselors attending. At the time the genetics of fragile X was murky. Only chromosome testing was available as the molecular basis (i.e. DNA mutations) had not yet been identified. Females could have chromosome testing but some known carriers had negative results and so if a girl tested negative it wasn’t definitive or always accurate. Also only a percentage of cells showed the “fragile X Chromosome” and labs used a different percentage to declare who was “positive.” It was confusing that some girls showed features of fragile X syndrome and some did not. It was highly unusual for girls to have X-linked conditions (like hemophilia or muscular dystrophy). The other complicated issue in genetic counseling was “transmitting males.” We knew some men must be “carriers” because related unaffected men had affected grandsons. This was unheard of with other X-linked conditions. If a man had the abnormal gene, he was affected. Genetic counseling sessions were full of “hopefully we’ll understand this more in a few years...”

Zoom ahead 20 years. The gene and its mutations have been identified. Now we now have a new language including DNA testing, pre-mutations, full mutations, methylation, mosaics, intermediate alleles and FMRP protein analysis. We could now explain “transmitting males (pre-mutation carriers), affected females (full mutations), premature ovarian failure, and why the disorder seemed to occur more frequently in each generation (mutations expanding). Genetic counseling sessions are longer, with fewer unknowns but more complicated information to explain and digest. Counseling issues have become more complex such as: “should I test my ‘normal’ daughter?” Or “what if an amniocentesis shows a full mutation in a girl, how affected will she be?” Many genetic counselors have now seen many families with Fragile X syndrome and are better equipped to answer questions and help wade through the decisions and issues that arise with this diagnosis. In fact, now at the Fragile X conference there will be many more familiar faces, those of genetic counselors who have learned alongside with the other professionals dedicated to serving families with Fragile X Syndrome. And now onto FXTAS and the next 20 years....

EDITOR’S NOTE: Liane Abrams is a genetic counselor who has been supportive of the NFXF and the Northern California Fragile X Resource Group for many years. She currently volunteers in the NFXF office one day per week in addition to her East Bay Genetic Counseling & Consultation business (www.ebgenetics.com).