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Removed from the *DSM-5*: Considerations for Counselors Treating Individuals With Rett Syndrome

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Abstract

Rett syndrome is a relatively rare neurological disorder found primarily in females with implications in various areas of independent functioning. A diagnosis in its relative infancy, Rett syndrome results in a diverse array of symptoms including developmental deficiencies in communication, gross motor skills, social interactions, and intellectual abilities. Evidenced across various cultural groups, the rarity of occurrence makes Rett syndrome a relatively ignored topic in counseling literature. In light of the removal of Rett syndrome as a mental health diagnosis from the *DSM-5* and misconceptions regarding this decision, this paper investigates the uniqueness of Rett syndrome and provides suggestions for care. A case illustration is provided to illustrate some considerations for the counseling professional.

Parents' joy regarding the birth and healthy development of their children is likely to transition to concern when that development slows or even reverses. While these concerns may be the reaction of an overly anxious parent, in some cases, the significant changes in development are representative of something substantial. For parents of a child with Rett syndrome, fears become reality as the symptoms of the disorder become apparent. A rare and relatively newly discovered disorder, Rett syndrome first appeared in a journal article in 1966 but was not adopted until its second notation in 1983 (National Institute of Neurological Disorders and Stroke, 2014). Given this infancy, a dearth of research regarding Rett syndrome and its implication into counseling practice exists.

The acceptance into the *Diagnostic and Statistical Manual of Mental Disorders* (4th ed.; *DSM-IV*; American Psychiatric Association, [APA], 1994) provided the first, yet temporary, classification of Rett Syndrome as a mental health disorder. Included as one

of five distinct categories of autism related disorders, the diagnosis of Rett syndrome as a distinct entity was quickly questioned based on various concerns including the failure of such categorizations to accurately reflect the variability among the diagnosis. Despite persisting skepticism about the accurate and comprehensive portrayal of Rett syndrome as a disorder, its simple inclusion further shaped the direction of research.

In 1999, researchers grew closer to identifying the cause for Rett syndrome, a neurological disorder, found in approximately 1 out of every 10,000 to 15,000 live female births, by noting the presence of a mutation in the methyl CpG binding protein 2 (MECP2) gene (Leonard, Fyfe, Leonard, & Msall, 2001). While the presence of a MECP2 mutation appears to be the most common mutation associated with Rett syndrome, presence of a mutation in itself does not necessarily indicate that an individual has Rett syndrome. In fact, in cases where an individual is shown to have mutations on the MECP2 gene but does not show clinical symptoms of Rett syndrome, the individual is identified as being an asymptomatic carrier (International Rett Syndrome Foundation, 2014). Along the same lines, individuals noted as having atypical or congenital Rett syndrome are noted to not have mutations on the MECP2 gene but instead within other genes on the X chromosome.

With the growing understanding associated with the variability of symptoms, it may have come as no shock that 20 years after its inclusion with the *DSM-IV*, the controversy would again emerge with the removal of Rett syndrome as a unique diagnosis from the *Diagnostic and Statistical Manual of Mental Disorders* (5th ed.; *DSM-5*; APA, 2013). While the decision to remove Rett syndrome as a unique diagnosis within the *DSM-5* was the result of much research and consideration, however, its exclusion should not negate the importance of the disorder on those directly affected by it. As such, the following article serves as an opportunity to revisit what researchers know about Rett syndrome, to identify the potential implications of symptoms in counseling, and to develop considerations for mental health professionals working with individuals directly impacted by the disorder.

Medical Aspects of Rett Syndrome

Variations regarding mutations within the designated genes on the X chromosome are responsible for the diversity of symptom type and severity among individuals with Rett syndrome. According to the National Institute of Neurological Disorders and Stroke (2014), these mutations result in decreased functioning within the affected genes setting off a domino effect for the abnormal expression of various other genes, which results in the multiplicity of symptoms across individuals with Rett syndrome. Such variations in symptoms further complicate the diagnostic process.

Although researchers have identified a genetic cause of Rett syndrome, such causes are not linked to inheritance among populations (National Institute of Child Health and Human Development, 2014). Instead, researchers have indicated that in more than 99% of cases of an individual diagnosed with Rett syndrome, the mutations have occurred spontaneously and at random (International Rett Syndrome Foundation, 2014). This means that only a small portion of individuals with Rett syndrome had a relative who was also diagnosed with the disorder. Further complicating the research, a number of individuals with Rett syndrome were found to have a relative with mutations on their

MECP2 gene without demonstrating any clinical symptoms of the disorder, earning these individuals the title of asymptomatic carriers (Dolce, Ben-Zeev, Naidu, & Kossoff, 2013).

Because the presence of a mutation on the MECP2 gene does not necessarily mean a diagnosis of Rett syndrome, and the lack of a mutation does not rule out Rett syndrome completely, many children do not receive a diagnosis until symptoms become increasingly apparent. Appearance of these symptoms is more frequent and often occurs earlier in children where a greater number of their active X chromosomes carry the genetic mutation (International Rett Syndrome Foundation, 2014). Still, early signs of the disorder often go unnoticed.

According to the National Institute of Neurological Disorders and Stroke (2014), researchers have identified a series of four stages that an individual with Rett syndrome can anticipate going through. The first stage, referred to as early onset, is characterized by subtle symptoms that often occur between the ages of 6 and 18 months. Demonstrated by slight reductions in development (e.g., decreased eye contact, reduced interest in age appropriate toys, and gross motor skills), the majority of individuals are not diagnosed with Rett syndrome at this age as the changes do not appear significant enough to warrant attention.

As a child with Rett syndrome enters into the second stage, the rapid destructive stage, symptoms become more evident and gain the attention of parents and caretakers. Usually occurring between the ages of 1 and 4, this stage often involves the appearance of autistic-like symptoms (e.g., reduced or eliminated social interaction, poor or absent communication skills, etc.) coupled with difficulties in motor movement, loss of purposeful hand skills, increase in repetitive nonpurposeful hand movements, and development of breathing difficulties, all of which appear more noticeable during the child's awake periods as opposed to their sleep (International Rett Syndrome Foundation, 2014). Because of these symptoms, it is often this stage where a child will receive a diagnosis, although often the child may be improperly identified as having an autistic disorder or cerebral palsy.

Improvements in social interactions and behavior within the third stage, pseudo-stationary stage, are often met with additional concerns of apraxia, seizures, and enhanced motor difficulties. This stage, with an onset of between 2 and 10 years, is where individuals with Rett syndrome will stay for the majority of their lives (Bianciardi et al., 2013). This stage often represents the pinnacle of physical functioning and plateau for cognitive functioning and communication, which generally remain constant throughout the final stage, the late motor deterioration stage. Established to have a notable drop in physical strength and stamina (e.g., spasticity, muscle weakness, rigidity), individuals within this final stage may witness increases in their eye gaze and reductions in their repetitive hand movements (United Cerebral Palsy, 2015).

Given the desire to learn more about the disorder and increase the accuracy and timeliness of diagnosis, physicians will often employ a two prong approach that couples a genetic test, seeking information about the presence of a mutation, and a clinical diagnosis, which involves three criteria types. While the main diagnostic criteria provide the opportunity for a physician to identify the potential presence of Rett syndrome, it is the exclusion criteria that provide the most absolute criteria for the diagnosis. According to the International Rett Syndrome Foundation (2014), individuals

who meet any of the following criteria can be excluded from a classic Rett syndrome diagnosis: (1) traumatic brain injury, (2) neurometabolic disease, (3) neurological problems caused by a severe infection, and (4) significant abnormalities in gross development prior to the child reaching 6 months of age. If none of these criteria apply to the individual, and the child has been identified to have a period of regression accompanied by a period of stabilization, the physician will then review whether the child meets the main criteria for diagnosis. The criteria include (1) partial or complete loss of acquired purposeful hand skills, (2) partial or complete loss of spoken language acquisition, (3) abnormalities of gait, and (4) stereotypic repetitive hand movements, which must all be present to indicate a Rett syndrome diagnosis (Neul et al., 2010). Finally, the third category of symptoms, supportive criteria, which includes scoliosis, breathing problems when awake, abnormal sleep patterns, inappropriate laughing, and diminished response to pain, provide more symptoms that may be evident in some but not all of those with Rett syndrome and are therefore not required components of the diagnosis.

Gender and Cultural Factors

The occurrence of Rett syndrome has been observed to exist fairly equally across all racial, ethnic, and cultural groups but has been noted to be significantly more common in female births as opposed to male births. According to the International Rett Syndrome Foundation (2014), the presence of a gene mutation within the X chromosome often has more significant effects on males given that they do not have an unaltered copy of the gene to compensate for the mutated gene. In these cases, the male child is often unable to survive long after birth even though they do not necessarily show the clinical signs of Rett syndrome. In very rare cases, boys may experience a more sporadic mutation of their MECP2 gene, resulting in developmental or intellectual disabilities (National Institute of Neurological Disorders and Stroke, 2014). Given the scarcity of reported cases, however, there is minimal research on what may cause this to happen.

Prognosis

While questions remain about the factors leading to the gene mutation associated with Rett syndrome, researchers have also been stunted in their abilities to provide a clear prognosis for individuals with Rett syndrome beyond the ages of 40 and 50. According to United Cerebral Palsy (2015), the lack of research targeting adults with Rett syndrome, especially those over 40, provides little insight into the life expectancy for an individual with Rett syndrome. While researchers continue to target the causes and research for potential treatment, one thing is known for sure. There is no cure for Rett syndrome at this time. Instead, treatment outcomes are targeted around the successful management of symptoms with the hope that this will increase positive experiences for those affected by the disorder (National Institute of Neurological Disorders and Stroke, 2014). Management of these services must target their multidimensionality. While research has noted the potential impact of medical doctors to monitor and prescribe medications for various physical conditions including seizures and abnormal heart beat (e.g., Krajnc, Župančič, & Oražem, 2011), occupational therapists to target self-directed activities such

as feeding and dressing (e.g., United Cerebral Palsy, 2014), physical therapists to target mobility (e.g., Lotan & Hanks, 2006), nutritionists to target weight management (e.g., Reilly & Cass, 2001), and assistive technology including braces or wheelchairs to increase performance (e.g., Lotan, 2007), no study found to date targeted the use of counseling to target overall outcomes of adaptation to the disorder. The dearth of this information provides ample opportunity for further investigation into its effectiveness.

Comparing Rett Syndrome to Autism Spectrum Disorders

According to DeWeerd (2011), there exists a misconception within the field of counseling that the removal of Rett syndrome as a mental health diagnosis reflects an exclusion of importance. In reality, however, the removal of the diagnosis from the *DSM-5* (APA, 2013) serves more as a testament to the complexity of the associated symptoms and need for counselors to identify individualized plans to working with those with Rett syndrome. While a clinical assessment of an individual with Rett syndrome may indicate diagnostic criteria for an ASD diagnosis, this is not always the case. Moreover, even individuals with Rett syndrome who meet diagnostic criteria for an ASD diagnosis may experience additional symptoms which should be targeted in practice. As a result, the removal of Rett syndrome as a diagnoses in the *DSM-5* successfully removed the automatic categorization of individuals with Rett syndrome as individuals with ASD and provided the opportunity to reflect clinically significant symptoms that an individual may experience. In effect, the establishment of Rett syndrome is associated with physiological symptoms, while the notation of an Autism Spectrum Disorder addresses neurodevelopmental issues

Common Treatment Approaches

Historically, treatment of Rett syndrome has focused on the effective management of symptoms through pharmacology, physical skill building, and nutritional education. Because difficulties with breathing often hamper an individual with Rett syndrome's ability to swallow, studies have identified the importance of targeting nutrition through education as well as through the use of natural supplements (Percy, 2013). By targeting this very basic aspect of an individual's development, an individual's attentiveness, growth, and social interactions are optimized. Coupled with treatment for breathing issues, treatment for seizures is often a significant concern for medical providers. As the British Epilepsy Association (2015) pointed out, however, attempts to address seizures among individuals with Rett syndrome rarely are successful with eliminating the seizures completely and instead focus on the reduction of their occurrence. Hearing, vision, cardiac functioning, and muscle stiffness are also monitored regularly.

According to the DNA Exchange (2015), a Web site that allows individuals directly impacted by a genetic disorder to share their stories in a hope of increasing discussion, the multidisciplinary approach to targeting Rett syndrome can become somewhat overwhelming. Coupled with the uncertainties in the research and prognosis of this disorder, families are often faced with more questions than answers. Kaufmann et al. (2012) noted that the removal of Rett syndrome as a diagnosis of pervasive developmental disorders, changed to autism spectrum disorders in the newest version of

the *Diagnostic and Statistical Manual of Mental Disorders (DSM-5, APA, 2013)*, reflects on not only the uncertainties of the condition but also on the lack of saliency of symptoms. While Rett symptoms in early development may mirror autism spectrum disorder (ASD) features, individuals with Rett syndrome do not necessarily meet diagnostic criteria for an ASD diagnosis. As a result, providing treatment associated with ASD among individuals with Rett syndrome is not always a clinically sound judgment. Even in cases where an ASD diagnosis is appropriate, treatment of Rett syndrome should take into account various other factors.

A Comprehensive Team Approach

Historical approaches to treating individuals with Rett syndrome have relied heavily on taking an approach influenced by ASD research. The resulting evidence-based services for working with individuals with ASDs are both varied and plentiful. According to the American Speech-Language-Hearing Association (2015), these services and interventions are often centered around goals of reducing language impairments, improving social communication, and modifying social behaviors. Behavioral interventions and techniques, cognitive behavioral therapy, play therapy, and auditory/sensory integration training serve as just a handful of the most commonly used treatment approaches for work with this population (Rett Syndrome Research Trust, 2015).

While these approaches have been shown to be effective in the treatment of ASDs, the potential complexities of other symptoms associated with Rett Syndrome indicate the need for a more multimodal and comprehensive team approach in order to improve effective outcomes for those effected by Rett syndrome. As Galloway, Buckenmaier, Gallagher, and Polamano (2011) pointed out, disorders which involve both physiological and psychological symptoms are best addressed through these more comprehensive approaches.

Referral and Diagnosis

As the National Institute of Stroke and Neurological Disorders (2014) revealed, one of the most instrumental elements in the effective treatment of Rett syndrome is proper diagnosis. In removing Rett syndrome from the *DSM-5, APA (2013)* underscored the potential presence of physiological and psychological diversity within the disorder. Therefore, counselors must be cognizant of the potential presence of Rett syndrome among the individuals to whom they provide services in order to ensure that an accurate diagnosis is obtained.

APA (2013) revealed that while some individuals with Rett syndrome meet the diagnosis of an ASD, many others do not. Still others may be mislabeled as having a general ASD without addressing the physiological causes. As standard practice, it may become beneficial for counselors working with individuals with ASD to make medical referrals in order to have the potential presence of Rett syndrome be addressed. Because results from Rett syndrome research indicate genetic markers, a medical assessment will likely serve as a proactive approach to treatment. Results of the assessment may provide insight into services that may need to be put in place to help the individual and family.

Education and a Consistent Message

While the exclusion of Rett syndrome within the *DSM-5* (APA, 2013) served multiple purposes, the misinformation and misconceptions about these reasons serves as a significant risk factor to the quality of treatment individuals with Rett syndrome may receive within the counseling profession. The DNA Exchange (2015) revealed that these frequent misconceptions cause a Rett syndrome diagnosis to be communicated as a worst case diagnosis. These worst case projections for individuals with Rett syndrome do not accurately reflect the potential lives they are capable of living (Rocky Mountain Rett Association, n.d.).

As the National Association of Mental Illness (NAMI; 2014) revealed, psychoeducation is an important element of working with individuals with chronic diseases as well as their family members. Learning about one's illness and how to effectively receive treatment is central to this approach. Because the type and severity of symptoms varies tremendously, this must be effectively communicated with the family.

While previously identified as the most severe of the pervasive developmental disorders, symptoms of Rett syndrome range from mild to severe in nature (International Rett Syndrome Foundation, 2014). Individuals with more severe symptoms of Rett syndrome are likely to see the most significant limitations in the areas of independent living skills. According to United Cerebral Palsy (2015), individuals with severe gross motor symptoms often require support in order to successfully complete activities of daily living. Some may require breathing or feeding tubes, have difficulty dressing themselves, and struggle with going to the bathroom independently. In these cases, residential or 24-hour care may be required.

Huppke et al. (2006) pointed out, however, that while Rett syndrome represents the second largest cause of intellectual disability among girls next to Down syndrome, not all individuals experience these symptoms. In these cases, individuals diagnosed with Rett syndrome demonstrated the ability to perform numerous independent living tasks including feeding themselves and dressing themselves. While these individuals meet the diagnostic classification of Rett syndrome, the mildness of symptoms may often cause such cases to go undiagnosed (Huppke et al., 2006), further supporting the need for additional collaboration to ensure accurate diagnosis.

Targeting Specific Symptoms

For counselors, it is important to understand the maximum functional capacity of an individual with Rett syndrome in order to develop a treatment approach. Given the potential for cognitive, social, developmental, and physical symptoms (International Rett Syndrome Foundation, 2014), counselors must evaluate what symptoms apply to a given individual and develop approaches based on this evidence. For example, an individual with Rett syndrome who meets criteria for an ASD would likely benefit from integrating evidence-based ASD approaches, while one who meets criteria for an intellectual disability may benefit from treatment approaches, including early intervention, which have been integrated in work with individuals with Down syndrome or other intellectual disorders to increase opportunities for independent functioning (National Institute of Child Health and Human Development, 2014). Physiological symptoms, on the other hand, would require evidence-based intervention associated with specific areas of chronic

health care. Taking into account the multiple areas of need and developing integrated interventions based on these areas of assessment are important.

One prevalent area which therapeutic approaches should target among individuals with Rett syndrome is self-esteem, given the multiple risk factors for low self-esteem. Specifically, issues related to the potential for lowered self-esteem have been linked to females (Orenstein, 2013), women with mental illness (Rusch, Lieb, Bohus, & Corrigan, 2006), individuals with learning disabilities (Hellenbach, Brown, Karatzias, & Robinson, 2015), individuals with physical disabilities (Nosek, Hughes, Swedlund, Taylor, & Swank, 2003, and individuals who perceive themselves as a burden (Nosek, Robinson-Whelen, Hughes, & Mackie, n.d.). For individuals with Rett syndrome, there is a significant potential for all of those characteristics to apply. Therefore, it is imperative that treatment focus on potential issues of self-esteem for increased success in treatment and optimizing the functional capabilities of individuals with this disorder.

Systemic Approaches

While targeting the individual with Rett syndrome through various physical and psychological avenues is important, counselors must also provide support and education to members of the family and other support systems. As the DNA Exchange (2015) noted, doctors often have a difficult time discussing issues regarding Rett syndrome with family members. While some doctors attempt to provide hope through making promises about the functioning level of the individual with Rett syndrome, these may not be accurate and may leave family members with disappointment. On the other hand, doctors' approaches that portray the Rett syndrome diagnosis as a worst case scenario could eliminate hope in the first place, resulting in unclaimed potential.

Because individuals with Rett syndrome may require supports in independent living or with developing increased self-esteem, the family system is an important component of any recovery plan. As United Cerebral Palsy (2015) pointed out, this can often be overwhelming for the family tasked to care for their child. Because of this, education and support are important aspects to consider. As Jeon, Brodaty, O'Neill, and Chesterton (2006) identified, respite care should also be explored, though focus may be placed on identifying more extended supports given that the quality of care offered through respite services has been largely questioned in the literature and is often a question of the caretaking family members.

Case management is an important element of ensuring systemic collaboration. According to America's Health Insurance Plans (2007), case management is an invaluable tool in the treatment of chronic medical conditions. Because treatment options may be plentiful, they are also often overwhelming. Therefore, the counselor has the unique opportunity to navigate clients through the process, to identify resources in the community, and when absent to develop them. While researchers have not targeted the importance of case management in working with individuals with Rett syndrome, the similarity between this disorder and other chronic medical conditions signals the potential effectiveness of such an approach.

Assistive Technology

Although numerous questions and uncertainties remain regarding the underlying causes of Rett syndrome, its prognosis, and the outcomes of treatment due to its relative

infancy in research, the potential to optimize functioning through the use of technology has been noted. Specifically, physical devices that provide support, can be contorted to the individual's shape, allow mobility, and reduce back pressure have all been identified to have possible implications in the effective treatment for individuals with Rett syndrome (Lotan, 2007). While some of these devices reduce symptom effect, others focus on increased independence. Augmentative devices used to target communication skills may also be beneficial in increasing individuals with Rett syndrome's ability to interact with others and communicate their needs (International Rett Syndrome Foundation, 2014). When paired with other therapeutic approaches (e.g., speech therapy, physical therapy, occupational therapy, counseling, etc.), these tools can provide increased opportunities for individuals with Rett syndrome that would not have been possible otherwise.

Identifying how to best support individuals with Rett syndrome both in the classroom and on the job is important in increasing their functioning level and reducing the psychological difficulties these individuals and their families face. According to the International Rett Syndrome Foundation (2014), increased independence through the use of assistive technology can reduce the burden. Therefore, as a counselor, it is important to understand and integrate a multidimensional approach when working with families of an individual with Rett syndrome.

Case Illustration

Pablo and Jamie were beyond excited when they learned that they were expecting their first child. After years of unsuccessful attempts, trips to the specialists, and increasing stress, they found out that they were having a baby girl. Life was amazing for the first year. Their daughter, Izabel, was the light of their eyes. She was the ideal baby. She slept through the night and met all of her developmental milestones, sometimes in advance. She learned to roll over at 3 months, sit at 7 months, crawl at 9 months, and walk at 11 months. Her first words were followed by many more.

Suddenly, at about 13 months, Jamie became concerned. Izabel appeared to begin losing her happy disposition. She was no longer entertained by her toys and stopped talking completely. After taking their daughter to the pediatrician, they were referred to various specialists. About six months later, they finally received the diagnosis. Their daughter was diagnosed with Rett syndrome. Doctors told the family, "I wish I had better news. It is only going to get worse from here." Devastated, the couple seeks out counseling.

A new counselor, Bryanna, recognizes the issues that the family faces regarding the impact of their daughter's diagnosis on their hopes and dreams for her in the future. Bryanna, herself, lost a daughter shortly after birth. She knows that she needs to develop rapport with the family and to instill a sense of hope in them. As a strength-focused counselor, Bryanna, believes that the family has the abilities to make it through this difficult time and would benefit from counseling to help them utilize these skills. She notes the following things that she must take into consideration and do in order for a successful treatment outcome.

Acknowledge Grief

Being informed that a loved one has a significant illness can be a significant time of grief for many individuals (Waldrop, 2007). While the loved one is still alive, family members are often faced with letting go of their expectations and the person their loved one was before the illness. Given that the family has just learned their child has this condition, grief would be normal and expected. Therefore, it is important for Bryanna to take into account that the family is likely to enter the various stages of grief.

Provide Education and a Consistent Message

As the International Rett Syndrome Foundation (2014) noted, each case of Rett syndrome is unique in its course, symptoms present, and the severity of these symptoms. While Izabel's symptoms are likely to get worse, the family has some ability to affect the outcome through early intervention. Percy (2013) identified the value of early intervention in treating symptoms and increasing optimal levels of functioning. Through the use of research, Bryanna is able to provide the clients with tools to keep them better informed and allow them to have a more realistic understanding of the potential needs of their daughter.

A Comprehensive Picture

Establishing professional communication between the counselor and medical provider allows for comprehensive assessment of Izabel's limitations. Through this assessment, an individualized treatment plan can be developed. Establishing goals with the family should encompass the already known strength of the family's resiliency (Retzlaff, 2007). As Nolan, Luther, Young, and Murphy (2014) revealed, parents of children with disabilities are more likely to want to develop goals that integrate optimism while their physicians often focus more on the potential setbacks that a client may experience. Knowing this, Bryanna needs to utilize the family's optimism to set realistic goals for themselves and their daughter, while leaving open the possibility that goals may need to be adjusted without representing that something is a failure. By doing so, the counselor can support in providing realistic expectations to support the family in maintaining their hope and optimism.

Establish Supports

While there is no way to anticipate the amount of supports that the family may need, Bryanna knows that the more information and resources she can provide the family, the better. She begins by creating a resource guide for the family that lists local physical therapists, occupational therapists, speech therapists, and neurologists. She notes the importance of providing professional resources for the family and also provides the family with information on various support groups. While there are no groups that specifically target Rett syndrome, she identifies groups that address other conditions and may provide the family with additional resources and shared experiences. These community tools may be beneficial to the family at no cost. Finally, Bryanna helps the family complete a genogram to identify informal family supports which may also be helpful.

Assess Family Self-Care

Given Weiner's (2014) notation of the prevalence of caregiver burnout and the potential limitations that an individual with Rett syndrome may experience, it is important to ensure that the family maintains a sense of balance and self-care. Because caregivers prioritize the needs of their loved ones often above their own, this becomes an important element for counselors to continue to assess. In the event that self-care becomes an issue, the counselor must revisit the supports in place.

Case Reflection

Izabel's diagnosis of Rett syndrome offers a cursory notation of symptoms which could apply to her development. Under classification in the *DSM-IV* (APA, 1994), Izabel would automatically be categorized with other ASD disorders and treatment would be developed based on this assessment. Under *DSM-5* (APA, 2013) classification, however, the counselor must gain more information in developing effective treatment approaches. By dissecting the various symptoms associated with Izabel and applying evidence-based treatment approaches based on these symptoms, the counselor has additional tools to add towards treatment success.

Conclusion

While research on Rett syndrome is underdeveloped as opposed to research on other genetic conditions and implications for counselor virtually neglected, counselors must be prepared to work with families and individuals affected by this condition. Understanding the diversity of symptoms and symptom intensity is important in providing a realistic hope, while providing a multifaceted approach can offer support in coping with the effects of Rett syndrome. Future research may be a valuable tool in delivering effective services.

References

- American Health Insurance Plans. (2007). *Innovations in chronic care: A new generation of initiatives to improve America's health*. Retrieved from <https://www.ahip.org/Innovations-in-Chronic-Care>
- American Psychiatric Association. (1994). *Diagnostic and statistical manual of mental disorders* (4th ed.). Washington, DC: Author.
- American Psychiatric Association. (2013). *Diagnostic and statistical manual of mental disorders* (5th ed.). Washington, DC: Author.
- American Speech-Language-Hearing Association. (2015). *Autism*. Retrieved from <http://www.asha.org/Practice-Portal/Clinical-Topics/Autism/>
- Bianciardi, G., Acampa, M., Lamberti, I., Sartini, S., Servi, M., Biagi, F., & ... Pastorelli, M. (2013). Microvascular abnormalities in Rett syndrome. *Clinical Hemorheology & Microcirculation*, 54(1), 109–113. doi:10.3233/CH-131707
- British Epilepsy Association. (2015). *Rett syndrome: Epilepsy action*. Retrieved from <https://www.epilepsy.org.uk/info/syndromes/rett-syndrome>

- DeWeerd, S. (2011). Reclassification of Rett syndrome diagnosis stirs concerns. *Simons Foundation Autism Research Initiative*. Retrieved from <http://sfari.org/news-and-opinion/news/2011/reclassification-of-rett-syndrome-diagnosis-stirs-concerns>
- Dolce, A., Ben-Zeev, B., Naidu, S., & Kossoff, E. H. (2013). Rett syndrome and epilepsy: An update for child neurologists. *Pediatric Neurology*, 48(5), 337–345. doi:10.1016/j.pediatrneurol.2012.11.001
- DNA Exchange. (2015). *The DNA exchange*. Retrieved from <http://thednaexchange.com/tag/rett-syndrome>
- Galloway, K. T., Buckenmaier, C. C., Gallagher, R. M., & Polomano, R. C. (2011). Multimodal and multidisciplinary therapy for pain management. *American Nurse Today*, 6(9), 13–18.
- Hellenbach, M., Brown, M., Karatzias, T., & Robinson, R. (2015). Psychological interventions for women with intellectual disabilities and forensic care needs: A systematic review of the literature. *Journal of Intellectual Disability Research*, 59, 319–331. doi:10.1111/jir.12133
- Huppke, P., Maier, E. M., Warnke, A., Brendel, C., Laccone, F., & Gartner, J. (2006). Very mild cases of Rett syndrome with skewed X inactivation. *Journal of Medical Genetics*, 43(10), 814–816.
- International Rett Syndrome Foundation. (2014). *Rettsyndrome.org*. Retrieved from <http://www.rettsyndrome.org/>
- Jeon, Y.-H., Brodaty, H., O'Neill, C. and Chesterson, J. (2006), 'Give me a break'—respite care for older carers of mentally ill persons. *Scandinavian Journal of Caring Sciences*, 20, 417–426. doi:10.1111/j.1471-6712.2006.00423.x
- Kaufmann, W. E., Tierney, E., Rohde, C. A., Suarez-Pedraza, M. C., Clarke, M. A., & Salorio C. F. (2012). Social impairments in Rett syndrome: Characteristics and relationship with clinical severity. *Journal of Intellectual Disability Research*, 56, 233–247.
- Krajnc, N., Župančič, N., & Oražem J. (2011). Epilepsy treatment in Rett syndrome. *Journal of Child Neurology*, 26(11), 1429–1433. doi: 10.1177/0883073811408422.
- Leonard, H., Fyfe, S., Leonard, S., & Msall, M. (2001). Functional status, medical impairments, and rehabilitation resources in 84 females with Rett syndrome: A snapshot across the world from the parental perspective. *Disability & Rehabilitation*, 23(3/4), 107–117. doi:10.1080/09638280150504180
- Lotan, M. (2007). Assistive technology and supplementary treatment for individuals with Rett syndrome. *Scientific World Journal*, 12(7), 903–948.
- Lotan, M., & Hanks, S. (2006). Physical therapy intervention for individuals with Rett syndrome. *Scientific World Journal*, 10(6), 1314–1338.
- National Association of Mental Illness. (2014). *Psychosocial treatments*. Retrieved from <https://www.nami.org/Learn-More/Treatment/Psychosocial-Treatments>
- National Institute of Child Health and Human Development. (2014). *Rett syndrome*. Retrieved from <http://www.nlm.nih.gov/medlineplus/rettsyndrome.html>
- National Institute of Neurological Disorders and Stroke. (2014). *Rett syndrome fact sheet*. Retrieved from http://www.ninds.nih.gov/disorders/rett/detail_rett.htm

- Neul, J. L., Kaufmann, W. E., Glaze, D. G., Christodoulou, J., Clarke, A. J., Bahi-Bussoin, N., ... Percy, A. K. (2010). Rett syndrome: Revised diagnosis criteria and nomenclature. *Annals of Neurology*, 68, 944–950. doi:10.1002/ana.22124
- Nolan, R., Luther, B., Young, P., & Murphy, N. A. (2014). Differing perceptions regarding quality of life and inpatient treatment goals for children with severe disabilities. *Academic Pediatrics*, 14(6), 574–580. doi:10.1016/j.acap.2014.02.012
- Nosek, M. A., Hughes, R. B., Swedlund, N., Taylor, H. B., & Swank, P. (2003). Self-esteem and women with disabilities. *Social Science and Medicine*, 56, 1737–1747.
- Nosek, M. A., Robinson-Whelen, S., Hughes, R. B., & Mackie, P. (n.d.). *Self-esteem and women with physical disabilities*. Retrieved from www.spinalcord.org/pdf/body-image-self-esteem-webinar.pdf
- Orenstein, P. (2013). *Schoolgirls: Young women, self esteem, and the confidence gap*. New York, NY: Knopf Doubleday Publishing Group.
- Percy, A. K. (2013). Path to treat Rett Syndrome. *Science*, 342(6156), 318–322. doi:10.1126/science.1245657
- Reilly, S., & Cass, H. (2001). Growth and nutrition in Rett syndrome. *Disability and Rehabilitation*, 23(3-4), 118–28.
- Rett Syndrome Research Trust. (2015). *Education*. Retrieved from <http://www.rsrt.org/rett-and-mecp2-disorders/education/>
- Retzlaff, R. (2007). Families of children with Rett syndrome: Stories of coherence and resilience. *Families, Systems & Health: The Journal of Collaborative Family Healthcare*, 25(3), 246–262. doi:10.1037/1091-7527.25.3.246
- Rocky Mountain Rett Association. (n.d.). *What is Rett?* Retrieved from <http://rmrett.org/what-is-rett/>
- Rusch, N., Lieb, K., Bohus, M., & Corrigan, P. W. (2006). Self-stigma, empowerment, and perceived legitimacy of discrimination among women with mental illness. *Psychiatric Services*, 57(3), 399–402.
- United Cerebral Palsy. (2015). *My child without limits*. Retrieved from <http://ucp.org/what-we-do/public-education/my-child-without-limits/>
- Waldrop, D. P. (2007). Caregiver grief in terminal illness and bereavement: A mixed-methods study. *Health and Social Work*, 32(3), 197–206.
- Weiner, L. (2014). *Caregiver research at the NIH clinical center*. Retrieved from <http://clinicalcenter.nih.gov/wecare/research.html>

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