

2006

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Recommended Citation

Boyd, Susan V. (2006) "Rett Syndrome," *LC Journal of Special Education*: Vol. 1, Article 12.

Available at: <https://digitalshowcase.lynchburg.edu/lc-journal-of-special-education/vol1/iss1/12>

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Running Head: Rett Syndrome

Rett Syndrome

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Inclusion and co-teaching are becoming predominant in school systems across the country. Educators, both special education and general education teachers, are being introduced to children with disabilities that are caused by rare disorders. Requirements for these children to be placed in the least restrictive environment mandates that educators become knowledgeable of these disorders. One such disorder, which educators may not be familiar with, is Rett syndrome.

In 1966, Dr. Andreas Rett, an Austrian physician, published a report describing this disorder (Lee, 1999). According to a review of history by the International Rett Syndrome Association (IRSA, n.d.), Dr. Rett had become intrigued several years earlier when he had noticed two girls in his office with similar symptoms. After further inquiry, he discovered that there were six other girls in his practice that had the same characteristics. His assumption was that these girls shared the same disorder. He continued his research by filming the girls and traveling around Europe to see if he could locate others with the same symptoms. Around the same time, Dr. Bengt Hagberg, a Swedish physician, had discovered patients of his own who had these symptoms. He collected data and his plan was to study these data at a time in the future (IRSA, n.d.).

When the report from Dr. Rett was published it appeared mostly in German medical journals so the research about the disorder did not become well-known fact. In 1983, however, Dr. Hagberg published his own report and it was in a mainstream journal. It was at that time that Rett syndrome became a recognized disorder and the name it was given gave credit to the original physician to discover the disorder (IRSA, n.d.).

Rett syndrome, which affects 1 in 10,000-15,000 girls, is an X-linked progressive neurodevelopmental disorder. Affected girls usually develop normally until 6-18 months of age

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(Baker, 1999, p.7). The purpose of this paper is to explore the causes, characteristics, and interventions associated with Rett syndrome.

Causes

Although this disorder was identified as early as 1966, it was not until 1999 that the gene involved was discovered. A team, under the direction of Dr. Huda Zoghbi, discovered a mutation on the methyl CpG-binding protein 2 (MeCP2) gene found on the Xq28 chromosome (Baker, 1999). Locating the gene on the X chromosome gave the proof needed to show that the disorder was a X-linked disorder and also, because only one X chromosome needs the mutation for the disorder to occur, it is a dominant disorder. This scenario explains why the disorder is mostly diagnosed in girls. Irby (2000) explains that the MeCP2 mutation is usually fatal to boys. One exception to this rule is the diagnosis of Klinefelter syndrome. In this disorder the boys have an extra X chromosome making them XXY rather than XY. Rett Syndrome can occur if one of the X chromosomes has the MeCP2 mutation.

According to Baker (1999), the purpose of the MeCP2 gene is to silence transcription of other genes. A number of genes that affect normal development can be overexpressed when there is a loss of functioning in MeCP2. The International Rett Syndrome Association (n.d.) offers a good analogy to understanding the process. The organization states the syndrome [leaves]:

....those genes to act like overzealous electricians ignoring the wiring plans for a new house. Instead of installing a network of carefully placed wires

and switches, these neuronal electricians create a hodgepodge of wires that cause short-circuits and blown fuses (p. 4).

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This analogy gives a simplified answer to a very complex problem.

The International Rett Syndrome Association (n.d.) states that scientists have determined that MeCP2 is active throughout the body, but is abundant in the brain. When there is a mutation in MeCP2 and Rett syndrome occurs, specific areas of the brain are affected. These areas are the frontal, motor, temporal cortex, brainstem, basal forebrain, and basal ganglia. All of these areas are critical to controlling basic functions. They are also critical to higher brain center, the cortex, during development in late infancy.

Research continues in the area of Rett syndrome. Mouse models are proving to be beneficial in the research of the progression of the disorder. Scientists are hopeful that they can discover ways to cure or slow down this disorder. They are focusing on the prenatal and early infancy stage, prior to six months, as their Awindow of opportunity@ (IRSA, n.d.).

Characteristics

As mentioned earlier, females diagnosed with Rett syndrome appear to develop in typical fashion until around the age of six to eighteen months. There also appears to have been normal prenatal and perinatal development (Graziano, 2004). Hypotonia (i.e., loss of muscle tone) appears to be the first symptom. Apraxia, which is the inability to perform motor functions, is the most disabling symptom. The most stereotypical behavior of females diagnosed with Rett syndrome is that of hand-wringing or washing and repetitive hand-to-mouth movements. According to Irby (2000), other symptoms include toe walking, sleep problems, difficulty chewing, and breathing problems. Fear also seems to be associated with Rett syndrome. When

these girls are upset, they have a shakiness in their body.

Understanding the progression of Rett syndrome is made easier by reviewing the four

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stages that these females go through. These four stages give a guideline for the disorder.

Characteristics of the first stage begin at approximately 6 to 18 months of age. The National Institute of Neurological Disorders and Stroke (NINDS, n.d.) refers to this as the early onset stage. According to Van Acker (1991), it is sometimes hidden due to the speed of development in infancy. Some of the symptoms are decreased head growth, floppy muscle tone, and decreased interest in play and environment. Gross motor skills may also be affected and delays in sitting and crawling may be observed.

Stage two is referred to as the rapid destructive stage and occurs between the age of 12 and 36 months (NINDS, n.d.). This is a period of regression for the child. According to Harris, Glassberg, and Ricca (1996), language, social, and motor skills deteriorate quickly. The stereotypical behavior of hand wringing and washing will begin during this stage. Periods of sleep apnea and hyperventilation will also begin to occur. This stage is also when social aloofness begins and females will begin to demonstrate what are thought to be autistic traits. This is the stage when they are often misdiagnosed with having autism. Seizure activity begins at this time and it is seen in one-fourth of the females. Parents report that their children are more irritable and that tantrums are common during this regression period. This stage may last for weeks or months (Van Acker, 1991).

Between the ages of two and ten stage three begins, the plateau or pseudo-stationary stage (NINDS, n.d.). As the name implies, this stage can last for years and will be the stage that most females are in for the majority of their lives. The autistic features of Rett syndrome seem to fade

but motor functioning becomes more of a problem. Seizure activity is prominent during this stage. The females do appear to have more of an interest in their environment and attention span

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and alertness may improve.

Stage four, the motor deterioration stage, can last for years or decades (NINDS, n.d.). As one would assume by the title, this stage features muscle weakness, stiffness, spasticity, posturing difficulties, and scoliosis. Many times this stage requires the use of a wheelchair if there has not been one prior.

Rett syndrome is a very challenging disorder as can be seen by reviewing the various stages. There are many areas of concern. These stages not only give a general idea of what may occur during a given period in the child's life, but it makes one aware of what lies ahead. Being knowledgeable about the progression of the disease will allow time for preparation to meet the needs of the child when these various problems occur.

Interventions

Rett syndrome presents many complications for the children diagnosed with it, yet they very often live into adulthood. The severity of the disorder may vary among individuals, but most interventions can be utilized across the Rett syndrome population. Areas for which interventions can be important are self-injurious behavior, nutrition, seizures, communication, and various areas for physical and occupational therapy.

Self-injurious Behavior

Self-injurious behavior has been identified in children with Rett syndrome. This usually occurs due to the rapid hand-to-mouth movement that is a stereotypical characteristic. According to Van Acker (1991), there have also been reported cases of self-biting, self-pinching, and self-

hitting. According to Oliver (1999), all of these behaviors serve as sensory stimulation to these children.

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Various interventions have been implemented to deal with self-injurious behavior and the rapid hand-to-mouth movements. These interventions have included elbow orthosis (i.e., splint) to decrease the hand to mouth movement and interventions such as differential reinforcement, response interruption techniques, and mitts to reduce self-injurious behavior. Use of the elbow orthosis did increase toy contact in one study but the orthosis can also present a discomfort to the child (IRSA, n.d.). Katsiyannis, Ellenburg, Acton, and Torrey (2001) reported on a study which showed that the most successful intervention for self-injurious behavior was differential reinforcement paired with response interruption. The benefit to decreasing these behaviors is that the child becomes more alert and focused when hand movements and self-injurious behaviors are controlled.

Nutrition

Girls with Rett syndrome have difficulty maintaining adequate weight. One of the first symptoms noticed by Rett was a wasting affect and growth retardation (Lee, 1999). Therefore, nutrition is a very important area of intervention.

The loss of purposeful hand use makes it difficult to teach self-feeding techniques. Some adaptations to feeding utensils have proven effective, but maintaining weight can present difficulties. According to Lee (1999), Kathleen J. Motil, pediatrician and nutritionist, has had success with fitting patients with a gastrostomy button. This surgically implanted device allows nutrients to be administered while the child is sleeping. Use of the button has proven successful in increasing the weight and reversing the downward trend of poor growth. Also of importance

is consulting with a nutritionist to develop a weight management plan.

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Seizures

Seizures are very common in girls with Rett syndrome. This is not a surprising fact because the syndrome affects the nervous system, however, the cause of seizures in this disorder is unknown. According to Van Acker (1991), there are numerous EEG abnormalities for these girls between the ages of three and ten.

Information retrieved from The International Rett Syndrome Association (n.d.) states that determining whether a seizure is occurring in a girl with Rett syndrome may be difficult. What may appear to be a seizure could actually be uncontrollable movements that are characteristic of the disorder. If it is determined the child is having seizures there are medications, such as Tegretol, available to treat them. However, it has been found that many children are sensitive to the medications and may overreact (IRSA, n.d.).

Communication

Lack of communication abilities in a child diagnosed with Rett syndrome create a major obstacle in their education. According to Woodyatt and Ozanne (1992), communication skills are severely impaired and most girls are at a preintentional level of communication (i.e., caregiver will assign a meaning). An important aspect is to determine if the child is attempting to communicate using verbal or nonverbal behaviors.

Augmentative and Alternative Communication (AAC) can be very beneficial when dealing with children who have Rett syndrome. Some examples of AAC are eye gaze and voice output devices. Using these can lead to formal and effective communication (Van Acker, 1991).

According to Gaines (2005), eye gaze may be difficult for some girls, but spacing items far apart may help. By placing Velcro on a frame, pictures or objects can be attached

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and the child can choose based on gazing at the object. Eye gaze vests are also available. Pictures can be placed in pockets on a vest that can be worn by an adult. The advantage of the vest is that it is portable and can be used in different settings.

Gaines (2005) also reports that voice output devices range from low to high tech. The low tech systems use digitized speech and only use a limited amount of vocabulary. The high tech systems are much more advanced and can allow for more vocabulary, they are also more expensive. The low tech systems can be activated by a single switch. Van Acker (1991) points out that there have been successes by implementing single switches to activate toys and computers. The low tech devices are inexpensive, which make them accessible to the school systems. The advantage to the voice output devices is that the child's response can be heard by everyone. Shotko, Koppenhaver, and Erickson(2004) recommend that further studies be conducted to explore the possibilities of initiating communication, enhancing communication, and contextualizing it over multiple environments.

Physical Therapy

Physical therapists are a very important resource for children with Rett syndrome. Although they cannot alter the course of the disorder, they are successful in maintaining and improving function (Katsiyannis et al, 2001). One aspect they must keep in mind is that they can only treat the symptoms and they must individualize each intervention for the specific child they are working with (Van Acker, 1991).

According to The International Rett Syndrome Association (n.d.), physical therapy should

be one of the first sought after interventions. Physical therapy plays an important role in each stage of the syndrome. During stage one, they should focus on independent sitting, standing, and walking. Stage two requires the therapist to focus on range of motion and ambulation. Scoliosis may be more prominent in stage three and focus should be placed on seating, to assist with positioning, and transporting. Stage four requires that all aspects from the previous stages be incorporated (IRSA, n.d.). The role of the physical therapist is crucial to the well being of a child with Rett syndrome throughout their lives.

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Occupational Therapy

Occupational therapists also play a crucial role. Their main focus is on hand use and eating skills. Although feeding skills may be difficult to teach, they can assist the child with adaptive eating utensils and work toward a possible goal of independent feeding (IRSA, n.d.).

Occupational therapy can also work with the child on sensory processing issues. Sensory information is not well organized for girls with Rett syndrome. Integrative therapy, such as tactile activities and joint compression, can help the child better organize information and be better able to respond. The occupational therapist is also an excellent source to assist with accommodations and adaptive equipment in the classroom (IRSA, n.d.).

Discussion

Educators should be aware that, with the emphasis of least restrictive environment, public schools will be addressing the needs of children with Rett syndrome (Katsiyannis et al, 2001). It would be beneficial to educators to be aware of certain aspects when working with a child diagnosed with Rett syndrome. The International Rett Syndrome Association offers helpful hints for the educators who have children with the syndrome in their classroom (see Appendix A).

As with most children with disabilities, early intervention is vital (Harris et al, 1996). Personnel should be aware of behavioral interventions, focus on individualized approaches, and employ least intrusive interventions (Katsiyannis et al, 2001). Due to the fact that physical and occupation therapies are crucial for these children school personnel should also have the educational technology for reaching these children (Harris et al, 1996). Educators should be aware that effective collaboration will be very important for the success of a child with Rett syndrome in the classroom. The most important point for educators to remember is offered by Shotko et al (2004), A students with Rett Syndrome, as well as other disabilities, can and should be educated with the expectation that they can learn@ (p. 155).

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Appendix

Appendix A: Education Helpful Hints (IRSA, n.d.)

Education Helpful Hints

- Assume that she understands you.
- She may need a security symbol, such as a blanket or doll.
- Show consideration for her fears and hesitations.
- Limit outside stimuli to those that are necessary.
- Explain everything to her before you do it (especially if you are moving her).
- Make situations meaningful.
- Choose activities that appeal to her emotions and her senses.
- Make sure activities are age-appropriate.
- Structure activities in a fixed sequence.
- Give her one task at a time.
- Schedule her activities with tangible reminders (bag by the door).
- Choose signals that she understands (words, signs, pictures).
- Combine several signals so that she gets more than one cue.
- Don't say it, sing it.
- Allow her to move about her environment; she learns from taking it in.

Source: International Rett Syndrome Association.

