

Note: This is one of the best undergraduate papers I've ever received.

Enjoy!

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Fragile X Syndrome

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Abstract

Fragile X Syndrome, the most common form of inherited mental retardation, is a disorder that is linked to a mutation on a gene of the X chromosome (Jewell, J., 2004). When the mutation of the gene becomes a complete one, it completely turns off the gene and does not produce the protein necessary to ensure that all of the other genes function properly (What Causes Fragile..., n.d.). Many characteristics are associated with Fragile X Syndrome. They include physical, cognitive, and behavioral. Some of the physical features include a long, thin head, protruding ears, low muscle tone, connective tissue problems, and many more (Fast D. & Abrams L., n.d.). These, however, are the least of the worries when dealing with these children. The cognitive and behavioral traits are by far the more important to focus on because these affect how the child will learn and function. Children with Fragile X are commonly oversensitive to stimuli such as noise and the things going on around them. When they become over-stimulated by these kinds of stimuli, they often will react inappropriately in order to get out of the situation they are in. They will often throw tantrums and be aggressive (Braden, M., n.d.). Although Fragile X is incurable, they can be helped through early intervention in some areas. These intervention methods include speech and language therapy, occupational therapy, behavior therapy, and medications (Fast D. & Abrams L., n.d.). Law mandates that everyone be able to receive an education. Students with Fragile X Syndrome can be placed in special education programs that allow them to be with both children with disabilities as well as “normal” children. Children with disabilities are also required to have what is called an Individual Education Program. These programs set up individual

goals and objectives for the student to work on throughout the school year (Harris-Schmidt, G. (a), n.d.). Many forms of special classes are available to these students as well. They include classes that teach the child about every day living such as personal hygiene, transportation, work skills, and shopping (personal interview, March 16, 2005). As they get older, students often can take classes that allow them to gain independence. These classes often take them out into the community where they receive on the job training. Fragile X Syndrome has no cure, but with the help of teachers, therapists, aides, parents, and volunteers, the effects of Fragile X Syndrome can be minimized.

Fragile X Syndrome

Fragile X syndrome is the most common inherited form of mental retardation (Jewell, J., 2004). The fully mutated gene affects about 1 in every 3600 males and 1 in every 4000 females. Fragile X syndrome, also known as Martin-Bell syndrome, was named after two men, Martin and Bell, who discovered, in 1943, that the condition is X-linked (Fast, D. & Abrams, L., n.d.). People with this syndrome have a large number of defects and disabilities including physical, cognitive, and neurobehavioral features (Jewell, J., 2004). These disabilities affect boys more often than girls and are caused by a gene that has been turned off in the X chromosome. There is no cure, however many options are available to people who have this disorder. Speech and language therapy, occupational therapy, behavioral therapy, and, probably the most well known, special education, are a few of the most common forms of treatment (Fast D. & Abrams L., n.d.). Cognitive and learning delays are associated with this disability. For this reason, education can be more challenging than normal. These delays vary for those affected and special accommodations must be made in order to meet the needs of each individual.

Fragile X syndrome is a mental retardation caused by the mutation of a gene (What Causes Fragile X, n.d.). This gene is called the FMR1 gene and is located on the X chromosome. Repeats of three bases make up the gene and are arranged in a cytosine, guanine, guanine (CGG) pattern. Normal variations of the FMR1 gene repeat the CGG pattern 7 to 60 times with the average being right around 30 repeats. People with Fragile X Syndrome, however, have an unusually large number of CGG repeats. A person with this syndrome has more than 200 repeats in their genes. When such high numbers are

found, methyl groups binds to the cytosine, or C bases, causing the gene to be turned off. Under normal circumstances, the FMR1 gene makes a protein called FMRP, or Fragile X Mental Retardation Protein, that is necessary to provide normal function (What Causes Fragile..., n.d.). This gene is present in everyone and is responsible for the development of the brain and testes by producing a protein that allows other genes to work normally (Saunders, S., 1999). The turning off of this gene does not allow it to make this protein causing impairments and other characteristics of Fragile X to develop (What Causes Fragile..., n.d.). This is how Fragile X gets its name. This mental retardation has many different kinds of physical, cognitive and behavioral characteristics.

Dr. Esther Nimchinsky is a researcher of Fragile X at the Cold Spring Harbor Laboratory located in New York. According to Dr. Nimchinsky, during normal development unnecessary connections in the brain are eliminated. This does not occur in persons with Fragile X Syndrome, leading to incorrect development of the brain (Nimchinsky, E., n.d.). This could be the main reason for the cognitive delays that people with Fragile X experience. Dr. Jennifer Jewell, a clinical assistant professor at the University of Vermont School of Medicine, suggests that children often have trouble meeting normal milestones such as speech and language. Attention Deficit Disorder (ADD) and Attention Deficit Hyperactivity Disorder (ADHD) are also very common traits of a child with Fragile X Syndrome. The average IQ is approximately 100. Those who score below 70 are considered mentally retarded. People with Fragile X tend to have an IQ between 35 –70 (Jewell, J., n.d.).

Along with these cognitive problems come some physical characteristics as well, most of which are only noticeable after the onset of puberty. According to Dr. Jewell,

children with Fragile X grow to be normal to slightly below normal height. Craniofacial abnormalities are also very common. These include long, thin faces with head circumferences much larger than the average. Other facial features include oversized, protruding ears as well as dental overcrowding. People with Fragile X often have connective tissue problems. Double-jointedness is one of these. Often they will be able to extend their fingers, thumbs, and extremities much farther than normal due to loose connective tissues (Jewell, J., 2004). Before puberty, inner ear infections are very common (Fast, D. & Abrams, L., n.d.). Heart murmurs are also common during adolescence and late childhood (Jewell, J., 2004). A heart murmur is when a valve in the heart does not close completely after contraction and blood leaks back through this valve (Heart Murmurs (a), n.d.). Although some heart murmurs can be very serious, most of them pose no need for concern (Heart Murmurs (b), n.d.). Another very common physical characteristic of Fragile X is flat feet. This is also a result of the loose connective tissues (Fast, D. & Abrams, L., n.d.). Macroorchidism, or oversized testicles, is a characteristic specific to males. Many times, men with Fragile X will have testicles that are two times and more the size of normal testicles. Back and chest problems are also frequently found. Pectus excavatum, where the chest caves into the body, and scoliosis are examples of this (Jewell, J., 2004).

Another set of characteristics that people with Fragile X possess is behavioral problems. People with Fragile X Syndrome are said to be sweet and loving with a strong desire for social interaction. However, about 90 percent of boys with Fragile X have some sort of sensory defensiveness (Fast, D. & Abrams, L., 2004). Sensory defensiveness is defined as a neural processing disorder in which an inappropriate

response occurs from an otherwise harmless stimulus (Adults With Sensory..., n.d.).

According to Dr. Dale Fast of St. Xavier University and Dr. Liane Abrams of the National Fragile X Foundation, this could include loud noises, bright lights, anxiety, and a feeling of being overwhelmed. These fairly harmless stimuli can evoke very inappropriate responses from them. For instance, a child may whine, cry, throw tantrums, misbehave, have aggressive behavior and do whatever it takes to get out of an overwhelming situation. Yet another very regular behavior problem with people with Fragile X, as mentioned earlier, is ADD and ADHD. Eighty to ninety percent of boys with this are considered impulsive and distractible. These children also show some autistic-like behaviors including hand flapping, skin biting, and avoidance of eye contact (Fast, D. & Abrams, L., 2004).

Dr. Jewell (2004) gives information on the genetics of the disorder. Fathers who have the pre-mutated gene, less than 200 repeats, pass the pre-mutation on to all of his daughters because he gives them an X chromosome. He does not, however, pass the pre-mutation on to his sons because they get the Y chromosome. If the daughter with the permutation has a child, depending on how close the gene is to full mutation, her child may have Fragile X Syndrome. The CGG pattern is unstable and increases in size during formation of the egg. Until now, mostly males have been discussed with the syndrome. The reason for this is simple. Females have two X chromosomes. Even if a woman has Fragile X Syndrome, she may not be as affected by it because she has an extra X chromosome which can make up for the abnormality of the other. Males on the other hand have only one X chromosome. If this chromosome is defective, there is nothing to make up for it. For this reason, males are affected by Fragile X syndrome more

frequently and more severely than females (Jewell, J., 2004). The risk for getting the disorder is directly related to the number of repeats of the CGG bases. A higher number of repeats means a higher risk for acquiring the disorder.

A number of intervention options are available to the person dealing with Fragile X Syndrome, one of which is speech and language interventions. According to Dr. Gail Harris-Schmidt of Xavier University in Chicago, both males and females may have problems with pragmatics, or conversational skills. Many times difficulty with language is one of the first indicators that something is not right with the child. Testing of a child should be done right away and should include hearing, oral-motor, and receptive and expressive vocabulary. Assessments should also include speech, voice, and fluency. A parent interview is also helpful in getting the individual assessed properly. A conversational analysis may also be performed to determine how well an individual can communicate with others. It is very important to do tests such as these in a very calm and quiet setting as well as with someone they are familiar with. This is to ensure that the child feels comfortable when he or she speaks. Testing for speech and language should be an ongoing process (Harris-Schmidt, G. (c), (n.d.)). Speech pathologists and special education teachers should set goals for communication that focus on social and learning interactions and incorporate them into the student's daily lesson (Casella, P. & McNamara, K., 2005). Along with seeing a speech and language therapist, medical intervention may also be needed to ensure that the child can communicate. Dr. Harris-Schmidt also informs that ear infections can be a common problem and can cause hearing loss. Insertion of tubes into the child's ears to drain fluid may be necessary in order for the child to be able to hear and listen properly. Another problem that could require

medical intervention is the issue of ADD and ADHD. Lack of attention is another part of speech and language that can interfere with communication (Harris-Schmidt, G. (c), n.d.). Medication could possibly be an effective way of addressing this situation. Some children with Fragile X Syndrome understand more than they can verbalize. Some may never speak at all. In this situation, another form of communication needs to be available to them. These alternative methods include gestures and sign language, picture cards, communication boards, and computerized interactive devices. No two children are the same. So, like in all situations with people with mental retardations, an Individualized Education Program or IEP is a must and is actually required by law (Harris-Schmidt, G. (a), n.d.). Speech-language pathologists are very important during the early years of people with Fragile X Syndrome, as well as the help of other professionals.

Behavior therapy for children with Fragile X Syndrome can be an immensely helpful to not only the parents, but to everyone who is a part of that child's life. Behavior therapy parallels speech and communication therapy in some ways. Dr. Marcia Braden, a psychologist and special education consultant from Colorado Springs, Colorado, writes that children with Fragile X often throw temper tantrums, which usually involve aggressive behaviors. Most people misunderstand these fits of aggression as unnecessary aggressive behavior, but this is usually not the case. Most of the time, these children will use this behavior to get out of an undesirable situation. Situations that may induce these sorts of behaviors include transition, loss of privilege, and a loud, noisy environment (Braden, M., n.d.). According to Dr. Vicki Sudhalter, a clinical psycholinguist, people need to be aware of the hyper-arousal and anxieties that children with Fragile X tend to experience. She believes that it must be addressed; whether it is through occupational

therapy, dance therapy, music therapy, aquatic therapy, or whatever other forms of therapy seem to alleviate that uncomfortable anxiety (Sudhalter, V., n.d.). Dr. Braden suggests that many times the best way to resolve inappropriate behavior is to remove the child from or altogether avoid that type of situation. Coping strategies are another beneficial and effective way of teaching the child to deal with the situation. This may even eliminate inappropriate behaviors. When dealing with a child who is having behavior issues, it is important to remember that overreacting to the problem will only worsen the situation and cause the negative behavior to escalate. Dealing with the situation calmly and indirectly are the best ways to alleviate the aggression (Braden, M., n.d.). Dr. Braden provides proactive behavior strategies that include several ways to structure the environment and lesson plans that take into account the fact that children with Fragile X Syndrome do not respond well to certain situations. Some of the strategies include providing structure and predictability, reducing the level of environmental noise, allowing additional processing time, providing calming activities, and encouraging down time and physical activity. It is important to these children that the environment be appropriate for them in order to prevent these aggressive behaviors. Another way in which a person with Fragile X Syndrome's behavior can be modified is by the use of medical intervention. Medicine can be a very effective tool in managing a child's behavior. It can stabilize their disposition and make behavior therapy more effective (Braden, M., n.d.). Along with these strategies is the idea of giving points or rewards when a student has done something good, such as following the rules. Using this method will not only encourage appropriate behavior, but also motivate the students to learn (Cruz, L. & Cullinan, D., 2001).

According to Dr. Harris Schmidt, an important part of determining what forms of intervention are needed is a test of cognition. There are many forms of tests, which are useful for telling a parent or teacher where a child needs help developmentally. The most common and recognized form of testing is the intelligence quotient, or IQ, test. Often, though, this test cannot be given to very young children or to children who are very cognitively challenged because a proper assessment cannot be made. In these circumstances, there are special tests or assessments that can be given. Many of these tests exist and include the Bayley Scales, the Stanford-Binet Intelligence Scale-4th Edition, the Wechsler Scales, and the Kaufman Assessment Battery for Children. All of these assessments are for children at different stages in their life. The one that has been shown to be very helpful for children with Fragile X Syndrome is the Kaufman Assessment Battery for Children. This test examines two types of processing, simultaneous processing and sequential processing. Most of the time, children with Fragile X have a difficult time with sequential processing. As the name suggests, sequential processing has to do with placing items in order such as numbers, alphabet, and tasks that require a certain order of events in order to be done correctly. Of account course, when performing assessment tests, other factors must be taken into. The fact that the child may have ADD, they become anxious very easily, and they may not have well developed verbal skills are just a few of these factors (Harris-Schmidt, G. (b), n.d.).

These characteristics of Fragile X Syndrome feed over into the area of education. Special education services are available for these children and the Individuals with Disabilities Education Act mandates that these children receive a free education in the least restricted environment (Harris-Schmidt, G., n.d.). Since each student is different, no

two lesson plans should be the same. Each student with any form of disability is required to have an Individualized Education Program (IEP) in which annual goals and objectives are set and are to be worked on throughout the school year. Law also requires that a meeting be held with all of those who will be a part of the student's education in order to determine the best possible way of reaching these goals and objectives (Huettig, C., Jan. 25, 2005).

Students with Fragile X Syndrome more often than not have difficulty learning and require assistance and a modified environment in order to assure that learning is as easy as possible for the child. For this reason, the quality of the classroom is a very important factor when teaching these children. According to research, the classroom quality and the level of engagement of children with Fragile X Syndrome are significantly correlated (Symons, F., Clark, R., Roberts, J., & Bailey, D., 2001). In other words, the more appropriate the teaching and the better the classroom practices, the more the students engaged in learning activities. This correlation shows the importance of an appropriate environment when teaching students with this disorder. There are many ways in which this can be done and should be included in the child's IEP. According to a study done by Suzanne Saunders, lecturer at Westminster College in Oxford, involving surveys and interviews from several teachers who have had children with Fragile X in their classrooms, there are many factors that go into an environmentally appropriate classroom. Teachers who responded to these surveys noted that it was important to provide activities that were physical in nature, involved a minimum amount of sitting and listening, and required very little direct communication with others. The atmosphere in the classroom should be quiet, calm, and free of distractions. The lesson plan should be

very structured and routine and allow the student their own area of the classroom that allows them to move around. This study also provided many different strategies that will help to facilitate learning in the classroom. One-on-one assistance, in many cases, can be effective in helping the child to stay on task and learn. It is very important, however, that this assistant or teacher be calm and quiet so that they themselves do not become a distraction. Teachers also pointed out that these students have a preference for visual learning. Any kind of pictures or symbols that can be used may be a very helpful tool in retrieving and storing information (Saunders, S., 1999).

The IEP changes as the student progresses from one activity to the next and they begin to master each task. The student's IEP also changes as they progress through life. A student who is three years old is not able to do activities that a nine year old may be able to do and the services and activities must change to be age appropriate. Dr. Harris-Schmidt has put together a list of services that students of different age groups might be involved in to be sure that they get the most out of their education. For infant to toddler aged children, services may be offered in the home where the professionals teach the parent or caregiver how to work with the child when they are not there. The Individuals with Disabilities Education Act of 1997 mandates that people with disabilities be able to learn in the most natural environment to ensure the best learning. This home environment is usually the most natural for children. Examples of professionals who may work in this environment are speech and language therapists and occupational therapists. Children in the early childhood age group might be best educated in a nursery school designed for "regular education", where the therapist might come once or twice a week. Other children of this age may be best off in a class that has a mixture of both children

with special needs and those without, while others may do better in a class of only children with special needs. Students with Fragile X Syndrome sometimes model after other students in the class, so often it is good for the student to be placed in a classroom with “normal” children. Placement, though, depends on where the child is most comfortable and where they function best. This environment is referred to as the least restrictive environment (LRE) and is mandated by law. Many students may have mild to moderate needs and can be in the general education classroom for most of the day with only a small amount of time set aside to work with a specialist, such as a speech or occupational therapist. Students who spend less than 50 percent of their day in a special education classroom are said to be in a “resource” room setting. These are the children who need only minimal amounts of special education. On the other hand, there are the students who are in special education classes all the time with the exception of maybe physical education, lunch, art, and music. These classes are called “self-contained special education” classes and include the students with more severe mental retardation. Every student in this class would have an IEP and several professionals, such as therapists, teachers, and aides, working to achieve the student’s goals. As the student gets older and progresses into high school and college, it is very important to teach the now young adult the skills necessary to be independent (Harris-Schmidt, G. (a), n.d.). Lori Randolph, the mother of a student with Fragile X Syndrome, discussed a few of the classes that her son took throughout his education to help prepare him for independent life. Throughout elementary school and junior high, he took a “Life Skills” class that was designed to teach the students about every day living, such as transportation, shopping, personal hygiene, work skills and manners. In high school he began a Community Based

Instruction program. In this program, students actually learn job skills as they are trained in the workplace. Students also learn in regular education classrooms where class content is modified by the instructor and an aide is present to help facilitate learning. The high school also provided modified physical education classes in which the students are partnered up with accelerated high school students. The high school students not only serve as aides but also surround the students with a good peer group (personal interview, March 16, 2005). There are a number of ways in which the school can help these students learn, as seen by the personal experiences of Mrs. Randolph and her son. These serve as just a few examples of how the school can provide services to students with disabilities.

Fragile X Syndrome is the most common form of inherited mental retardation (Jewell, J., 2004). It is caused when a mutation of the X chromosome occurs, turning off a gene necessary for normal function and development (What Causes Fragile..., n.d.). Many common characteristics occur in children with Fragile X Syndrome. They include ADD or ADHD, a long, thin face, protruding ears, low muscle tone, connective tissue problems and many more (Fast D. & Abrams L., n.d.). These are merely the physical characteristics. Many behavioral and cognitive characteristics are also common among children with Fragile X Syndrome. They will often have trouble with language and speaking (Harris-Schmidt, G. (c), n.d.). They are overly sensitive to certain stimuli and will often over-react when overwhelmed with these stimuli, such as too much noise or too much activity around them. These will often bring about responses that are not appropriate, such as tantrums and aggression (Braden, M., n.d.). There are, however, many intervention plans that can help these children to function normally and to be able

to learn with few difficulties, such as speech and language therapy, occupational therapy, behavioral therapy, and medication (Fast D. & Abrams L., n.d.). Law requires that each individual student have an Individual Education Program that contains annual goals and objectives for the student to work on throughout the school year (Harris-Schmidt, G. (a), n.d.). The schools often provide many various classes and services that allow the student to achieve the goals set for them. Schools also provide specialized classes that work on the skills the student will need in order to become more independent (Harris-Schmidt, G. (a), n.d.). Although there is no cure for Fragile X Syndrome, the things we do for the students now, whether it is in the classroom, at home, or anywhere else, can and will have a positive impact on their lives.

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