

Supporting Individuals with Rubinstein-Taybi Syndrome in Education

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Abstract

Educators will be tasked with planning for children with many different needs. The aim of this article is to provide specific information about Rubinstein-Taybi Syndrome (RTS) to those who may be interested. This syndrome is rare and has not been well researched. This article contains information about the physical, cognitive, and developmental differences of a child with RTS. There are also recommendations for working with children who have RTS.

Over one billion people in the world are living with some form of a disability that impacts their quality of life (Cooc, 2019). Addressing and supporting physical, mental, intellectual, and sensory impairments can be a challenging task. Rubinstein-Taybi syndrome (RTS) is a rare syndrome that is not well known. Individuals with RTS have well-defined physical features, motor development delays, common behavioural traits, and varying degrees of intellectual disability (López et al., 2018). Gaining a better understanding of this syndrome will improve educators' self-efficacy and allow them to provide support to individuals with RTS.

RTS is not well researched, and many individuals are unfamiliar with it. The lack of knowledge would make it very difficult for educators to support a child with RTS; therefore, it is important to gain a better understanding of the syndrome. There are specific physical characteristics, motor abilities, behavioural characteristics, and cognitive abilities that educators should be aware of. RTS occurs with one in every 100 000 to 125 000 children (Milani et al., 2015). There is less than a 1% chance that families will have a second child with RTS (Stevens, 1997). Two possible gene mutations are known to cause RTS (Cazalets et al., 2017). Over half of the RTS cases are caused by a mutation on the CREBBP gene ("Rubinstein-Taybi Syndrome," 2019). The other gene mutation known to cause RTS is the EP300 gene, which accounts for approximately 8% of cases ("Rubinstein-Taybi Syndrome," 2019). The remainder of RTS cases are caused by unknown factors ("Rubinstein-Taybi Syndrome," 2019). This syndrome can be diagnosed after birth with genetic testing, but the majority of cases are confirmed by the physical features of individuals with RTS (Milani et al., 2015).

Individuals diagnosed with RTS have very distinct physical features. A couple of distinctive features are their broad angulated thumbs and enlarged toes (López et al., 2018). Broad nasal bridge, slanted eyes, and high arched eyebrows are some other prominent physical characteristics (Waite et al., 2015). Based on these physical differences, individuals are often diagnosed at birth or in their early stages of life (López et al., 2018). Height, weight, and head circumference are often normal in the prenatal stages, but are in a very low percentile during infancy and the early stages of life (Sescleifer & Stevens, 2018). Those with RTS will face a large range of medical issues, and individuals will have their own sets of obstacles to overcome. Most individuals will have some form of feeding difficulty, including reflux and vomiting (Roome & Ade, 2013). RTS is not something that is curable, but having an understanding of the syndrome will help those involved with these individuals to be supportive. Beyond obvious physical characteristics, children with RTS have delayed motor development.

Motor development delays are common for most individuals with RTS, due to low muscle tone (Char et al., 2019). For instance, an average time for those with RTS to begin walking is two and a half years of age (Edens Hurst, 2017). These individuals are often considered clumsy and they deal with poor coordination throughout their lives (Cazalets et al., 2017). Interestingly, motor difficulties are closely intertwined with cognitive development (Cazalets et al., 2017).

Motor problems were considered to be related to postural or gait deficiencies, but they actually have more to do with poor attention span and visuomotor impairment (Cazalets et al., 2017). Taking this into consideration, it is helpful for these individuals to have access to early intervention services and play-based therapies (Char et al., 2019). In addition to motor development delays, individuals with RTS have common behavioural characteristics.

There are similarities in the behaviours of RTS individuals with Autism Spectrum Disorder and Obsessive Compulsive Disorder (Char et al., 2019). There are repetitive behaviour tendencies, including repetitive questioning (Waite et al., 2015). This repetitive questioning could be to gain caregiver attention, or to help with memory, because it has nothing to do with social-communication issues. Some of the behaviour issues that occur could be in relation to a major life event (Char et al., 2019, p. 15). Examples include surgeries, schedule changes, lack of routine, or a new respite worker. Large crowds or new situations can cause over stimulation and lead to impulsiveness or moodiness (Sescliefer & Stevens, 2018). Although there are similarities to other disorders, RTS is a unique syndrome and should be treated accordingly. More commonly, people with RTS are described as friendly, loving, and very happy by those around them (Milani et al., 2015). They certainly know how to connect with others through physical touch and eye contact. Social and emotional development is actually considered advanced in some respects with these RTS individuals (Char et al., 2019). Although social and emotional development is positive, there are some cognitive struggles faced by those with RTS.

The cognitive abilities of those with RTS range greatly, but moderate intellectual disability is most common (Waite et al., 2015). Approximately 90% have speech problems, including speech delay and articulation issues (Stevens, 1997). Sign language is used by nearly half of the RTS population, and another 6% use it as their only form of communication. Reasoning, learning, and problem solving are a challenge for some individuals (Char et al., 2019). The majority of those with RTS have very short attention spans, which affects overall skills development (Stevens, 1997). The young brain is very adaptable; therefore, early intervention is encouraged (Char et al., 2019). These individuals are very capable of learning new skills throughout life (Stevens, 1997), and therefore need to be adequately supported by those involved with them.

Determining the individual needs of a child with RTS is important because it is a rare syndrome that has unique characteristics (Char et al., 2019). Teachers and school therapists need to make a connection with parents. Early intervention is recommended, but can be very overwhelming for both the child and the parents (E. Mangin, parent of a child with RTS, personal communication, June 12, 2019). With this connection, challenges can be approached as a unified team. Every child with RTS will need some sort of special needs support and would benefit greatly from an Individual Education Plan. As a team, plans can be made to support the motor, behavioural, and cognitive needs of the child (Char et al., 2019). Physical therapy, speech therapy, and social skills training will all benefit a child with RTS. In terms of language development, technology and communication devices could be a huge asset. Teachers should also work to create a welcoming and inclusive classroom environment. Educators can start by becoming familiar with the child's specific needs. A useful way to become more familiar with a rare syndrome is to join an online support community (E. Mangin, personal communication, June 12, 2019). These communities may be able to answer specific questions, and suggest tools that may help support the child. With a better understanding of the syndrome, educators can create a learning environment to help the children meet their full potential. Unfortunately, there is very little research-based information on RTS in education. Taking the initiative to join in professional development, and gaining the confidence to apply new information in an inclusive classroom setting, will make all the difference in supporting a child with RTS.

Conclusion

A large percentage of our population is faced with some form of a disability. With such a wide range of disabilities, it is important that teachers seek out specific information about the

students in each new class. RTS is one of many rare syndromes, and many are unfamiliar with the characteristics and needs of a child with RTS. Individuals with RTS have unique physical characteristics and motor development delays. They also have some intellectual setbacks and behavioural differences. It is very important to be open minded, and work to find new ways to support these children and the needs of all students during any given year.

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