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The Educational Experiences of Children and Adolescents with an RYR1-Related Disease

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THE EDUCATIONAL EXPERIENCES OF CHILDREN AND ADOLESCENTS WITH A RYR1-RELATED DISEASE

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By

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ABSTRACT

THE EDUCATIONAL EXPERIENCES OF CHILDREN AND ADOLESCENTS WITH A RYR1-RELATED DISEASE

By

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August 2024

Dissertation supervised by Tammy Hughes, PhD, ABPP.

This paper investigates the educational experiences of children and adolescents diagnosed with RYR1-related diseases, a spectrum of congenital myopathies affecting muscle function. Despite these conditions typically sparing cognitive abilities, they profoundly impact physical capabilities, presenting challenges in school environments such as participation in physical activities and social interactions. The study utilizes data collected through the RYR1-Related Disease Educational Impact and Satisfaction Survey (RDEISS) and the Behavioral and Emotional Screening System (BESS) from 49 parents of 51 children aged 3-18 years. Findings reveal prevalent classroom difficulties including handwriting issues and written expression difficulties, with 78% of children receiving special education services, primarily through Individualized Education Programs (IEPs) and Section 504 Plans. Parental satisfaction with school services is generally high (80%), despite challenges in communication and

implementation. Most children exhibit normal behavioral and emotional profiles, yet some experience heightened internalizing behaviors. These insights underscore the necessity for tailored educational support and interdisciplinary collaboration to optimize educational outcomes for children with RYR1-related diseases.

Keywords: RYR1-related diseases, congenital myopathies, educational experiences, special education, parental satisfaction

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CHAPTER I

INTRODUCTION

In the 2008 National Hockey League (NHL) draft, with the 10th pick in the first round, the Vancouver Canucks selected Cody Hodgson. After being drafted, Cody Hodgson, a Canadian-born hockey player, went on to play 328 games, tallying 64 goals and 78 assists, totaling 142 points in just seven seasons. His promising NHL career was unfortunately cut short when he suddenly retired from the game in 2016, following his seventh season in the NHL; he was only 28 years old at the time. In 2016, Cody was diagnosed with Malignant Hyperthermia, a rare muscle disorder caused by a genetic mutation in the RyR1 gene, also classified as a RYR1-related disease. Cody said, "the doctors basically told me it was too dangerous for me to play. It wasn't really a choice to stop playing. It really wasn't anything that I could control" (Vingan, 2018, p. 2).

Cody is extremely fortunate to have ever played in the NHL at all, considering the severity of the symptoms that can be associated with RYR1-related diseases. However, RYR1-related diseases impact each individual differently. Unlike Cody's case, most symptoms of RYR1-related diseases are present at birth or early infancy (National Organization for Rare Disorders, 2021).

Growing up with a RYR1-related disease can be detrimental to a child's quality of life and educational experience at school. Accordingly, educational staff, families, and children need to be aware of the types of support necessary for children to be successful at school. In this chapter, RYR1-related diseases will be reviewed, core symptoms and review how these symptoms can impact a children's development and educational experience and success.

RYR1-Related Diseases

RYR1-related diseases are inherited forms of congenital myopathy potentially resulting in a wide range of symptoms, particularly in varying degrees of muscle weakness. These diseases may also impact bones, joints, breathing, eating, swallowing, and speaking. In addition, RYR1-related diseases can contribute to a potentially fatal reaction to certain anesthetics or excessive physical exertion, known as malignant hyperthermia. The wide range of symptoms of RYR1-related diseases are typically either non-progressive or very slowly progressive. Individuals inherit a defective mutation from one (autosomal dominate) or both parents (autosomal recessive), or a spontaneous mutation (de novo). The RyR1 receptor is a channel in muscle cells that regulate the flow of calcium, a critical component of muscle contraction, and thus a critical component in how individuals move. A reduced number or abnormal RyR1 channels lead to dysfunctional muscle contraction, and consequently weak or impaired movement.

RYR1-Related Diseases Symptoms

Since individuals are born with this disease, it is important to consider the progression of their symptoms and how such symptoms may affect their educational experience. As previously discussed, RYR1-related diseases have a wide range of severity and symptoms (Clinical Care Guidelines, 2019). Additionally, different symptoms can be present even among affected individuals within the same family (National Organization for Rare Diseases, 2021). General or common symptoms present in RYR1-related diseases are weakness of the eye muscles, weakness of the facial muscles, weakness of muscles close to the torso (proximal muscle weakness), increased susceptibility to muscle cramping and pain, and decreased tolerance to exercise and heat (National Organization for Rare Diseases, 2021).

Skeletal Muscles

Typically, RYR1-related diseases predominantly affect skeletal muscles, which are the muscles attached to bones used for voluntary movement. This weakness is due to the core function of the RyR1 channel and its relationship with calcium. The inability of the RyR1 channel to properly regulate calcium throughout the body results in weak muscle contractions. Largely, individuals with RYR1-related diseases experience muscle weakness all the time (Clinical Care Guidelines, 2019).

Bones and Joints

People suffering from a RYR1-related disease may also experience orthopedic problems including overall contractures, hip dislocations, and spinal deformities (i.e., scoliosis; Clinical Care Guidelines, 2019). A contracture refers to a joint that does not have a full range of motion. Hip dislocations occur when the hip bone is out of its joint. Individuals with a RYR1-related disease may also have a variety of spinal deformities such as scoliosis, Kyphosis, and Lordosis, all of which have a negative effect on an individual's posture (Clinical Care Guidelines, 2019).

Breathing

An individual's breathing may also be negatively impacted by an RYR1 gene mutation resulting in a dysfunctional RyR1 receptor. In the body, all cells need oxygen to survive, which causes cells to produce energy. When cells produce energy, they expel carbon dioxide. Breathing is an essential component for cell function, considering that breathing brings oxygen into the body, allows cells to survive and produce energy, and releases carbon dioxide, the by-product of this process (Clinical Care Guidelines, 2019). The diaphragm is the primary breathing muscle located underneath the lungs. It is responsible for pulling oxygen into the body and expelling carbon dioxide. RYR1-related diseases can cause weakness in the diaphragm, making it difficult

to clear the airways during breathing (Clinical Care Guidelines, 2019). This may contribute to negative outcomes ranging from mild symptoms in which individuals need help breathing when they have a respiratory infection or common cold to severe problems in which they will need a ventilator to help them breathe.

Eating, Swallowing, and Speaking

Another symptom of RYR1-related diseases is difficulties eating, swallowing, and speaking. This is due to muscle weakness in the face, mouth, and throat, also known as the bulbar muscles. When eating and swallowing, the muscles in the mouth and jaw help you chew and break down the food. Once the food is prepared to be swallowed, the muscles in your mouth and throat work together to direct the food towards the esophagus and away from the nose and trachea (windpipe). The eating and swallowing process may be adversely affected for individuals with bulbar muscle weakness such that, it may take longer to eat, they may drool, and food could enter the nose and/or trachea when eating. Additionally, bulbar muscle weakness may negatively impact speaking. When individuals speak, they use their lungs, and larynx (voice box) to produce sound. They then use their facial muscles (i.e., jaw, lips, and tongue) to form that sound into words. Individuals with a RYR1-related disease with bulbar muscle weakness may suffer from a weak voice, problems controlling breathing, and structural abnormalities in the mouth. (Clinical Care Guidelines, 2019).

Malignant Hyperthermia

Malignant Hyperthermia (MH) is a severe and dangerous reaction to certain anesthesia. This reaction causes the body to overheat to the point that the muscle tissue breaks down. This reaction also causes muscle spasms, rapid heart rate, and rigid muscles (Mayo Clinic, 2020). If an individual with MH is not treated quickly, they can suffer from kidney failure, brain damage,

cardiac arrest, additional organ failure and even, in extreme instances, death. Individuals with a RYR1-related disease are known to have Malignant Hyperthermia Susceptibility (MHS), which describes their susceptibility to MH due to a mutated gene. Unfortunately, MHS in individuals can cause muscle breakdown, severe muscle cramping, muscle stiffness, and heat intolerance upon significant exertion (Clinical Care Guidelines, 2019).

Contemporary literature suggests that RYR1-related diseases have no direct impact on a child's cognitive abilities and intelligence (Topaloglu, 2020). Despite this fact, the physical symptoms associated with RYR1-related diseases can be detrimental to a child's quality of life and functioning within a school.

RYR1-Related Diseases Impact on Educational Experience

A child with a RYR1-related disease may struggle with such routine activities as standing in the lunch line, eating lunch within a short timeframe, walking up and down stairs, participating in gym class or other extracurricular activities that require strenuous movement. Furthermore, this lack of participation and engagement in school activities (i.e., physical education, field day, team sports) may have a negative impact on this child's social interactions throughout the school day. Less opportunity to engage in social interactions results in less opportunity to make, develop, and maintain friendships. This becomes particularly difficult for youths whose friendships often center around sports and other physical activities. Additionally, individuals with RYR1-related diseases report significantly more impaired alertness, social interaction, higher levels of somatization, poorer physical functioning, social functioning, pain, and general health when compared to healthy individuals (Ruitenbeek et al., 2019).

A substantial literature base supports RYR1-related adverse effects on an individuals' physical abilities (Amburgey et al., 2013; Bharucha-Goebel et al., 2013; Clinical Care

Guidelines, 2019; Klein et al., 2012; Lawal et al., 2020; Todd et al., 2018; Voermans et al., 2016; Zhou et al., 2007). Contemporary studies also support that RYR1-related diseases can harm individuals' mental health and social functioning (Ruitenbeek et al., 2019; Lue et al., 2016) suggesting the increased likelihood of a diverse, strenuous, and adversely affected educational experience. Consequently, in order to help advance the extant literature, I will investigate the school experiences of children with a RYR1-related disease. Therefore, providing valuable knowledge to inform both families and related professionals (i.e., school personnel, health service providers, medical professionals) of how a RYR1-related disease might impact a child's educational experience. Through this research, I hope to answer the following questions:

Research Questions

1. What are the common classroom problems experienced by children with a RYR1-related disease?

2. With what frequency do children with RYR1-related diseases receive special services in schools (i.e., formal special education. Section 504 Plan, informal services, no services at all)?

3. If a child does receive special education services, under what IDEA category(ies) does the child qualify?

4. For children who do receive special services (i.e., IEP or Section 504 Plan) at school, what services do they receive and what is the parental satisfaction with those services?

5. Do children with a RYR1-related disease experience behavioral and emotional problems as reported by parent/caregivers on the Behavioral and Emotional Screening System (BESS)?

Summary

Throughout this chapter, I provided the framework and foundational knowledge for understanding RYR1-related diseases and their impact on a child's physical and mental

wellbeing. This brief introduction into RYR1-related diseases supports that the various negative outcomes of this disease can be detrimental to a child's educational experience and overall wellbeing. Cody Hodgens says that he is very fortunate to have played in the NHL for six years, and while playing, he made many great memories and friends (Vingan, 2018). Some individuals with a RYR1-related disease will not get the opportunity to become professional athletes; some may not get the chance to play sports at all. Growing up with a RYR1-related disease can be detrimental to a child's quality of life and educational experience at school. The foundational knowledge presented in this chapter paves the way for the importance of further research into children with RYR1-related diseases, specifically relating to their experiences in childhood and education. This research can inform educational staff, medical professionals, families, and children about this disease's effects on childhood experiences and help inform the support necessary to help these children succeed at school. In the next chapter, I will thoroughly review the literature on RYR1-related diseases and the biological mechanisms of these diseases. In addition, I will discuss relevant school-related services that are useful to help promote the overall well-being and educational experience of individuals with a RYR1-related disease.

CHAPTER II

LITERATURE REVIEW

Historical Background and Significance

RYR-1-related diseases result in the muscles' inability to regulate calcium throughout the body, a necessary mechanism of muscle contraction. Thus, individuals with a defective mutation of the RYR1 gene experience mild to severe muscle weakness. Although there is a wide range of symptoms of RYR-1-related diseases, they are typically either non-progressive or very slowly progressive. Limitations in physical movement is often detrimental to a child's educational experience at school. Consequently, it is important for educational staff, families, and children to be aware of the types of support needed to help foster a positive and successful educational experience. This chapter, provides an in-depth description of RYR1-related diseases introduced previously, including their prevalence, range of severity, developmental course of the disease, inheritance, and diagnosis. This chapter will further elaborate on the clinical features of RYR1-related diseases. It will also discuss potential treatments available for individuals with a RYR1-related disease. Furthermore, this chapter will discuss the impact that RYR1-related diseases have upon students' learning and their educational experience. Finally, this chapter will discuss the various types of school-related services available for individuals suffering with a RYR1-related disease.

RYR-1 Related Diseases

Ryanodine receptors (RYRs) are intracellular calcium release channels located in the sarcoplasmic reticulum (SR) within a muscle cell. The SR is an intracellular storage unit holding positively charged calcium ions (Ca^{2+}). Type 1 ryanodine receptor (RyR1) is the primary Ca^{2+} release channel on the SR. RyR1 facilitates the quick, precise, and coordinated release of Ca^{2+} from the SR to activate skeletal muscle contraction (Santulli et al., 2017). This serves a crucial

function for releasing calcium into the muscle cell, initiating a process called excitation-contraction coupling (ECC) that allows our muscles to contract and relax, permitting body movement (Jungbluth et al., 2016). To put it simply, the RyR1 channel serves as the doorway for the SR and Ca^{2+}, opening when the body moves, and closing when the body relaxes. Mutations in the RyR1 gene can contribute to a defective RyR1 channel, resulting in complications in muscle contractions and various neuromuscular disorders. These mutations can bring about constant calcium leak at rest, defective ECC, and increased mitochondrial oxidative stress, all impeding skeletal and smooth muscle contractions and body movement (Dowling et al., 2012).

Congenital Myopathies

Congenital myopathies are defined as a group of inherited, non-dystrophic neuromuscular disorders classified by their unique clinical features and skeletal muscle histopathology (Lawal et al., 2020). Typically, congenital myopathies are present and identified during childhood, considering individuals inherit such diseases. Congenital myopathies are almost always disorders consisting of a disrupted ECC (Lawal et al., 2018). Over the last decade, dominant and recessive RYR1 mutations have emerged as the most common cause of congenital myopathies. Various congenital myopathies with an RYR1 gene mutation include central core disease (CCD), centronuclear myopathy (CNM), multi-minicore disease (MmD), congenital fibre type disproportion (CFTD), and malignant hyperthermia susceptibility (MHS; Jungbluth et al., 2016). This group of disorders that are associated with RYR1 gene mutations resulting in a dysfunctional RyR1 channel is referred to as RYR1-related diseases.

Prevalence

RYR1-related diseases are rare and classified as orphan diseases. An orphan disease refers to the lack of marketability, funding, and resources available for the disease due to the

negligible portion of the population affected. An estimated 1 in 90,000 individuals in the US are reported to have a RYR1-related disease (Amburgey et al., 2011). It is important to note, however, that this estimate was based on a pediatric study. Considering this, the overall frequency in the general population is likely to be higher.

Range of Severity

Snoeck and colleagues (2015) investigated the full spectrum of RYR1 related disorders throughout life. In a cohort of 272 non-related patients with congenital myopathies, 77 patients were identified with one or more RYR1 mutations. In total, of the 77 patients identified with one or more RYR1 mutations and 61 different mutations were identified, thus, providing support of the wide spectrum of RYR1 mutations (Snoeck et al., 2015). The range in RYR1 mutations can lead to a wide variety of severity and symptoms. RYR1-related diseases have symptoms ranging from mild to severe muscle weakness, in which individuals may require a wheelchair or breathing support (Clinical Care Guidelines, 2019).

Developmental Course of RYR1-Related Diseases

Symptoms of RYR1-related diseases are often present from birth or appear in early infancy. Symptoms experienced by individuals with RYR1-related diseases are highly inconsistent. However, RYR1-related diseases are often non-progressive or very slowly progressing (National Organization for Rare Disorder, 2021). This indicates that the symptoms that individuals experience at early infancy are typically the same symptoms they will experience later in life. The lifespan for individuals affected by RYR1-related disease is generally average, despite that there is no cure or approved treatment (National Organization for Rare Diseases, 2021).

Genetics

The central implication of RYR1-related diseases lies in the core function of the RyR1 channel. This channel is responsible for releasing Ca^{2+} from the SR into the muscle cell when the muscles contract. Upon receiving electrical signals from the brain informing the muscles to move, the RyR1 channel will open, and release positively charged Ca^{2+} ions from the SR into the muscle cell. This increase of positively charged Ca^{2+} in the muscle cell stimulates muscle contractions, allowing the body to move. When the body is ready to relax, the RyR1 channel closes, stopping the flow of positively charged Ca^{2+} ions into the muscle cell. In response to the decrease of Ca^{2+} in the muscle cell, the muscle relaxes. With the RyR1 channel closed during rest, the SR fills with positively charged Ca^{2+} to be used when the RyR1 channel reopens, allowing the body to move again (Dirksen, 2018). Genetic mutations in the RYR1 gene result in dysfunctional RyR1 channels, leading to RYR1-related diseases.

Mutation can be defined as pathogenic changes in the DNA sequence of a gene that affect the function of that gene and its corresponding proteins (Clinical Care Guidelines, 2019). In RYR1-related diseases, the RyR1 receptor is the protein that corresponds with the RYR1 gene. The RYR1 gene is located on chromosome 19. This gene is responsible for properly encoding amino acids to proteins to build the RyR1 receptor; the RyR1 receptors are the building blocks for the RyR1 channel (Klein et al., 2012). RYR1 gene mutations can result in a protein that does not work properly and/or causes a reduction in the amount of RyR1 protein that is produced in the muscle cell (Clinical Care Guidelines, 2019). Recent research indicates there are more than 200 RYR1 gene mutations that can cause RYR1-related diseases (Jungbluth et al., 2016).

There are two types of genetic mutations that are most prevalent in RYR1-related diseases: nonsense and missense. Nonsense mutation is a mutation in which the DNA informs

the cell to stop making the RyR1 receptor before it is completed. This can result in a small RyR1 receptor or fewer RyR1 receptors produced in the cell, thereby resulting in an abnormal function of a RyR1 channel. Missense genetic mutation is a mutation in the DNA in which the wrong amino acid is being fused into the RyR1 receptor. This genetic error can result in an abnormal RyR1 channel (Clinical Care Guidelines, 2019).

Inheritance

RYR1-related diseases are inherited forms of myopathies, which means that individuals are born with these diseases and can inherit these mutations from one or both of their biological parents. Additionally, individuals can attain an RYR1 gene mutation spontaneously with neither biological parent having the mutation. The three types of inheritance for RYR1-related diseases are autosomal dominant, autosomal recessive, and de novo. Autosomal dominate inheritance indicates that one copy of a mutated RYR1 gene is passed down directly from one parent. In other words, one biological parent carries an RYR1 gene mutation and passes it down directly. If an individual with autosomal dominant RYR1-related disease conceives a child with an individual without a RYR1-related disease; there is a 50% chance that the child will have a RYR1-related disease.

Autosomal recessive inheritance indicates that both biological parents carry a mutated RYR1 gene, however, they typically do not show signs or symptoms of RYR1-related diseases. If two individuals carrying a mutated RYR1 gene, without showing signs of a condition, and conceive a child, there is a 25% chance that the child will have a RYR1-related disease. In addition, there is a 50% chance that the child will be a carrier of the mutated gene; meaning the child will have a mutated RYR1 gene but will not show signs or symptoms of a condition. Lastly, de novo inheritance refers to a brand-new mutation that is present for the first time in a

family, meaning both biological parents do not have a mutated RYR1 gene, however, their conceived child has a mutated RYR1 gene. Individuals who inherit a mutated RYR1 gene in this manner have a 50% chance of passing down a mutated RYR1 gene to their biological child, like autosomal dominant inheritance (Clinical Care Guidelines, 2019).

Different types of RYR1-related diseases are passed down through different types of inheritance. For instance, research reveals that autosomal dominant inheritance contributes to CCD, a more common RYR1- related disease, and MHS, while autosomal recessive inheritance contributes to MmD (Jungbluth et al., 2016; Klein et al., 2012; Zhou et al., 2007). Furthermore, different types of inheritance have been shown to contribute to a variety of symptomatology. In particular, individuals with autosomal dominant mutations typically experience symptoms in childhood and adulthood, while individuals with autosomal recessive mutations tend to first experience symptoms in infancy and childhood (Clinical Care Guidelines, 2019; Klein et al., 2012; Zhou et al., 2007). Finally, symptoms such as weak eye muscles, affected speech, swallowing, chewing, and more severe weakness are typical symptoms associated with an autosomal recessive mutation (Clinical Care Guidelines, 2019; Lawal et al., 2020; Todd et al., 2018; Klein et al., 2012; Zhou et al., 2007).

Diagnosis

Recent development in research have generated accurate genetic diagnosis of congenital myopathies including RYR1-related diseases. Unfortunately, given the fact that congenital myopathies have such low incident rates, they remain unfamiliar to a majority of care providers (Wang et al., 2012). Historically, congenital myopathies such as RYR1-related disorders were classified and diagnosed by histopathologic findings on muscle biopsy. In recent years, however, more RYR1-related disease phenotypes have emerged due to the implementation of next-

generation sequencing methods. Next-generation sequencing methods include the complete RYR1 gene coding sequence when compared to a histopathological approach, which just focuses on hotspot regions. These methods significantly enhance diagnostic capabilities (Lawal et al., 2018). Despite the recent advancement of diagnosis procedures, the different types of RYR1-related diseases can be difficult to distinguish from each other and other muscle diseases. Doctors may utilize lab tests, a muscle biopsy, an MRI or ultrasound, and genetic testing to diagnose RYR1-related diseases (Clinical Care Guidelines, 2019).

Characteristics of RYR1-Related Diseases

RYR1-related disease is referred to as a neuromuscular disease because it directly hinders a person's muscles (Clinical Care Guidelines, 2019). As previously discussed, current literature suggests RYR1-related diseases have a wide range of severity and thus exhibit a wide range of symptoms. Occasionally, different symptoms occur even among affected individuals within the same family (National Organization for Rare Diseases, 2021). General or common symptoms present in RYR1-related diseases are weakness of the eye muscles, weakness of the facial muscles, weakness of muscles closet to the torso (proximal muscle weakness), increased susceptibility to muscle cramping and pain, and decreased tolerance to exercise and heat (National Organization for Rare Diseases, 2021). Signs and symptoms are often present at birth or appear in early infancy. Various early infancy indicators include significant motor delays, poor eating, difficulties walking and climbing stairs, poor respiratory involvement, scoliosis, and kyphosis (a curvature of the spine or hunchback; Amburgey et al., 2013; Bharucha-Goebel et al., 2013). In addition, dynamic symptoms observed are heat intolerance, exercise induced muscle breakdown, muscle pain, and fatigue (Voermans et al., 2016). Considering that dynamic symptoms can occur in response to certain triggers, recent literature has explored types of

triggers that spark dynamic symptoms among individuals with a mutated RYR1 gene. Voermans and colleagues (2016) discovered exercise and heat as primary triggers, while viral infections, alcohol, and drugs were detected as less frequent triggers Research indicates that RYR1-related diseases can impact many different functions of the human body, including symptoms such as proximal muscle weakness, breathing complications including respiratory weakness, delayed motor milestones, bone and joint complications including joint contractures, Kyphoscoliosis to scoliosis (spinal deformities), ophthalmoplegia (weakness in eye muscles), facial weakness, and Malignant Hyperthermia (Lawal et al., 2020; Todd et al. 2018).

Skeletal Muscles

Typically, RYR1-related diseases predominantly affect skeletal muscles, which are the muscles attached to the bones that we use for voluntary movement. RYR1-related diseases may also inhibit smooth muscles, which are the muscles that humans do not voluntarily control (i.e., lungs, bladder, stomach, and esophagus). This weakness is largely due to the interaction of the RyR1 channel and its relationship with calcium. The dysfunctional relationship between the RyR1 channel and calcium results in weak muscle contractions. Largely, individuals with RYR1-related diseases often experience static muscle weakness, which means the muscle weakness they are experiencing is always present (Clinical Care Guidelines, 2019). RYR1-related diseases also have dynamic symptoms, which means they come and go in response to certain triggers (National Organization for Rare Diseases, 2021; Voermans et al., 2016).

Bones and Joints

Research supports that mutated RyR1 receptors are associated with skeletal disease in humans, as well as many other bone complications (Amador et al., 2013). Individuals suffering from a RYR1-related disease can experience orthopedic problems including overall muscle

weakness, contractures, hip dislocations, and spinal deformities (i.e., scoliosis; Clinical Care Guidelines, 2019). A contracture refers to a joint that does not have a full range of motion, specifically it is stuck in a flexed or bent position. Causes of contractures are issues with the joint capsule, muscles being replaced with stiff connective tissue over time, and the muscles being too weak or too short to allow for a full range of motion. Hip dislocations occur when the hip bone is out of its joint. Finally, individuals with a RYR1-related disease can suffer from spinal deformities such as scoliosis, Kyphosis, and Lordosis, all of which impair an individual's posture. It is important to note that a spinal deformity can impair an individual's ability to breathe deeply, thus, resulting in respiratory problems, as well (Clinical Care Guidelines, 2019). The medical literature reveals different common orthopedic features among different RYR1-related diseases. Individuals with CCD have been shown to have scoliosis, hip dislocations at birth, and mild contractures. Individuals with MmD tend to have progressive scoliosis. Furthermore, individuals with CNM commonly have scoliosis while those with CFTD commonly have hip dislocations, contractures, foot deformities, scoliosis, and lordosis (D'Amico & Bertini, 2008).

Breathing

It is evident that RYR1-related diseases have a negative impact on many functions of the human body. As previously examined, breathing may be negatively impacted by an RYR1 gene mutation resulting in a dysfunctional RYR1 receptor. All cells throughout the body need oxygen to function properly and make energy. When cells make energy, they produce carbon dioxide. Breathing is an essential component of this cell function, such that breathing brings oxygen into the body and releases carbon dioxide (Clinical Care Guidelines, 2019). Bearing in mind that dysfunctional RYR1 receptors contribute to weakness in muscle contraction, the diaphragm, the

primary breathing muscle, can be directly affected. During the breathing process, the diaphragm, located underneath the lungs, contracts, and moves downward while the chest expands. This allows air to be pulled into the lungs. Once oxygen is in the lungs, oxygen enters the blood and is carried to all internal organs and cells. While oxygen enters the blood, the carbon dioxide excreted from cells enters the lungs. When we exhale, the diaphragm relaxes, and the abdominal muscles assist in expelling the carbon dioxide out of the body through the mouth or nose. RYR1-related diseases can cause weakness in the diaphragm and abdominal muscles, rendering it difficult to clear the airways during breathing (Clinical Care Guidelines, 2019). Research suggests that individuals with CCD, MmD, CNM, and CFTD all experience respiratory difficulties, with MnD exhibited the most severe respiratory symptoms (D'Amico & Bertini, 2008).

Eating, Swallowing, and Speaking

Similar to a weakened diaphragm resulting in complications during breathing, a defective RyR1 receptor can negatively impact an individual's eating and speaking abilities. Unlike the breathing process there are about 26 different muscles that work together to allow people to chew, swallow, and articulate speech. These muscles are called the bulbar muscles and they are located in the face, mouth, and throat.

When eating and swallowing, the muscles in the mouth and jaw help you chew and break down the food after it enters the mouth. Once the food is adequately broken down, the muscles in your mouth and throat work in unison to push the food towards a long tube called the esophagus and away from the trachea, also known as the windpipe. A small but important muscle known as the soft palate flexes to block food from entering the nose and help direct the food down the esophagus. Additionally, another small muscle known as the epiglottis covers the trachea when

swallow to block any food from entering the lungs; like the soft palate, the epiglottis helps direct the food down the esophagus to eventually the stomach where the food can be digested. Unfortunately, among individuals with bulbar muscle weakness, the process of eating and swallowing may be impaired, such that, food or liquid could end up in the nose or trachea. Bulbar muscle weakness could also cause individuals to drool, take much longer to eat, and can lead to poor growth and difficulties gaining weight due to eating difficulties. Additionally, this could cause acid reflux (Clinical Care Guidelines, 2019)

Individuals also need bulbar muscles to speak. While speaking, people will utilize facial muscles such as the jaw, lips, and tongue to form words, while the diaphragm, lungs, and the larynx work together to produce sound. Like swallowing, these muscles work in unison to allow us to articulate speech. When these essential muscles are weakened due to a dysfunctional RyR1 receptor, individuals may suffer from a weak voice, causing communication difficulties. These individuals may also have problems controlling breathing, resulting in hypernasality, where too much air is leaking out of the nose during speech resulting in low quality speech. Others with a RYR1-related disease might suffer from slurred or slow speech also known as dysarthria. Some individuals may even develop structural abnormalities in the mouth. (Clinical Care Guidelines, 2019).

Medical professionals refer to weakness in the bulbar muscles as bulbar involvement; like the other characteristics of this disease, different types of inheritance contribute to different bulbar involvement features. Fascinatingly, research supports autosomal dominant forms of inheritance tend to be associated with more mild bulbar involvement, while autosomal recessive forms of inheritance tend to be associated with more severe symptoms (Clinical Care Guidelines, 2019; Jungbluth et al., 2018). Additionally, different types of RYR1-related diseases contribute

to different types of bulbar involvement; such that, bulbar involvement is typically only observed among individuals with CCD in severe cases, while it is common in MmD. Furthermore, structural abnormalities, such as a high arch in the roof of the mouth is seen in individuals with CNM and CFTD (Jungbluth et al., 2018).

Malignant Hyperthermia

Malignant Hyperthermia (MH) is a severe and dangerous reaction to certain anesthesia. This reaction causes your body to overheat to the point that your muscles break down. Additionally, this reaction causes muscle spasms, rapid heart rate, and rigid muscles (Mayo Clinic, 2020). If an individual with MH is not treated quickly, they can suffer from kidney failure, brain damage, cardiac arrest, additional organ failure, and possibly death. Individuals with a RYR1-related disease are known to have Malignant Hyperthermia Susceptibility (MHS), which indicates that they are more susceptible to MH due to a mutated gene. However, they are only at risk of MH if they are exposed to an anesthetic drug. Unfortunately, there are other symptoms of MHS in which individuals can experience muscle breakdown, severe muscle cramping, muscle stiffness, and heat intolerance when they engage in physical overexertion (Clinical Care Guidelines, 2019).

Potential Treatments

Despite the vast medical need, no approved treatments or therapies exist for RYR1-related diseases. Researchers are currently investigating various types of therapies and treatments; however, they are in the very early stages of clinical development (Lawal et al, 2018). Lawal and colleagues (2018) discuss the various drugs under development as potential therapeutics for RYR1-related diseases. N-Acetylysteine (NAC), Rycal, Sodium 4-phenylbutyrate (4BPA), 5-Aminoimidazole-4-carboxamide ribonucleoside (AICAR),

Salbutamol/ albuterol, Dantrolene, Carvediol, and Pyridostigmine are all current therapeutic compounds in early clinical development. However, NAC and Rycals are the most developed compounds with NAC having completed Phase I and II data collection while, Rycals have completed Phase I and are awaiting Phase II approval (Lawal et al., 2018).

NAC works to reduce oxidative stress. Oxidative stress is an imbalance of reactive oxygen species, also known as free radicals, and antioxidant defenses (Betteridge, 2000). Oxidative stress occurs in all of us; however, elevated oxidative stress is particularly problematic. Furthermore, it can be detrimental among individuals with a mutated RYR1 channel (Clinical Care Guidelines, 2019). Dowling and colleagues (2012) discovered that NAC significantly reduced oxidative stress among RYR1-related diseases in human myotubes (a developmental stage of a muscle fiber composed of a syncytium formed by fusion of myoblasts). The authors concluded that NAC is a successful treatment for individuals with a RYR1-related disease. Unfortunately, a recent study exploring this therapy yielded that NAC did not reduce oxidative stress and did not improve participants' endurance or fatigability (Todd et al., 2020).

Rycals are a new compound intended to stabilize the RyR1 receptor directly. This drug does this by improving the binding of the RyR1channel, thus repairing the calcium leak and improving muscle contractions (Clinical Care Guidelines, 2019). A recent study investigated Rycals among individuals with a RYR1-related disease. Results of this study revealed that Rycals can improve the binding of the RyR1 channel in muscle cells (Kushnir et al., 2020). This supports a positive first step to a potential therapeutic technique to treat individuals with a RYR1-related disease.

In addition to drug therapies, a consensus statement was released in 2012 that recommends regular exercise around two to three times per week for individuals with an RYR1

disease. Partaking in various athletic activities can improve strength, endurance, and maintain muscle function (Wang et al., 2012). Physical activity is extremely important for individuals with a RYR1-related disease, even among individuals with severe muscle impairment. Individuals with severe muscle impairment can remain active by using standing frames, assistance with walking, wheelchairs, arm supports, and avoiding bed rest for long periods of time (Clinical Care Guidelines, 2019).

Physical therapy is another technique for individuals suffering from a RYR1-related disease. Physical therapy can help prevent contractures, manage pain, avoid fatigue, and improve issues with cardiovascular and muscular endurance. Physical therapy can also improve mobility and slow the progression of a RYR1-related disease. However, it is important to note that there are currently no set standards of care for physical therapy for individuals with a RYR1-related disease (Clinical Care Guidelines, 2019).

RYR1-Related Diseases Impact on Student Learning and School Experience

The current literature confirms that RYR1-related diseases have no direct impact on a child's cognitive abilities and intelligence (Topaloglu, 2020); however, it is apparent that the physical symptoms are detrimental to a child's movement and life experience, especially when considering how they function within a school setting. For instance, a child with a RYR1-related disease with weak proximal muscles may have difficulties standing in lines for long periods of time and walking up and down stairs. Perhaps, if a child with a RYR1-related disease has difficulties eating, they may struggle to finish their lunch in the short, allotted time the school allows for children to eat. In addition, participation in physical education, field days, field trips, and recess will be negatively affected. Furthermore, children experiencing these symptoms will

not be able to participate in school athletics, inhibiting their involvement with extracurriculars and school community involvement.

Ruitenbeek et al. (2019) investigated the functional impairments, fatigue, mental health, and quality of life among individuals with RYR1-related diseases compared to a control group of healthy individuals. Patients with RYR1-related diseases had significantly more functional impairments when compared to the healthy control group; most notably patients with RYR1-related diseases had significantly more impaired sleep/rest, home management, alertness behavior, and social interaction than healthy individuals. In addition, individuals with a RYR1-related disease had significantly more chronic fatigue (i.e., perceived fatigue, concentration, motivation, activity) than healthy individuals. Furthermore, patients with a RYR1-related disease reported significantly higher levels of depression, anxiety, somatization, and obsessive-compulsive behavior when compared to the healthy controls. Lastly, patients with RYR1-related diseases had significantly worse physical functioning, social functioning, pain, and general health when compared to healthy individuals.

These results also challenge the notion that RYR1-related diseases are mild conditions considering the significant impact these diseases have on a person's life (Ruitenbeek et al., 2019). Although there is a lack of research on the impact of student learning with RYR1-related diseases, researchers have focused on students with other, muscular impairments such as Duchenne Muscular Dystrophy. Duchenne Muscular Dystrophy (DMD) is a progressive muscle weakness disorder that impacts a child's physical health similar to RYR1-related diseases (Lue et al, 2016). Lue et al. (2016) investigated the quality of life (QOL) in adolescents and young men with Duchenne Muscular Dystrophy. The authors utilized the health related QOL and the global QOL to assess the quality of life among participants, discovering that physical functioning and

social functioning domains among these individuals were significantly below average. These findings provide further evidence of the negative impact that muscular diseases can have on young children.

It is important to note that the many significantly elevated domains for individuals with RYR1-related diseases can have a direct impact on a student's learning and educational experience. Motivation, concentration, alertness behavior, social interaction, social functioning, depression, anxiety, and obsessive-compulsive behavior were all elevated among individuals with a RYR1-related disease. All elevated domains can negatively impact a child's learning. Despite RYR1-related diseases having no direct impact on intelligence, there is evidence that supports the existence of an indirect impact. For instance, a child with a RYR1-related disease experiencing high levels of fatigue and lack of alertness may have significant difficulty engaging in classroom material, thus impacting their learning. This research provides powerful evidence of the negative impact that these diseases can have on a child's learning and school experience.

Theoretical Framework

The biopsychosocial model has been a prominent framework for guiding clinical care. George Engel introduced the biopsychosocial model in 1977 and believed that to truly understand and respond appropriately to a patient suffering, a clinician must address the biological, psychological, and social dimensions of the disease or illness. In general, it is a way of understanding the patient's own experience as an essential contributor to an accurate diagnosis, healthy outcomes, and humane care (Borrell-Carrio et al., 2004). To put it simply, it is the connection between biology, psychology, and social environmental factors that can contribute to individuals' health. Unlike the biopsychosocial model, other theoretical models in psychology (i.e., biological, psychodynamic, behavioral, cognitive, and humanistic) focus on one

aspect of a person. Thus, not fully capturing the comprehensive complexity of a person's wellbeing. With the biopsychosocial model, we can address all facets of the human experience to truly conceptualize their experience. Additionally, this model, unlike the others, was introduced particular for serving a patient population experiencing distress (Borrell-Carrio et al., 2004).

In its application to RYR1 that would include an acknowledgement that RYR1-related diseases affect not only an individual's body (biology), but also their mental (psychology) and social life experiences, as well. The relationship between mental and physical health is complex. In order to properly and effectively treat individuals, clinicians must address the patients' holistic needs, connecting with the patient and assessing all levels of suffering that their disease brings (Borrell-Carrio et al., 2004). Considering RYR1-related diseases are inherited, individuals are born with their disease (Clinical Care Guidelines, 2019). As such, it is vital for early accurate diagnosis which will lead to early intervention services to help support these individuals. Treatment is not just medical in nature but also services to support development and functioning in school. Overall, the earlier the accurate diagnosis the earlier the proper service will be in place and the better the outcomes (Hebbeler et al., 2007). Furthermore, Kranzler and colleges (2020) discuss that school system-wide interventions are vital to support students and have a positive impact on student outcomes. The researchers go on to discuss that biopsychosocial models of human development that integrate the effects of genetics, personal characteristics and behaviors, environments, and broad social contexts are better and more informed frameworks for guiding future research in school psychology (Kranzler et al., 2020).

RYR1-Related Diseases in Schools

There is ample evidence of a growing literature base regarding the history, genetics, support, and treatments of and for RYR1-related disease. However, not much research has been

cunducted to explore the considerations of RYR1-related diseases for schools. Families and children often are unaware of the legal rights and protections they have supporting their quality of education, specifically to children with disabilities (Altshler & Kopels, 2003).

The Individuals with Disabilities Education Act (IDEA) is a federal law enacted to help find and protect children with disabilities. IDEA includes the "Child Find" mandate, which requires schools to find, identify, locate, and evaluate children with disabilities, no matter how severe the disability may be (Hughes, 2018). IDEA and the "Child Find" requirement protects all children with disabilities from birth to age 21. This requirement covers all children no matter where they attend school (i.e., public schools, private schools). In accordance with the 1999 revisions of IDEA, "Child Find" also does not discriminate against homeless children, migrant children (Altshler & Kopels, 2003), children who are wards of the state, those with disabilities, including those who are receiving passing grades (Clinical Care Guidelines, 2019; Hughes, 2018). School districts must identify all children with disabilities and provide appropriate support or special education and related services to eligible students identified (Lee, 2021). Once a child is identified as being at risk of having a disability in accordance with IDEA, a formal assessment will be completed. As specified by IDEA, either a parent of a child, a state educational agency, other state agency, or a local educational agency have the right to initiate a request for a comprehensive initial evaluation (Individuals with Disability Education Act, 2019).

This comprehensive initial evaluation is conducted by the school's multidisciplinary team and reviewed with the parents (Clinical Care Guidelines, 2019). A child is considered eligible for special education after the multidisciplinary team evaluates the child and determines that the child's school performance is adversely affected by the disability and the disability is categorized as an IDEA disability category (Lee, 2021). It is important to note that educational impact is not

defined as failing grades but rather interference in the ability to benefit from the educational environment (Individuals with Disabilities Education Act, 2017).

In the case of RYR1-related diseases, many school-related services can be useful to foster school success. For example, even before a formal psychoeducational evaluation, schools offer students support through Multi-Tiered System of Supports (MTSS) services, traditional parent, or teacher referral for support via Student Assistance Program (SAP) or other services as well as the use of screeners to help identify youth at risk even if they are not yet considered to need special education.

MTSS is a framework most schools use to provide targeted support and interventions to students in need. The goal of MTSS is to locate and intervene with struggling students so they do not fall behind and/or can catch up with their fellow peers (Rosen, 2022). Importantly, MTSS focuses of academic supports, while also addressing behavioral, social, and emotional needs of children. MTSS consists of 3 tiered levels comprised of many facets utilized to accomplish this goal. The levels become more intensive and individualized from one to the next. MTSS utilizes universal screeners for all students each year to detect struggling students, increasing levels of targeted support for those who are struggling, and individuals plans that address children's academic, behavioral, social, and emotional needs when applicable (Rosen, 2022). However, MTSS is not likely the best framework to support students with a RYR1-related disease due to the limited knowledge among school staff regarding the rare disorder.

SAP is a systematic team process used to utilize school resources to remove the barriers to learning. Such barriers addressed by SAP are issues including alcohol, tobacco, other drugs and mental health issues. SAP is designed to identify and address these barriers as they pose a threat to student success (Pennsylvania Department of Education, 2022). With regard to mental

health in youth, SAP teams typically identify referral sources to agencies for community-based support. This process often does not directly address support within the school (Swick & Powers, 2018).

Screeners are preferable because traditional practices have been shown to miss a third of students in need (Dowdy et al., 2013). There is limited information on the usefulness of screeners for use with students with a physical illness such as a RYR1-related disease. Information such as this might inform school supports such as a 504 Plan. It is an area in need of further research.

504 Plan

A 504 plan is one of the many services made available for students who require accommodations in the classroom based upon an eligible disability under the Americans with Disabilities Act. Specifically, 504 plans are guidelines for how schools can and will support a child with a disability (Lee, 2020). The 504 plan guides teachers and related school staff in supporting a student in the classroom (Clinical Care Guidelines, 2019). The goal of a 504 plan is to ensure that a child with a disability can fully participate in school to receive a Free and Appropriate Public Education (FAPE). Interestingly, since 504 plans are not under the umbrella of special education, there is no formal set of rules or guidelines for how they look and what they should include (Understood, 2020).

Typically, 504 plans can help a child with a disability by providing classroom accommodations in the general education classroom (Clinical Care Guidelines, 2019; Understood, 2020). Some examples of classroom accommodations that could be relevant for individuals with a RYR1-related disease are (but not limited to), extended time on test to help with fatigue and tired eyes, alternative seating options, and providing 2 sets of books; to prevent from having to carry a heavy backpack around school (Clinical Care Guidelines, 2019).

Due to there being no set rules mandating what a 504 plan looks like, the process of receiving a 504 plan differs from school to school. In most cases, parents and guardians may request a 504 plan through the school or the school will offer them directly to the child and family. A formal assessment is not required to receive a 504 plan. A school may provide classroom accommodations to any student who may need classroom supports to succeed in school (Understood, 2020). Considering the range in symptom severity among RYR1-related diseases, a 504 plan can be beneficial to support a child's academic success and educational experience.

IDEA Eligibility Categories

The Individuals with Disabilities Education Act requires public education agencies to provide special education and related services to students who need them. However, unlike a 504 plan, to receive formal special education through an educational agency, a student must qualify for special education (Lee, 2020). A comprehensive initial evaluation will determine if a struggling child qualifies for one of the 13 IDEA disability categories. The 13 IDEA disability categories are: Specific Learning Disability, Other Health Impairment, Autism Spectrum Disorder, Emotional Disturbance, Speech or Language Impairment, Visual Impairment (includes blindness), Deafness, Hearing Impairment, Deaf-blindness, Orthopedic Impairment, Intellectual Disability, Traumatic Brain Injury, and Multiple Disabilities (Individuals with Disability Education Act, 2019). After a comprehensive and holistic evaluation is conducted, and it is concluded that the child's school performance is adversely affected, they may qualify for special education and potentially receive services under the umbrella of one of these 13 disability categories. Children with a RYR1-related disease may qualify for Other Health Impairment and Orthopedic Impairment (Clinical Care Guidelines, 2019).

Other Health Impairment is defined as an individual who suffers from limited strength, vitality, or limited alertness with respect to their educational environment. Specifically, their educational performance is adversely affected by chronic or acute health problems such as (but not limited to) asthma, or attention deficit hyperactivity disorder (ADHD), diabetes, epilepsy, any heart condition, hemophilia, lead poisoning, leukemia, nephritis, rheumatic fever, sickle cell anemia, and Tourette syndrome (Individuals with Disability Education Act, 2017). RYR1-related diseases can be both chronic and acute health problems and therefore a child suffering from a RYR1-related disease could qualify under this disability category.

Orthopedic Impairment is often the most appropriate and applicable disability category for a child with a RYR1-related disease (Clinical Care Guidelines, 2019). Orthopedic Impairment is defined as a severe orthopedic impairment that adversely affects a child's educational performance. This includes impairments caused by a congenital anomaly, impairments caused by a disease (i.e., poliomyelitis, bone tuberculosis), and impairments from other causes (i.e., cerebral palsy, an amputation, and fractures or burns that cause contractures (Individuals with Disability Education Act, 2017). Notably, congenital myopathies, such as RYR1-related diseases, are both a congenital anomaly and an impairment caused by a disease. Moreover, RYR1-related diseases are impairments that cause contractures. If a child is suffering from a RYR1-related disease and it is deemed that their educational experience is being adversely affected, they will likely qualify for special education under an Orthopedic Impairment disability category, thus entitling this child to receive special education services to support their academic success.

IEP Related Services

Once a child is deemed eligible for special education, the next step the school must take to provide services is create an Individualized Education Program (Clinical Care Guidelines, 2019; Lee, 2020). An Individualized Education Program (IEP) is a legal document that verifies a child is receiving FAPE in the child's least restrictive environment (Clinical Care Guidelines, 2019). Similar to the comprehensive evaluation process, the IEP is developed by a multidisciplinary team to ensure the proper services are in place to address and support the child's needs. Each member of the IEP team plays an essential role in the development of the IEP (Lee, 2020).

The IEP team consists of the parent or legal guardian, a general education teacher, a special education teacher, a school district representative (i.e., principal, vice principal, special education director), an expert who can interpret the child's evaluation (i.e., school psychologist), any individual with quality knowledge or expertise needed to help the child (i.e., speech language pathologist, occupational therapist, school nurse, physical therapist) and depending on the age, the child (Clinical Care Guidelines, 2019; Lee, 2020). Each member plays a vital role in identifying the needs and appropriate services needed to support the child's educational experience. It is imperative that the IEP meets the child's individual needs. Also, once an IEP meeting occurs and an IEP is being implemented, IDEA requires the IEP team to meet once a year to discuss the IEP and make corrections or additions if necessary (Individuals with Disability Education Act, 2017).

One significant component of the IEP are related services, which include the non-academic areas in which the student requires assistance, which include such supports as counseling, parenting collaboration, and transportation. The related services included in the IEP should directly address the child's individual needs to help them reach their educational goals

(Clinical Care Guidelines, 2019). Some direct and related services that may be used to help students with a RYR1-related disease are adaptive physical education, speech-language pathology, occupational therapy, physical therapy, counseling services, parental supports.

Adaptive Physical Education is physical education that has been adapted or modified so that it is appropriate for an individual with a disability (Adapted Physical Education National Standards, 2008). IDEA states that physical education must be provided to students with disabilities if the educational agency provides physical education. IDEA defines physical education as the development of physical and motor skills, fundamental motor skills (i.e., throwing, catching, walking, running), and skills in aquatics, dance, and individual or team games/sports (Individuals with Disability Education Act, 2017). The Adaptive Physical Education teacher is a direct service provider because physical education for students with disabilities is a federal mandate (Adapted Physical Education National Standards, 2008).

Adaptive physical education is essential for promoting exercise and physical activity among children with a RYR1-related disease in schools. As mentioned previously, exercise and physical activity are vital for managing symptoms of RYR1-related diseases (Wang et al., 2012; Clinical Care Guidelines, 2019). The adaptive physical education teacher can provide various services such as planning, assessment of individuals and ecosystems, placement in the IEP, teaching, counseling, or coaching, evaluation services, coordination of resources and consulting, and advocacy (Adapted Physical Education National Standards, 2008). Considering the importance of physical activity for managing and preventing symptoms of RYR1-related diseases, it is imperative that students with a RYR1-related disease have opportunities to participate in physical education. Moreover, adaptive physical education should be seriously considered when considering IEP services.

Speech-language pathology services and occupational therapy services are also related services that can be included in an IEP for a student with a RYR1-related disease, considering that RYR1-related diseases can negatively impact the bulbar muscles, which are muscles in the face, mouth, and throat. Speaking and communicating can be an area of weakness among these individuals. In addition, eating and swallowing may also be adversely impacted. A speech-language pathologist or an occupational therapist are specialists that works in schools who can assess the speaking/communication and swallowing difficulties among children with a RYR1-related disease. A speech-language pathologist can help a child learn to control breathing and speak more clearly. In addition, the speech-language pathologist may teach sign language, gestures, writing, or how to use a communication tool/device to help communication skills. An occupational therapist can properly assess the swallowing and eating challenges that a child with a RYR1-related disease may experience. Occupational therapists can help determine the risk of eating certain foods and inform the family and school which foods are safe for the child to eat (Clinical Care Guidelines, 2019).

Wang et al. (2012) present a standard of care for congenital myopathies such as RYR1-related diseases. The authors discuss the promotion of independence and maximizing function through the promotion of physical activity and avoidance of inactivity, management of pain, and access to appropriate specialized orthopedic management when necessary. They encourage exercise, standing, maintaining joint range of motion, scoliosis management, and the proper use of mobility aids. These important guidelines for care can be addressed and supported by physical therapy. Physical therapy is a vital part of managing RYR1-related diseases. Working with a physical therapist can help prevent further contractures, mange pain and fatigue, improve

mobility, increase endurance, slow the progression of the disease, and help use assisted mobility aids correctly (Clinical Care Guidelines, 2019).

Ruitenbeek and colleges (2019) revealed individuals with a RYR1-related disease to report significantly more mental health problems (i.e., anxiety, depression, obsessive compulsive behaviors, social difficulties) when compared to the general healthy population. Addressing the mental health of children with a RYR1-related disease is important to consider when addressing the students' needs in the IEP. Indeed, the multidisciplinary evaluation process requires the consideration of the impact of a disability on the whole of a child's functioning. That is, information regarding the child's mental health needs should not be neglected in the evaluation of a child with RYR-1. If it is deemed necessary, counseling services or small social/emotional groups with a school counselor, social worker, or school psychologist may be considered.

Parents and legal guardians play a critical role in the IEP team. The parent or legal guardian is a partner in the education of a child, able to provide valuable knowledge about the child's strengths, weaknesses, and goals (Lee, 2020). The parent or legal guardian of a child with a RYR1-related disease knows the most about their child and how their disease is affecting them. Parents or guardians serve as an advocate for a child and are responsible for their child's wellbeing (Clinical Care Guidelines, 2019). Therefore, it is essential that parental supports be considered when developing an IEP for a child with a RYR1-related disease. Connecting the parents with resources and the community can beneficial to not only the parent but also the child. Schools should consider providing families with resources such as the Clinical Care Guidelines developed by the RYR-1 Foundation. The Clinical Care Guidelines are included in a manual and are a resource for patients and families to help understand RYR1-related diseases (Clinical Care Guidelines, 2019).

Summary

A significant amount of research supports that RYR1-related diseases have numerous negative impacts on individuals' biology and body function (Amburgey et al., 2013; Bharucha-Goebel et al., 2013; Clinical Care Guidelines, 2019; Klein et al., 2012; Lawal et al., 2020; Todd et al., 2018; Voermans et al., 2016; Zhou et al., 2007). Most literature addressing RYR1-related diseases focuses on the mechanisms, diagnosis, pharmaceutical treatments, and the biological features of the disease. However, researchers also suggest that RYR1-related diseases can negatively impact an individual's mental health and social environment (Ruitenbeek et al., 2019; Lue et al., 2016). In order to significantly generate valuable literature and promote the betterment of individuals suffering from RYR1-related diseases, psychologists and related healthcare providers must take a biopsychosocial approach to this disease and focus research on its psychology and social impact. Through this research project, my aim is to address the psychological and social needs of children with RYR1-related diseases. Specifically, I hope to generate useful information to help children and families suffering with RYR1-related diseases to fully utilize all resources available for them in schools; and thus, prosper in their educational environment.

CHAPTER III

METHODS

This chapter describes the methodology and proposed analyses used to assess the classroom experiences of individuals with a RYR1-related disease and the types of services they receive in school. As a means to identify the research questions identified in Chapter I, I will describe the characteristics of the participants, the research design, the procedures, and the proposed statistical analyses. Additionally, I will provide a rationale of the instruments selected in order to answer the research questions.

Methods

Participants

Participants in this study were parents or caregivers of children with a diagnosis of a RYR1-related disease enrolled in a public, parochial, or private school in the US. Participants in this study will be provided information about the study and invited to participate in the research through the RYR-1 Foundation's communications with their families (i.e., listserv and social media). Individuals were selected to participate for this study if they met the specific inclusion criteria, including being the primary caregiver for one or more children formally diagnosed with a RYR1-related disease who are attending a PreK-12 public or private school within the US.

Instruments

Data for this study will be gathered using two instruments, a newly-developed RYR1-Related Disease Educational Impact and Satisfaction Survey (RDEISS) which was adapted from other measures considering the impact of illnesses in school settings and the Behavioral and Emotional Screening System (BESS). For the purposes of this study, the RDEISS was

developed to gather demographic data, information specific to the child diagnosed with a RYRI-related disease and assess overall caregiver satisfaction with school services provided to the child. In addition, the BESS was utilized to assess the child's behavioral and emotional status. Accordingly, each instrument was utilized to answer the research questions previously discussed. A description of the survey and rating scale are provided below.

RYR1-Related Disease Educational Impact/Satisfaction Survey (RDEISS)

In this study, I developed a survey to gather data to help answer the research questions posed. The RDEISS was delivered through Qualtrics, a simple, well-established, and flexible survey platform. This survey platform may be used to send surveys to a desired audience on any channel, including email (Qualtrics, 2022). For this reason, it was a useful survey platform to distribute the measures for this study. The RDEISS has three sections: Demographics, Educational Information, and School Services Satisfaction.

The Demographics section was designed to gather valuable information of the parents/caregivers completing the survey and the children their children they are reporting on. This section of the survey was adapted from The National Association of Independent Schools (NAIS) Student Demographic Questionnaire (child demographics) and Bitterman and colleagues (2008) article (parent demographics). The National Association of Independent Schools (NAIS) Student Demographic Questionnaire was designed to be completed by parents and caregivers to assist schools in collecting accurate demographic information about their student populace. This instrument uses generic language, so it can be completed by any parent or caregiver. In addition to the NAIS Student Demographic Questionnaire, the Demographic section of this survey also adapted certain elements of the Survey Questions section of a dissertation published by Miglioretti (2019).

Similar to the goals of this study, the Miglioretti (2019) survey provided a tool to gather information regarding the classroom experiences of children with Pediatric Acute-Onset Neuropsychiatric Syndrome (PANS). This disorder affects school-age children, causing cognitive, social, emotional, and academic difficulties. Miglioretti's (2019) survey was designed to be completed by caregivers/parents to gather relevant information regarding the classroom experiences of their children. The Survey Questions section of Miglioretti's measure includes relevant questions related to the disorder of the child. Therefore, it is an appropriate instrument to use to gather relevant information to RYR1-related disease. Overall, both the NAIS Student Demographic Questionnaire and Miglioretti (2019) Survey Questions section were adapted and combined to form the Demographics section of the survey used in this study. Questions in the Demographic section of the survey included those such as the age of child, the gender of child, in which grade the child is currently enrolled, the type of school the child is attending (private, public), the age at which the child first experienced RYR1-related disease symptoms, age at diagnosis, and primary symptoms.

The Educational Information section of the survey used in this study was also adapted from the Miglioretti (2019) survey. In this section, I will focus on gathering information regarding the difficulties the child experiences in school and if they have been receiving any school-related services. In this section, the types of services are identified. Questions in the Educational Information section of this survey include the types of difficulties that the child is experiencing in the classroom, whether the child is receiving homebound instruction, if the child has ever been withdrawn from school due to their RYR1-related disease, if the child has been referred for an evaluation, what was the result of the evaluation, and what services is the child receiving. At the end of the Educational Information section, the parent/caregiver is

asked if their child is receiving any individualized school services, such as an IEP or a 504 plan. If the parent/caregiver responds yes and indicates that their child is receiving services or accommodations documented in an IEP or 504 plan, they are directed to the next section of the survey.

In part three of the survey, questions are organized into a theme of a School Services Satisfaction section. If the parent/guardian indicates that their child is not receiving any services, then they will be redirected to the next section of the survey. The School Services Satisfaction section of the survey used in this study was adapted from the IEP Satisfaction Survey developed by Slade and colleagues (2018).

Slade et al. (2018) developed a three-question IEP satisfaction survey to assess a parent's satisfaction with the IEP plan and services being provided to their child. Therefore, this survey was appropriate to assess the satisfaction of school services being provided to children with a RYR1-related disease. For the purposes of this study, Slade et al.'s (2018) IEP satisfaction survey was adapted to address all school services, including an IEP and a 504 plan. Questions in the School Services Satisfaction section of the survey include: (1) the satisfaction of the parent/caregiver with the school services plan, (2) the degree to which the parent perceives the school service plan being implemented as it is written, and (3) the satisfaction of the parent/caregiver with the services being implemented. A 4-point Likert scale ranging from not at all satisfied to very satisfied was utilized for Items 1 and 3, while a 4-point Likert scale ranging from no agreement to very good agreement was utilized for Item 2 (Slade et al., 2018).

Slade and colleagues (2018) investigated the satisfaction of parents of young children with Autism Spectrum Disorder (ASD) regarding their children's IEPs. The researchers

conducted a confirmatory factor analysis (CFA), using four factors, to examine the measurement model for the factors of their study. The first factor included was the IEP satisfaction composite (which was measured by the IEP satisfaction survey that was adapted and utilized for the current study) comprising of all IEP satisfaction items. The second factor used in the researchers' CFA was the child's developmental functioning, comprising the child's ASD symptoms, measured by the Autism Diagnostic Observation Schedule (ADOS) algorithm score. Additionally, this factor was comprised of two language variables measured by the Comprehensive Assessment of Spoken Language (CASL) sum of standard scores and the Child's Communication Checklist (CCC-2) global communication composite. Furthermore, this factor was comprised of intellectual functioning measured by the Wechsler Preschool and Primary Scale of Intelligence (WPPSI-IV) estimated full-scale IQ.

The third factor in the CFA was parent–school connectedness, comprising items assessing parent and teacher relationship quality (reported by both parent and teacher), teacher-reported parental school involvement, and parent-reported parental school involvement. Finally, the fourth variable used in the CFA was a family financial factor, including the family income at enrollment, family income at IEP assessment, and family financial hardship.

The confirmatory factor analysis based upon the four factors and their measures produced good model fit (CFI = 0.99). Standardized factor loadings for the IEP satisfaction factor ranged from 0.61 – 0.93, which was significant at $p \leq .001$ (Slade et al., 2018). This indicates that the IEP satisfaction survey used in Slade et al. (2018) was a valid measure for assessing the factor IEP satisfaction composite. In other words, such measurement offers evidence that the IEP satisfaction survey is a valid measure for assessing a

parent's/caregiver's satisfaction with their child's IEP; and therefore, is appropriate to use for this study.

Behavioral and Emotional Screening System (BESS)

The BESS, developed by Randy Kamphaus & Cecil Reynolds (2015) is an instrument designed to assist school personnel, such as school psychologists and other care providers, in determining students' behavioral and emotional strengths and weaknesses. This screener targets students between the ages of 3 and 18 years of age, and has 25-30 items, depending on the form used, that contribute to four behavioral and emotional functioning domains in children and adolescents. The four domains of the BESS are Externalizing Risk, Internalizing Risk, Adaptive Skills Risk, and Behavioral and Emotional Risk Index (Kamphaus, R. & Reynolds C., 2015). There is a parent, teacher, and student form available for use (Evidence Based Intervention Network, 2014).

For the purposes of this study, I utilized the parent form. The parent form is designed to be completed by parents and consists of 29 items (Kamphaus, R. & Reynolds C., 2015). I selected the BESS parent form due to its feasibility, quick response time, and strong reliability and validity. In addition to interpreting the scores on each domain, a total score can also be used to gather valuable information regarding the child's overall behavioral and emotional status. One can then convert the raw total score to a standard score that allows the responses to be easily compared to the whole normed population (Evidence Based Intervention Network, 2014).

There has been an ample literature base examining the reliability and validity of the BESS on students' behavioral, emotional, and academic status. Most literature has provided a review of the teacher form on elementary and middle school students (Evidence Based

Intervention Network, 2014). However, Dowdy and colleagues (2013) examined the parent form of the BESS and its criterion-related validity of score inferences to detect symptoms of prevalent mental health disorders in childhood (Dowdy et al., 2013). In other words, the authors investigated if the parent form of the BESS could effectively identify symptoms of mental health disorders in children. Dowdy and colleagues (2013) gave 99 parents of first-fifth grade students the BESS parent form. In addition, they gave all parents the Child Behavior Checklist (CBCL). The authors computed correlations based on the CBCL scales oriented to the Diagnostic and Statistical Manual (DSM). They discovered that the correlations between the BESS (parent form) total score and the CBCL DSM-oriented scales (conduct problems, oppositional defiant, attention-deficit/hyperactivity disorder, and affective problems) yielded moderate to high correlations. In addition, correlations between the BESS (parent form) and the CBCL externalizing, internalizing, and total problems composite scores also yielded moderate to high correlations (Dowdy et al., 2013). These results support that the BESS is a reliable measure for identifying children's at-risk behavior and emotional weaknesses. This research supports that high scores on the BESS may indicate the presence of symptoms of mental health disorders (Dowdy et al., 2013). Furthermore, these results are consistent with research conducted by Kamphaus, R. & Reynolds C. (2007) in which the BESS yielded strong reliability and validity. In addition, Kamphaus and Reynolds (2007) provided evidence indicating the BESS has high positive correlations with general measures of behavior and emotional problems, ADHD, and executive functioning. Considering this research, the BESS is a reliable and valid measure for assessing children's behavioral and emotional status and, therefore, is an appropriate measure for the current study.

Research Design

For this study, I will use the survey method for collecting data. This research method generates quantifiable information about individual characteristics of members of a target population. Through the current research design, I will survey parents/caregivers for the purpose of gathering information about children with a RYR1-related disease. Survey research designs allow researchers to generalize information about a population by drawing upon data gathered from a small portion of that same target population (Rea & Parker 2014). The expected number of participants to complete surveys for data collection was approximately 50.

Variables

All survey items were presented in a multiple-choice format; some survey responses prompted respondents to provide additional information in free text. Additionally, the School Services Satisfaction section and the BESS utilized a 4-point Likert scale for each question. See Appendix A to review the survey developed for this study.

Throughout this study there are four variables of interest. The primary variables of interest for this study are common problems in the classroom, types of school services being provided, the satisfaction of school services being provided, and the behavioral and emotional status of children with a RYR1-related disease. The common problems in the classroom variable is assessed through the Educational Information section of the survey in which the parent/caregiver is asked to indicate all areas of difficulties with which their child is experiencing in the classroom from a comprehensive list. This will provide valuable information for the types of struggles children with a RYR1-related disease are experiencing in the classroom.

In order to ascertain which types of services are being provided, there are two questions

in the Educational Information section of the survey. In the first question, parents/caregivers are asked if their child is receiving services at school. If yes, in the second question, the parent/caregiver is asked what types of services the child is receiving. Through this question, information will be provided regarding the types of services children with a RYR1-related disease are receiving. Additionally, parents/caregivers' satisfaction of school services being provided is assessed using the School Services Satisfaction section of the survey. Through these questions, parents/caregivers who have indicated that their child is receiving school services are asked if they are satisfied with the services. With a 4-point Likert scale, the lower ratings indicate less satisfaction and agreement while higher ratings will indicate more satisfaction and agreement. For those who indicate low satisfaction with school services; an open-ended question is provided asking the caregiver to briefly explain why they have low satisfaction with their child's school services.

Finally, the behavioral and emotional status of children with a RYR1-related disease is assessed using the BESS. The BESS consists of 29 questions, which require parents to rate their child's experiences on a 4-point Likert scale. Responses on items will load onto four different domains of behavioral and emotional functioning (Externalizing Risk, Internalizing Risk, Adaptive Skills Risk, and Behavioral and Emotional Risk). Elevated scores on these domains will provide valuable information on the behavioral and emotional status of children with a RYR1-related disease. It is important to note that the BESS parent form is assessing parents' perceptions of their child's behavior; as this is a valid measure for evaluating the behavioral and emotional status of children, it is likely that this measure is tapping behaviors most reflected in the home setting. However, despite this, considering its validity and relevant information obtained, this measure is still relevant in understanding the behavioral and emotional

functioning of children with a RYR1-related disease.

Procedures

An introductory statement about the purposes of the research and informed consent about research participation was provided before participants completed the RDEISS. Consent was considered to have been granted if participants chose to proceed to complete the survey questionnaire. Initial questions included screening criteria for inclusion, which was described previously. If participants met inclusion criteria, they would continue to complete the survey sections in the following order: Demographics, Educational Information, School Services Satisfaction, and BESS.

The RDEISS was delivered through Qualtrics, a well-established online research tool for administering surveys. The Qualtrics program is convenient and easy to use for both participant and researcher. It allows researchers to have participants respond to surveys remotely and stores the participants' responses in a way that ensures participant confidentiality. The survey was distributed to parents and caregivers of children with a RYR1-related disease with assistance by the RYR1 Foundation, who have access to many families with a RYR1-related disease diagnosis. Once the appropriate number of surveys were completed, and the data collection process was finished I analyzed the data to answer the research questions.

Data Analysis

After the data collection, the data was cleaned and coded. Data cleaning included the removal of data from participants who did not meet the criteria for participation in the survey previously outlined. Additionally, individuals who provided incomplete survey responses were

removed. The methods of analysis in this research included simple descriptive statistics and frequency counts from survey responses.

Research Questions

Through the current research project, I sought to answer the following research questions based on the parent/caregiver responses on the RYR1-related disease educational impact/satisfaction survey. An analysis for each research question and rationale for how they will be answered are documented in more detail below.

Research Question 1. What are the common classroom problems experienced by children with a RYR1-related disease?

This question will be answered using the Educational Information section of the RDEISS. Question 27 asks parents/caregivers to identify all difficulties that their child experiences at school. The parents/guardians are prompted to indicate all that apply from a comprehensive list of school problems. Descriptive statistics will provide the appropriate analyses to assess the types of school difficulties parents are indicating their child with a RYR1-related disease is experiencing.

Research Question 2. With what frequency do children with RYR1-related diseases receive special services in schools (i.e., formal special education (IEP), Section 504 Plan, informal services, no services at all)?

This question will be addressed in the educational information section of the RDEISS. Through Question 28 and 29, parents/caregivers are asked to indicate whether their child has been evaluated to determine eligibility for a 504 plan or formal special education services (IEP). Response options are “Yes” and “No”. If the parent/caregiver responds yes, question # asks the parent/caregiver to indicate the result of the evaluation. Response options are “my child is

eligible for special education and received an IEP", "My child is eligible for accommodations and received a 504 plan", or "The school found my child was not eligible for an IEP or 504 plan (no plan in place)." Descriptive Statistics of these set of questions will be calculated to provide the appropriate analysis to answer this research question.

Research Question 3. If a child does receive special education services, under what IDEA category(ies) does the child qualify?

This question will be answered if the parent/caregiver responds to question 30 indicating that their child has received an IEP. Question 30 asks the parent to select the IDEA disability category/categories the multidisciplinary team identified. The response options are the 13 IDEA disability categories (Autism, Specific Learning Disability, Blind, Visually Impaired, Deaf, Hearing Impaired, Other Health Impairment, Emotional Disturbance, Multiple Disabilities, Traumatic Brain Injury, Speech/Language Impairment, Intellectual Disability, Orthopedic Impairment) and an option if the parent/caregiver doesn't know (Don't know/aren't sure). Descriptive Statistics of these set of questions will be calculated to provide the appropriate analysis to answer this research question.

Research Question 4. For children who do receive special services (i.e., IEP or Section 504 Plan) at school, what services do they receive and what is the parental satisfaction with those services?

This question will be answered using the educational information and school service satisfaction sections of the RDEISS. Question 31 (part one and two) will ask the parents/caregivers to indicate what services are provided for their child as part of their IEP or 504 plan? They will be presented an abundant list of school services to select from. Descriptive statistics will be an adequate analysis of responses to fully grasp the types of school services in

place for students with a RYR1-related disease. Questions 34 will ask the parents/caregivers who indicated their child is receiving an IEP/504 plan "overall, how satisfied are you with the services currently outlined in your child's IEP or 504 plan?" In Question 35, parents/caregivers will be asked, "how much agreement is there, in your opinion, between the content of your child's IEP/504 plan document and services actually being provided?" Question 36 asks parents/caregivers "in practice, how satisfied are you with the quality of the services actually being provided to your child?" Each question has a 4-point Likert scale for scoring, with higher scores indicating more satisfaction and lower scores indicating lower satisfaction. Considering this measure has been previously found as a valid measure for assessing IEP satisfaction among parents/caregivers, it seems to be an appropriate instrument for this study (Slade et al., 2018). The responses and scores these questions yield will be an adequate measure to capture parents' IEP satisfaction. Descriptive statistics will be an adequate analysis of responses to the survey items. Moreover, this form of statistical analysis will be effective at answering this research question. Finally, question 37 asks parents "if you have low satisfaction (Not at all Satisfied or Not Satisfied) with this child's school services, briefly explain why below." This question allows participants to briefly explain why they might not be satisfied with the school services being provided. Thematic coding procedures will be utilized to analysis participants responses.

Research Question 5. Do children with a RYR1-related disease experience behavioral and emotional problems as reported by parent/caregivers on the Behavioral and Emotional Screening System (BESS)?

This question will be answered using the BESS. The BESS can be easily converted to the Qualtrics survey platform and can easily be added to the RYR1-related disease educational impact/satisfaction survey. Upon completion of the RYR1-related disease educational

impact/satisfaction survey, parents will be directed to answer the 29 items on the BESS parent form. Responses to these items will load to the four domains, Externalizing Risk, Internalizing Risk, Adaptive skills Risk, and Behavioral and Emotional Risk Index. There are four rating options for each item (never, sometimes, often, or almost always). Since this screener is normed on a whole population, the sum of raw scores on all items can be converted to a T-score. A T-score from 20-60 suggests a normal or average level of risk, 61-70 suggests elevated level risk, and 71 and higher suggests an extremely elevated level of risk (Dowdy et al., 2013). For the purposes of this study T-scores will be used to assess the behavioral, emotional, and academic status of individuals suffering from a RYR1-related disease.

Summary

Through this chapter, I have presented all the necessary components to execute the proposed study. In the current study, I will utilize a survey method for collecting relevant data to answer the research questions. This study will employ descriptive statistic data analysis on data obtained from the RDEISS collected from parents/caregivers of children with a RYR1-related disease. The completed surveys will be used to analyze the variables of common problems in the classroom, types of services being provided, and satisfaction of services being provided to better understand the educational experience of children with a RYR1-related disease. The school satisfaction section of the RYR1-Related Disease Educational Impact and Satisfaction Survey is adapted from the IEP satisfaction survey presented by Slade et al. (2018). This survey has been thoroughly investigated and has been shown to have strong psychometric properties, thus indicating that it is an appropriate measure to investigate the satisfaction of school services being provided. Furthermore, The BESS parent form will be utilized to measure the behavioral and emotional status of children with a RYR1-related disease. This is a well validated measure and

appropriate for identifying at-risk behavioral and emotional symptoms in children (Dowdy et al., 2013). In the next chapter, the results of the analyses will be examined to answer the proposed research questions.

CHAPTER IV

RESULTS

This chapter describes the results of the RDEISS survey used to assess the classroom experiences of individuals with a RYR1-related disease. Specifically, the demographic data of the children and their parents will be reported. Furthermore, the survey questions that directly correspond to the research questions identified in Chapters 1 and 3 will be presented.

Results

Sample

Parents of this study include the primary parent of an individual with a RYR1-related disease between the ages of 3-18. A total of 61 parents began the REDISS survey. Of the 61 parents who began the survey, four indicated that they did not wish to complete the survey. Additionally, three parents did not meet the inclusionary criteria outlined in Chapter 3 (i.e., primary parent of a child currently in grades k-12 and the child is between the ages of 3-18, diagnosed with a RYR1-related disease). Furthermore, five parents left the survey immediately after completing the consent form. This means 12 parents of the total 61 were eliminated and did not have responses to be analyzed. A total of 49 parents answered survey questions; however, seven of those parents withdrew from the survey at various points of the survey. Overall, 42 parents completed the survey in its entirety. With consideration of the rarity of this disease and the limited number of parents, it was decided to interpret the results of the 49 parents who completed some or all of the survey. This allows for more data to be included in the current research study of an otherwise under-researched population. Thus, the proceeding results are indicative of 49 parents who are the parent of an individual diagnosed with a RYR1-related disease between the ages of 3 and 18.

Moreover, of the 49 parents, three parents indicated that they have a second child diagnosed with a RYR1-related disease between the ages of 3 and 18 years old. Therefore, this survey is indicative of 49 parents reporting on 52 total children and adolescents with a RYR1-related disease. Notably, the parents who reported on 2 children finished the survey in its entirety.

Importantly, as this research utilizes descriptive statistics and frequency counts, the total number of responses reported will be no less than 42 or 45 (the 42 parents who finished the survey reporting on 45 children/adolescents) and no more than 49 or 52 (the 49 parents, including the 7 who didn't finish the survey, reporting on 52 children/adolescents). Results are described and depicted in detail below.

Parent Demographics

There was a total of 45 parents of individuals with a RYR1-related disease who were reported in the "Parent Demographics" section of the survey. 41 of the parents were female (91%) and 4 were male (9%). Additionally, 39 were white/Caucasian (87%), 2 were Hispanic, Latinx, or Spanish origin (4%), and 4 were either Asian (2%), Black/African American (2%), Middle Eastern (2%), or Native American or Alaska Native (2%). The majority of parents' household income was more than $40,001 (91%) and most parents' highest education level was some college attended (31%) or four-year degree or higher (62%). Furthermore, 84% of parents indicate there is another parent in the household.

Child Demographics

There was a total of 51 children/adolescents diagnosed with a RYR1-related disease that was reported on behalf of their parents. 4 parents indicated that they have 2 children diagnosed with a RYR1-related disease. The ages of the children varied, ranging from age 3 to 18 with at

least one child represented in each age. With respect to the gender identity of the children, 43% (N=22) identified as female, 55% (N=28) identified as male, and 2% (N=1) identified as non-binary. Similar to parent race, most children were white/Caucasian (80%; N=41) with 6% (N=3) Black/African American and Hispanic/Latinx, or Spanish origin, respectively, and 3% (N=2) being Middle Eastern, and 2% (N=1) being Native American or Alaska Native, respectively.

Moreover, 40 children (78%) attend school in the United States while 11 children (22%) attend school outside of the United States. Children's current grade level ranged from Pre-K to 12th grade with students represented in each grade except the 11th grade. A majority of children (76%; N=39) attend public schools and 10% (N=5) attend private/parochial schools. Lastly, 92% (N=47) have not been retained or required to repeat a grade.

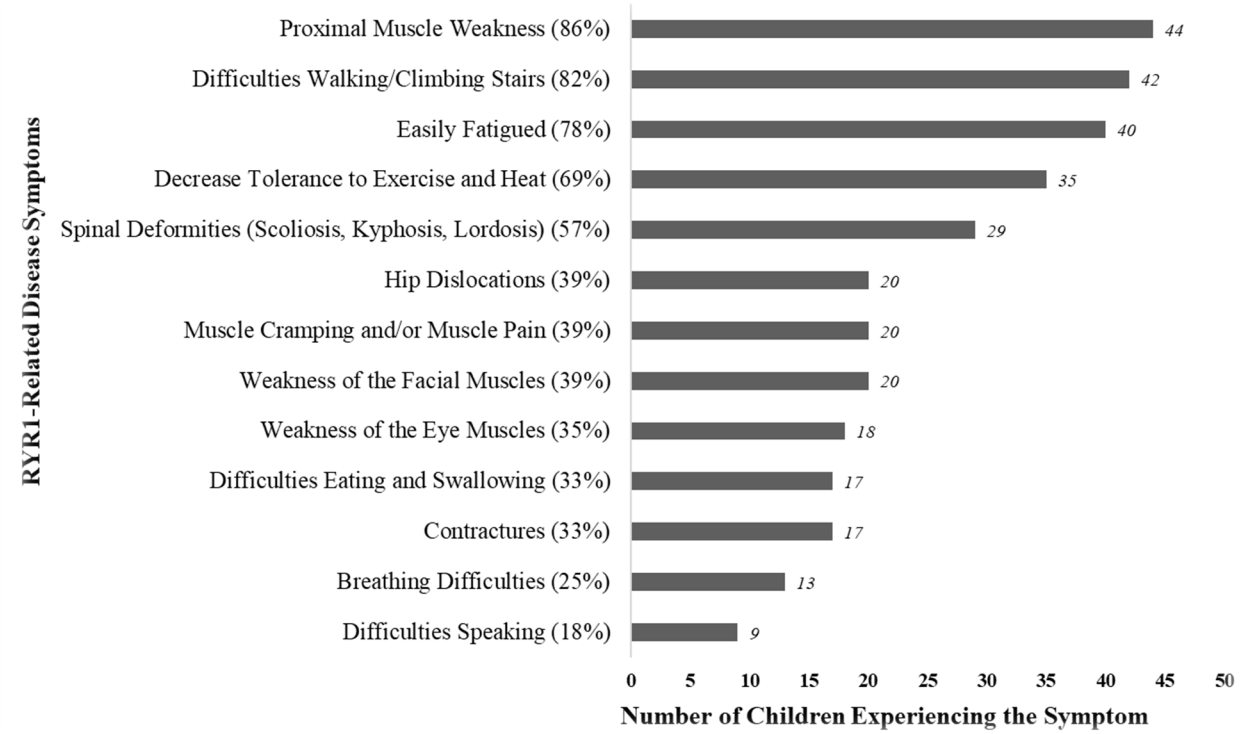
RYR1 Related Demographics

Over 98% of parents indicated that their children first exhibited RYR1-related disease symptoms before the age of 3 (N=50). Interestingly, only 49% of children were diagnosed with a RYR1-related disease before the age of 3 (N=25). Around 23% of children were diagnosed with a RYR1-related disease between the ages of 3 and 4 (N=12), 7% between the ages of 5 and 8 (N=4), 17% between the ages 9-12 (N=9), and less than 2% between the ages of 13-18 (N=1). Of the RYR1-related disease diagnoses, 27 of the total 51 children (53%) have been diagnosed with central core disease, 23 children (45%) are diagnosed with malignant hyperthermia (MH) or malignant hyperthermia susceptibility (MHS), 7 children (14%) diagnosed with congenital fiber type disproportion (CFTD), 4 (7%) are diagnosed with multi-minicore disease (MmD), and 1 child (2%) is diagnosed with centronuclear myopathy (CNM). Notably, children can be diagnosed with multiple RYR1-related diseases, and it is common for individuals who have a RYR1-related disease to also be diagnosed with MH/MHS (Clinical Care Guidelines, 2019).

This is why the total number of children diagnosed with a RYR1-related disease is more than the total number of children being reported (N=51). Additionally, 12 of the total 51 children (24%) were previously diagnosed with another disease that was later better explained under a RYR1-related disease.

Figure 1

Frequency of Primary RYR1-Related Disease Symptoms



Note. The total number of children in the sample is 51 (N=51).

As seen in Figure 1, 44 of the total 51 children reportedly experience weakness in the muscles closest to the torso (proximal muscle weakness). 42 children experience difficulties walking/climbing stairs, 40 children are easily fatigued, and 35 children have decreased tolerance to exercise and heat. The least common symptom reported is difficulties speaking, with 9 of the total 51 children experiencing this type of symptomology. Furthermore, regarding other

symptoms of a RYR1-related disease (not depicted in Figure 1), 17 children experience sensory or motor difficulties, 16 experience anxiety or depression, and 11 experience social difficulties. Only 5 children are reported to experience irritability, aggression, and severe oppositional behaviors.

Moreover, regarding additional mental health diagnoses, 7 of the 51 children have also been diagnosed with attention deficit/hyperactivity disorder, 7 children with an anxiety disorder (4 generalized anxiety disorder and 3 with separation anxiety disorder), 2 children with a specific learning disorder, and 1 with autism spectrum disorder and Major depression disorder, respectively. Importantly, over 84% of children received early intervention services. Lastly, the child's symptom severity as reported by the parents was reported. Overall, the RYR1-related disease symptoms of 19 children are reported as mild, 19 children's RYR1-related disease symptoms are also reported as moderate, and 13 children's RYR1-related disease symptoms are reported as severe.

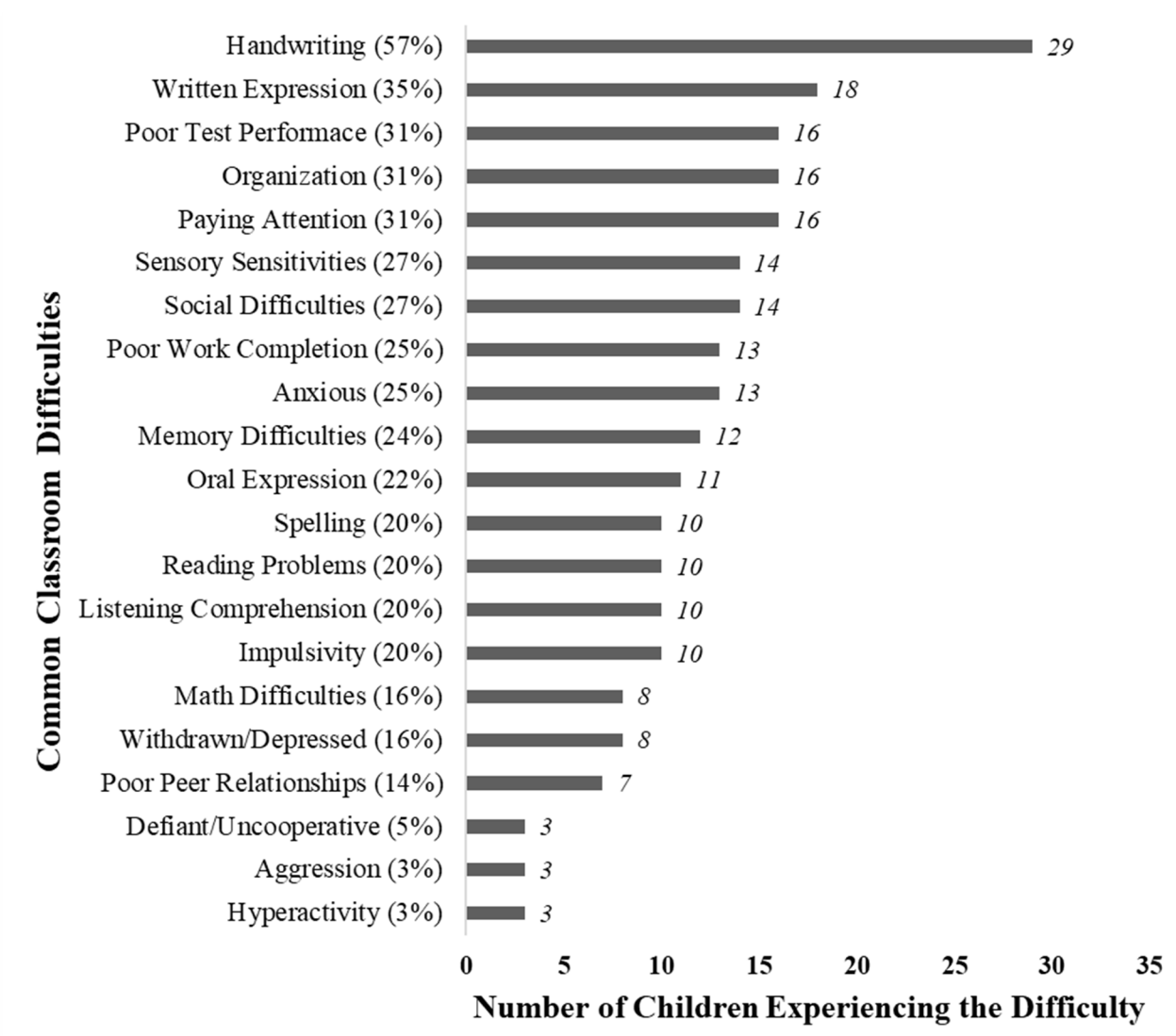
Common Classroom Difficulties

As noted in Chapter 3, research question 1, which asks "What are the common classroom problems experienced by children with a RYR1-related disease" is answered through question 27 of the RDEISS. As seen in Figure 2, which depicts the common difficulties children with a RYR1-related disease are currently experiencing in the classroom. The most common classroom problem was handwriting difficulties, with 29 of the total 51 parents (57%) indicating their child experiences this type of difficulty. Similarly, 18 parents indicated their child experiences difficulty with written expression, which served as the second most common classroom difficulty. Furthermore, 16 parents report that their child has difficulty paying attention in the classroom, has poor test performance, and poor organization. Also of note, 14 reported their

child experiences social difficulties and 13 parents indicate their child experiences anxiety. Moreover, only 3 parents (6%) indicate their child is observed to be hyperactive, aggressive, and/or defiant/uncooperative in the classroom.

Figure 2

Frequency of Common Classroom Difficulties of Individuals with a RYR1-related Disease



Note. The total number of children in the sample is 51 (N=51).

Frequency of Special Services in School

Research question 2 which asks "With what frequency do children with RYR1-related diseases receive special services in schools (i.e., formal special education, Section 504 Plan, informal services, no services at all)" is addressed in questions 28 and 29 through the RDEISS. As noted above, 40 out of the 51 children attend school in the United States to which this question applies. Of the 40 children being reported on, 21 (52%) children have had a Section 504 Plan throughout their educational careers. Additionally, 31 (78%) children have had formal special education services through an Individualized Education Program (IEP) under the Individuals with Disabilities Education Act (IDEA) throughout their educational careers. Notably, 12 parents report that their child has received both a Section 504 Plan and an IEP throughout their education. Furthermore, 19 (48%) children did not receive a Section 504 Plan and only 9 (23%) children did not receive an IEP throughout their education.

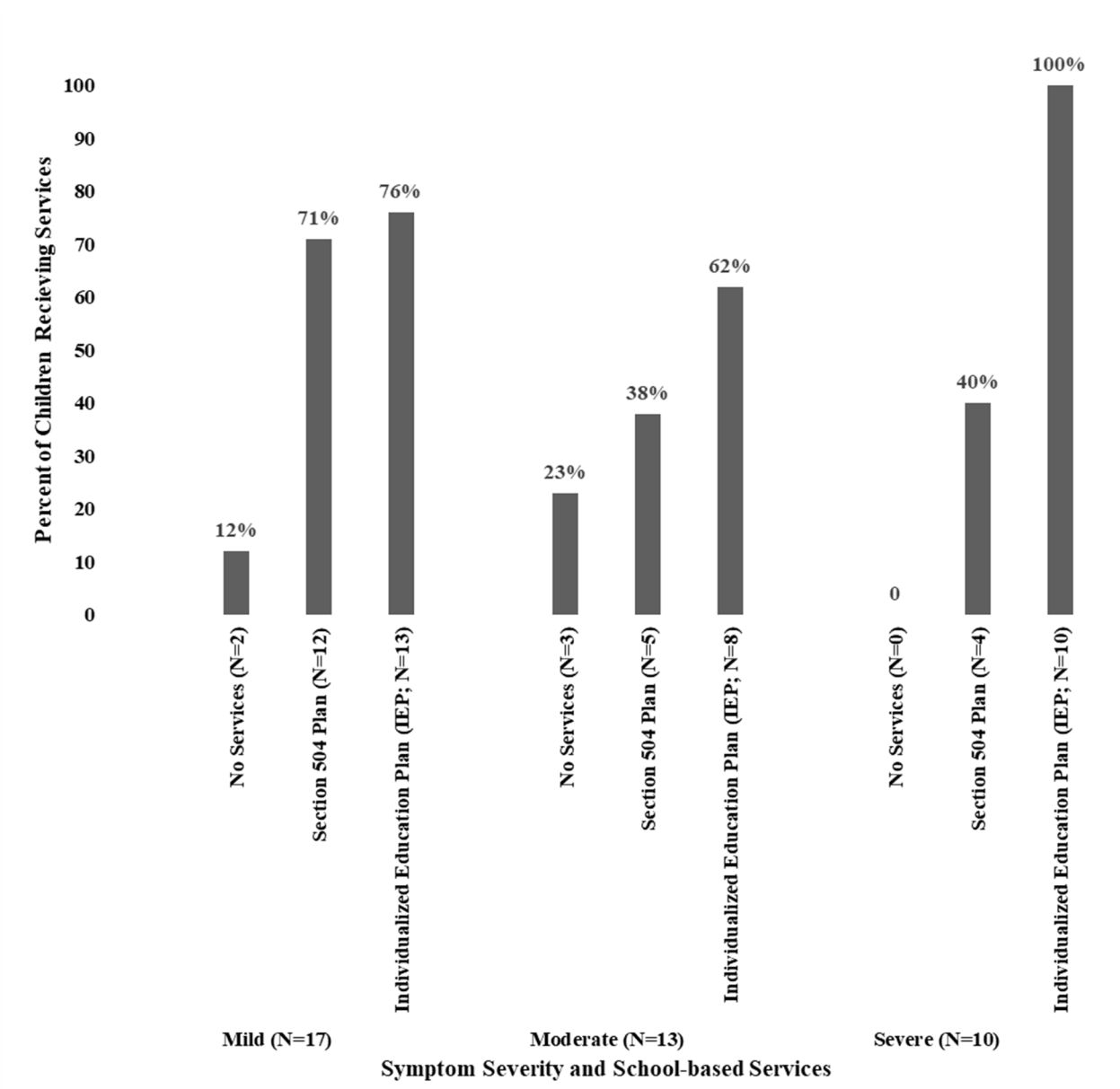
School-based Services and Symptom Severity

As this was not a focus of this study as it pertains to the research questions. It was worth investigating the frequency of school-based services (i.e., Section 504 Plan, IEP, or no services) with consideration of symptom severity as reported by the child's parent. As depicted in Figure 3, the total number of students represented in this sample is 40 with 17 having mild symptomology, 13 having moderate symptomology, and 10 having severe symptomology as reported by their parents. This analysis was only conducted on students attending school in the U.S. Notably, it is important to remember that participants were permitted to indicate their child has had both a Section 504 plan and an IEP. As discussed above, a vast majority of students are receiving services in school with a majority receiving an IEP. This is consistent across each severity group upon visual analysis. Also, all 10 students whose parents classify their symptomology as severe all receive an IEP, which is not the case for all students with mild and

moderate symptomology. Additionally, 23% of students (N=3) with moderate symptom severity do not receive services.

Figure 3

RYR-1 Symptom Severity and School-based Services



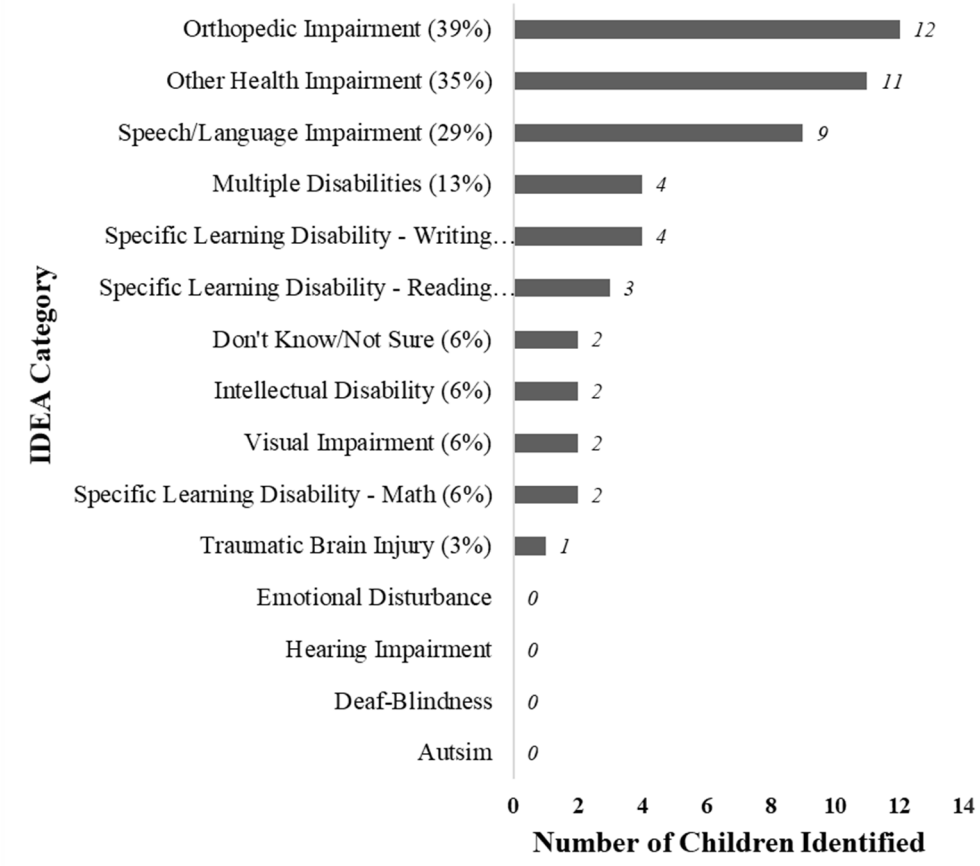
Note. Total children depicted in Figure 3 is N=40, as this was only presented to individuals residing in the U.S.

IDEA Category

Research question 3 which asks "If a child does receive special education services, under what IDEA category(ies) does the child qualify" is addressed through question 30 of the RDEISS. Question 30 of the RDEISS was provided to only the individuals who indicated that their child has received an IEP throughout their education. Thus, 29 parents were assessed on the IDEA category(ies) for which their child was found eligible for special education services. Since individuals can qualify for multiple IDEA disability categories, parents were instructed to report all categories for which their child qualified for. As such, the total frequency of categories will be greater than 31.

Figure 4

Frequency of IDEA Disability Categories among Individuals with a RYR1-related Disease



Note. The total number of children in this sample is 31 (N=31).

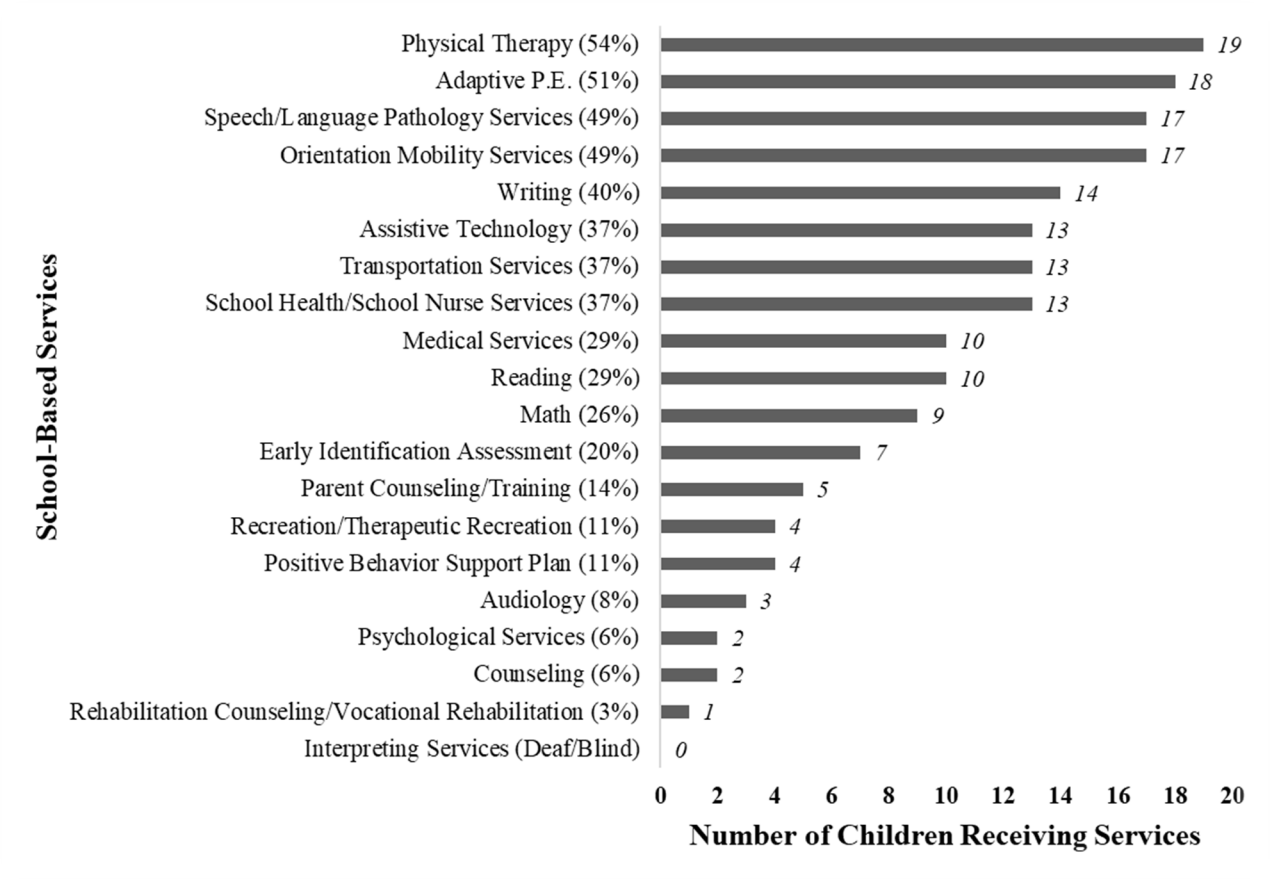
Figure 3 depicts the IDEA categories for which parents report their child has qualified for special education services. Out of the 29 parents reporting on 31 children, 12 (39%) children qualified for special education services under the IDEA disability category, orthopedic impairment. Similarly, 11 children (35%) qualified for special education services under the IDEA disability category other health impairment. The third most common IDEA disability category was Speech/Language Impairment with 9 children (29%) qualifying for services under this category. Lastly, 4 children (13%) qualified for services under multiple disabilities and a specific learning disability in writing, respectfully.

Types of School-Based Services

Research question 4 states "For children who do receive special services (i.e., IEP or Section 504 Plan) at school, what services do they receive and what is the parental satisfaction with those services?" The first part of this research question is addressed in Figures 4 and 5. Figure 4 depicts the school-based services provided to the child as part of their IEP or Section 504 Plan. Importantly, a total of 33 parents reporting on 35 children completed this portion of the survey. As seen in Figure 4, 19 children (54%) receive physical therapy as part of their IEP or Section 504 plan. Secondly, 18 children (51%) receive adaptive physical education, and 17 children (49%) receive speech/language pathology services. Additionally, 17 children (49%) also receive orientation mobility services and 14 children (40%) receive writing services. Moreover, 13 children (37%) reportedly receive school health/nursing services, transportation services, and/or assistive technology services, respectively. Lastly of note, less than 6% of children with a RYR1-related disease receive psychological or counseling services as part of their IEP or Section 504 Plan.

Figure 5

Frequency of Services Provided in School



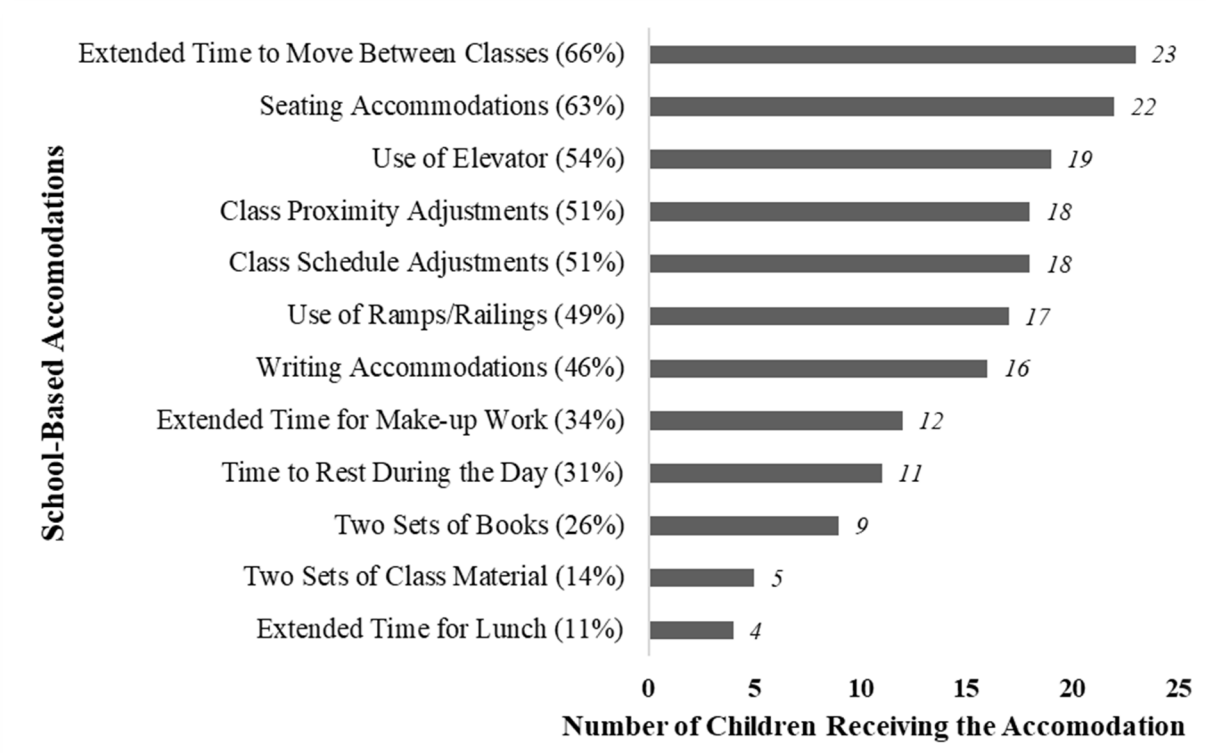
Note. The total number of children in this sample is 35 (N=35).

Similarly, Figure 5 depicts the common school-based accommodations that parents report their child receives as part of their IEP or Section 504 Plan. As seen in Figure 5, the most common accommodation is extended time to move between classes, with 23 (65%) children receiving this type of accommodation. Secondly, 22 children (62%) receive seating accommodations, and 19 children (54%) have access to and use an elevator in their schools. Furthermore, 18 children (51%) receive class schedule adjustments, class proximity adjustments (i.e., a class location closer to restrooms/cafeteria; all classes on ground level; etc.), and 17 children (49%) have access and use ramps/railings in school as part of their IEP or Section 504

Plan. Also seen in Figure 5, 16 children (46%) receive writing accommodations. Lastly, only 4 children (11%) receive extended time for lunch as part of their IEP or Section 504 Plan.

Figure 6

Frequency of School-Based Accommodations Provided to Individuals in School



Note. The total number of children in this sample is 35 (N=35).

Satisfaction of School-Based Services

The second part of research question 4 asks questions regarding parents' overall satisfaction with the services/accommodations provided to their child as part of their IEP or Section 504 Plan. Parents' overall satisfaction with services/accommodations was addressed through questions 34-37 of the RDEISS. Question 34 of the RDEISS asks "Overall how satisfied are you with the services currently outlined in this child's IEP or 504 Plan? Out of the 33 parents reporting their satisfaction with 35 children's school-based services provided through their IEP or

Section 504 Plan, 74% of parents report being satisfied with their child's services currently outlined in their IEP or Section 504 Plan. Additionally, 20% of parents report not being satisfied with their child's services, and less than 6% report being very satisfied. No parents indicate they are not at all satisfied with the services currently outlined in their child's IEP or 504 Plan. Question 35 of the RDEISS asks the parent "How much agreement is there, in your opinion, between the content of this child's IEP or 504 Plan document and services actually being provided? 74% of parents report that there is good agreement between the content of their child's IEP or 504 Plan and what is provided by their school. 17% of parents report that there is little agreement between what is written and what is provided, 9% of parents indicate there is very good agreement between what is written and what is provided, while no parents indicate that there is no agreement. Question 36 of the RDEISS asks parents "In practice, how satisfied are you with the school-related services actually being provided to this child?" Most of the parents (74%) indicate that they are satisfied with the school-related services actually being provided. 14% of parents indicate that are not satisfied, 8% report being very satisfied, while only 2% report they are not at all satisfied with the services actually being provided.

The parents who indicated they have low satisfaction (not at all satisfied or not satisfied) with their child's school services, were then asked to briefly explain why they have low satisfaction with the school services provided to their child. Their responses were pulled and themed as depicted in Table 1. Methods of evaluation of the qualitative research were derived from Guba & Lincoln (1994) and Castleberry and Nolen (2018). The following themes arose as overall concerns resulting in low satisfaction for these families. Communication breakdown; challenges in communication and understanding between parents, students, and school staff, particularly in schools lack of understanding of the child's needs. Advocacy; navigating disputes

and seeking necessary services through mediation with the school district, particularly in response to the school indicating they cannot "see" the disability and thus don't require services in school. Accessibility; families indicate they struggle with accessible facilities and environments, particularly during recess. Homeschooling versus public education; transitioning to homeschooling due to dissatisfaction with the quality of education and services provided in public schools. Implementation gaps; discrepancies between listed accommodations and their actual implementation, as well as differences between what services are put in practice compared to professional recommendations (i.e., medical doctors). Lastly, specific challenges; and difficulties encountered with feeding/mealtimes and snacks at school.

Table 1

Parents' Low Satisfaction Themes

Theme	Example Quote
Communication Breakdown (N=2)	"Poor compliance. Lack of understanding about my child's needs. Poor communication with substitute and support staff."
Advocacy (N=2)	"School claims that there is not enough impact on education to provide specific services, such as OT, due to dx and fatigue. Because the dx is mild and they are not able to “see” it effecting performance than they cannot service if they can’t see it. Currently going into mediation with school district."

	"We no longer have an IEP after no longer qualifying for speech services. Everything is now an accommodation and we have to fight for each one."
Accessibility (N=1)	"We still struggle with recess. The playground is not accessible."
Home School vs. Public Education (N=2)	"After years of being in public school and not being educated, I pulled my child to homeschool and had to start with the basics listed at 6 grade but it's at a 2-3rd grade level education. The services are fewer at home but he is progressing fast better than the public school where it was treated like a day care."
Implementation Gaps (N=2)	"They pick and choose which accommodations to actually give him, compared to what doctors have recommended." "Often are listed but not actually put into practice."
Specific Challenges (N=1)	"We are having a lot of problems around feeding/mealtimes and snack, but have a meeting with the school next week."

Note. A total of 10 parents indicated a reason for their low satisfaction with their child's school-based services.

Behavioral, Emotional, and Adaptive Functioning

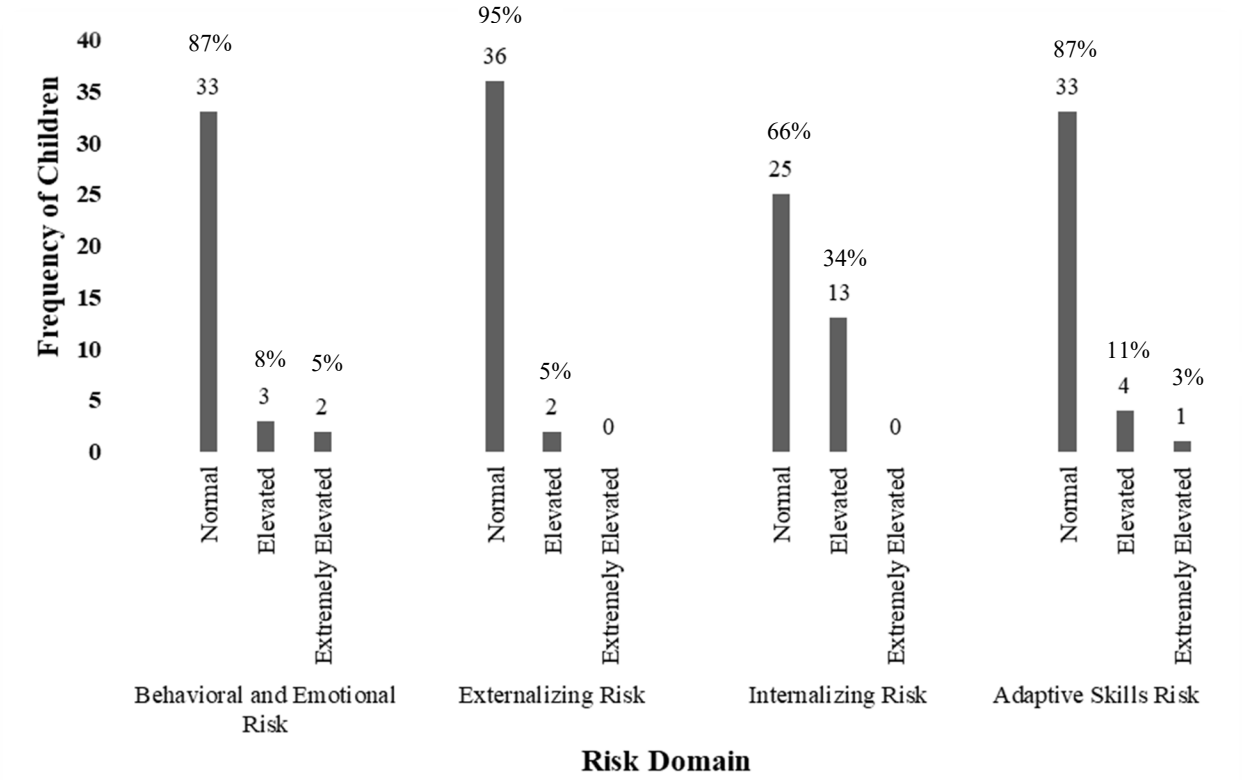
The last section of the survey assessed the extent individuals with a RYR1-related disease are at risk for behavioral and emotional difficulties. This section of the REDISS answers

research question five: "Do children with a RYR1-related disease experience behavioral and emotional problems as reported by parents on the Behavioral and Emotional Screening System (BESS)?" As noted in Chapter 3, the BESS is a 29-question screener that provides four total indexes (i.e., Externalizing Risk Index, Internalizing Risk Index, Adaptive Skills Index, and Behavioral and Emotional Risk Index). A total of 38 parents responded on behalf of 38 children/adolescents ranging from ages 6-18 years old. Results are depicted in Figure 6. Regarding the overall behavioral and emotional index, 33 of the total 38 children have a normal amount of risk for behavioral and emotional problems. Furthermore, 3 children were rated as having an elevated risk for behavioral and emotional difficulties, and 2 children were rated as having an extremely elevated risk for behavioral and emotional difficulties.

Moreover, 35 of the total 38 children were reported as having a normal level of externalizing/behavioral risk. While 2 of the 38 are exhibiting elevated risk for behavioral problems. Additionally, 25 of the total 38 children are exhibiting a normal amount of risk for internalizing/emotional difficulties. While 13 children are exhibiting elevated risk for emotional difficulties. Lastly, 33 of 38 children exhibit normal risk levels for adaptive functioning skills, while 4 children experience elevated risk, and 1 child is experiencing extremely elevated risk for adaptive skill difficulties.

Figure 7

Behavioral and Emotional Risk



Note. The total number of children in this sample is 38 (N=38).

Summary

This chapter details the results collected from the REDISS which assessed the educational experiences of children/adolescents diagnosed with a RYR1-related disease. Specifically, the results depicted in this chapter addressed and answered each research question outlined in Chapter 3. Chapter 5 will analyze and interpret the results summarized in this chapter in more detail, to further the understanding and meaning of the data presented. Additionally, the limitations of the current study will be presented as well as the future implications of the findings and their utility in the field of school psychology. Lastly, future research directions will be

discussed to inform possible next steps to aid this population's experiences in schools through research.

CHAPTER V

DISCUSSION

This chapter analyzes the results depicted and described in the previous chapter. Interpreting the data collected will provide further meaning to the results and their impact on the field of school psychology. Further, connections between the results and the literature provided in Chapter 2 will also be presented. Lastly, the limitations of the current research and future research directions are explored to help expand the field of school psychology in supporting families, students, school staff, and medical staff in serving children and adolescents with a RYR1-related disease.

RYR1-Related Disease Demographics

Importantly, the demographics collected regarding RYR1-related disease diagnosis and symptoms were collected. This is important to discuss to understand the population further. As discussed in Chapter 4, over 98% (N=50) of caregivers indicated that their child first exhibited RYR1-related disease symptoms before the age of 3. This is consistent with current literature as these individuals are born with this disease and are likely to exhibit symptoms at a young age. However, less than half the sample were diagnosed with a RYR1-related disease before the age of 3. This difference between observed symptoms and diagnosis is likely due to the vast diversity of symptomology, rarity of the disease, limitations of diagnostic methods, and unfamiliarity of disease by care providers depicted in the literature (Clinical Care Guidelines, 2019; Snoeck et al., 2015; Wang et al., 2012; Amburgey et al., 2011). Moreover, consistent with current literature on the disease, children are experiencing vastly diverse symptoms as illustrated in Figure 1 in the previous chapter. Thus, providing further evidence of the unique impact this disease has on

individuals. However, also consistent with current literature, the most common RYR1-related disease symptoms experienced by individuals were weakness in the muscles closest to the torso, difficulty walking/climbing stairs, and becoming easily fatigued. Lastly, information gathered through this research indicates that 31% of children in this sample experience anxiety and depression, and 22% experience social difficulties. This is also consistent with the findings of Ruitenbeek and colleagues (2019) and Lue and colleagues (2016) which indicate that individuals with a RYR1-related disease experience mental health and social difficulties. Lastly of note, the current study found a limited number of children have mental health diagnoses in addition to their RYR1-related disease. Specifically, only 14% of children also carry a diagnosis of ADHD, and 14% also carry a diagnosis of anxiety. Therefore, in this sample, a majority of children with a RYR1-related disease are not experiencing comorbid mental health conditions.

Common Classroom Difficulties

One of the primary goals of this literature was to measure the classroom difficulties of children with a RYR1-related disease. As such, Figure 2 (depicted in Chapter 4) illustrates the common reported classroom difficulties for these children. Thus, according to the current findings, children with a RYR1-related disease are most likely to experience difficulty with handwriting and written expression. This finding aligns with extant literature that demonstrates the weakness in muscle contractions is a primary feature of the disorder (Amburgey et al., 2013; Bharucha-Goebel et al., 2013; Clinical Care Guidelines, 2019; Klein et al., 2012; Lawal et al., 2020; Todd et al., 2018; Voermans et al., 2016; Zhou et al., 2007). Handwriting requires fine motor control to produce letters and words in order to communicate meaning (i.e., written expression). In turn, handwriting is known to be a significant predictor of writing performance (Graham et. al., 1998). Writing is a primary skill used within the classroom across most subjects

(i.e., math, spelling, written expression). Considering its importance, children and adolescents diagnosed with this disease should be prepared for handwriting and written expression difficulties as they develop in school.

Moreover, the least common difficulties experienced by students with a RYR1-related disease are defiance/uncooperative behaviors, aggression, and hyperactivity. Thus, we can conclude that within this sample of children with a RYR1-related disease, externalized behavior problems were not prevalent. Thus, it appears that behavior problems are not correlated with this disorder. Again, consistent with the literature, this motor disorder has not historically been associated with behavioral dysregulation (Clinical Care Guidelines, 2019). The present study has not found evidence of this either.

RYR1-Related Disease and Special Education

The next goal of this study was to assess the frequency in which children/adolescents receive special services in schools whether through a Section 504 Plan or special education services through an IEP. Importantly, these questions only applied to individuals who resided and attended school in the United States. Findings suggest that most (78%) students received special education services through an IEP. In comparison, 52% of children have had a Section 504 Plan. Notably, 30% of children have received a Section 504 Plan and an IEP in their educational careers. Overall, these findings suggest that children/adolescents diagnosed with a RYR1-related disease may benefit from accommodations, modifications, and/or specially designed instruction to support their education, depending on the presentation of the disease. Literature also indicates that the frequency of chronic health conditions in childhood is increasing and school-based professionals regularly encounter children with chronic health conditions in the school setting. These students with chronic health conditions often require

accommodations, assessment, intervention, and intensive collaboration with medical providers, families, and school staff to effectively care for these students (Dempsey, 2020). The current study continues to provide evidence of the importance of parent, school, and medical communication and collaboration, as well as an understanding of parents' rights regarding their child's education (Clinical Care Guidelines, 2019).

Furthermore, as most students in this sample have been identified as being eligible for special education services through an IEP, it is relevant to explore which disability category individuals with a RYR1-related disease are identified with. As noted in Chapter 2, there are 13 disability categories through IDEA in which children can be found eligible for special education services under. As illustrated in Figure 4 in the previous Chapter a majority of children/adolescents with a RYR1-related disease are identified as eligible for special education services under the disability category of orthopedic impairment and other health impairment. As noted in the Clinical Care Guidelines (2019), these are the two most relevant categories in which children with a RYR1-related disease may qualify for. Notably, the next most common disability category identified was speech/language impairment. As weakness in the bulbar muscles is an evident symptom of a RYR1-related disease, individuals have been known to experience difficulties speaking (Clinical Care Guidelines, 2019). Thus, speech/language impairment is an appropriate category for individuals experiencing difficulty with speech. In terms of school-based services and symptom severity. As expected, most individuals receive IEP's across symptom severity. Notably, all students with severe RYR-1 related symptoms receive IEPs. Furthermore, 5 individuals do not receive services in schools. Interestingly, a majority of these individuals (N=3) were classified as having moderate symptomology while only 2 had mild symptomology. This is interesting as we would expect students with mild symptoms to not

require/receive services. Results from this sample show that individuals with mild and moderate symptomology don't receive services in schools.

Types of School-Based Services

As we now know a vast majority of children and adolescents with a RYR1-related disease are identified as benefiting from school-based services to support their educational success, most commonly through an IEP, we look to investigate the types of school-based services provided for these students. As noted in previous chapters, school-based services can be broken down into two categories; accommodations (typically addressed through a Section 504 Plan) and modifications to instruction (addressed through special education and an IEP). Importantly, accommodations do not alter the content/standards of work in which the student is expected to complete, rather supports their environment to aid in their learning and educational success. Modifications, on the other hand, alter the standards and expectations to meet the child where they are to help support their educational success. For example, simply allowing a student more time to complete a spelling test does not change the standards and expectations of how many words they are expected to spell and thus they are graded on the same scale as their general education peers. This is an example of accommodation typically addressed through a Section 504 Plan. While modifying a child's spelling curriculum such that their spelling test only consists of 10 words instead of 25 words is an example of a modification or specially designed instruction, as the standard or expectation is modified to meet the child where they are. Modifications are addressed through an IEP.

Accommodations

As illustrated in Figure 5, students with a RYR1-related disease receive a variety of accommodations. Most commonly, students with a RYR1-related disease benefit most from

extended time to move between classes, seating accommodations, use of elevator, class proximity adjustments, class schedule adjustments, and use of ramps/railings. As this disease primarily impacts movement (Amburgey et al., 2013; Bharucha-Goebel et al., 2013; Clinical Care Guidelines, 2019; Klein et al., 2012; Lawal et al., 2020; Todd et al., 2018; Voermans et al., 2016; Zhou et al., 2007), it is to be expected that children will benefit from accommodations to support their ability to navigate around the school building effectively.

Modifications

As illustrated in Figure 5, students with a RYR1-related disease also receive a variety of modifications and specially designed instruction as part of their IEP. Children with a RYR1-related disease primarily receive physical therapy and adaptive physical education as part of their IEP. Furthermore, just under half of the students with a RYR1-related disease receive speech/language pathology services and orientation mobility services as part of their IEP. Again, these findings are consistent with current literature presented in Chapter 2 considering the physical impact and speech impact a RYR1-related disease can have on an individual. Interestingly, Research presented in Chapter 2 suggests that having a RYR1-related disease can have a negative impact on mental health and social functioning (Ruitenbeek et al., 2019; Lue et al., 2016). However, the research findings less than 6% of children with a RYR1-related disease receive psychological services or counseling as part of their IEP. Considering over 30% of this sample population report their child experiences anxiety and depression and over 20% report experiencing social difficulties; less than 6% receiving psychological services and counseling is notable. Current findings in unison with relevant literature suggest that students with a RYR1-related disease may benefit from counseling and psychological services in school.

Satisfaction of School-Based Services

Thus far we have explored the common classroom difficulties experienced by students with a RYR1-related disease, the identification of school-based services through a Section 504 Plan and an IEP, and the types of school-based services provided through a Section 504 Plan and an IEP. The next area of exploration regarding the educational experiences of students with a RYR1-related disease is satisfaction with school-based services being provided. Similar to all the data gathered through this study, satisfaction of services was measured by the caregivers of children with a RYR1-related disease.

Overall, a vast majority of caregivers of a child with a RYR1-related disease (74%) report being satisfied with their child's services currently outlined in their IEP or Section 504 Plan, while 20% indicate they are not satisfied with the services currently outlined in their child's IEP or Section 504 Plan. Similarly, a vast majority of caregivers of a child with a RYR1-related disease (74%) report that there is good agreement between the services outlined in their child's IEP or Section 504 Plan and the services actually being provided to their child. Therefore, we can conclude that the services outlined in a student's IEP or Section 504 Plan are consistent with the services actually being provided to them. Similarly, a vast majority of caregivers (74%) are satisfied with the services actually being provided for their child in school. Overall, the findings of this study suggest that more than 70% of caregivers of a child with a RYR1-related disease are satisfied with the school-based services being provided for their child in school.

Of those individuals who were not satisfied with the school-based services being provided for their child in school, they indicated that communication is a significant barrier to accessing quality services for their child. Most notably, they indicate that school staff lacks understanding of their child's RYR1-related disease and thus their needs. This is again consistent with the literature indicating that a majority of care practitioners are unfamiliar with the disease

and its impact on children (Wang et al., 2012). Thus, we can conclude that this trend is also, at times, prevalent in the school setting as well. Additionally, they indicate that they struggle to receive services as the school cannot "see" the disability and thus does not require services in school. This difficulty speaks to the vastly diverse symptomology of this disease, particularly its wide-ranging severity. For those individuals who present with mild symptom severity, it is likely school staff are unable to see the impact and can view the behavior as willful noncompliance rather than a manifestation of their disability (Hughes, 2018). Moreover, caregivers with low satisfaction report having difficulties with accessibility in schools particularly regarding recess thus inhibiting their child's ability to engage with peers during free play environments. Lastly, caregivers expressed specific concerns regarding lunch at school, such that they have struggled to access extended time to eat lunch as children with RYR1-related disease experience weakness in their bulbar muscles and thus may struggle with eating and swallowing (Clinical Care Guidelines, 2019). Similarly, as discussed above, this research found that the least common accommodation provided for individuals with a RYR1-related disease was extended time for lunch.

Behavioral, Emotional, and Adaptive Functioning

As noted in previous chapters, the BASC-3 Behavioral and Emotional Screening System (BASC-3 BESS) was utilized to measure the extent to which children and adolescents with a RYR1-related disease are experiencing behavioral and emotional difficulties. The BESS assesses behavioral and emotional strengths and weaknesses in children and adolescents. The BESS is not considered a comprehensive assessment but rather is a measurable tool to determine a child's risk level for developing emotional and behavioral difficulties (Kamphaus & Reynolds, 2015). Primary caregivers of children/adolescents completed the BESS parent form on behalf of their

children. The BESS provides four total scores to interpret. The four scores are the Behavioral and Emotional Risk Index, Externalizing Risk Index, Internalizing Risk Index, and Adaptive Skills Index. The results depicted in Chapter 4 are analyzed below.

The Behavioral and Emotional Risk Index provides information on a child or adolescent's risk of having or developing a behavioral or emotional problem (Kamphaus & Reynolds, 2015). Of the sample in the current study, over 86% of children with a RYR1-related disease experience a normal amount of risk. This does not mean that these children/adolescents are at an absence of risk for having or developing behavioral or emotional difficulties as all children are at risk for behavioral and emotional challenges, however, some may have a small amount of risk (Kamphaus & Reynolds, 2015). That being said, a significant majority of children with RYR1-related disease are within the normal range of risk for behavioral and emotional concerns. Overall, this result demonstrates a similar result to Ruitenbeek et al. (2019) who found that older individuals with a RYR1-related disease report comparable levels of anxiety, depression, interpersonal sensitivities, and hostility compared to healthy individuals. Thus, supporting that this trend is not only evident in adulthood but also development.

Similarly, the Externalizing Risk Index measures the level of risk in which children exhibit externalizing behaviors such as hyperactivity, aggression, and defiance. A vast majority of children with a RYR1-related disease are present with an age-appropriate amount of risk for externalizing behaviors. This research is consistent with previous research (Ruitenbeek et al., 2019) and the current classroom difficulties reported by caregivers in this sample. That is, a significantly small number of children present with hyperactivity, defiance, and aggression.

Moreover, the Internalizing Risk Index measures the amount of risk a child has of having or developing internalizing behaviors, such as anxiety, depression, and somatization. When

addressing these behaviors specifically, 65% of children with a RYR1-related disease experience an age-appropriate amount of risk for internalizing problems. As this is still the majority of children and adolescents in this sample it is important to note that 35% of children with a RYR1-related disease experience elevated internalizing behavior risk and are therefore at a higher risk for having or developing anxiety, depression, and somatization concerns.

Lastly, the adaptive skills risk index measures the level of risk children/adolescents experience adaptive skills behaviors such as, adapting to their environment and changes in routine, completing everyday activities, functional communication skills, study skills, and social skills. As noted in Chapter 4 a vast majority (86%) of children/adolescents with a RYR1-related disease experience a normal amount of risk for having/developing adaptive skill deficits. This finding offers a complementing perspective to Ruitenbeek et al. (2019). The authors found that compared to the general healthy population, older individuals with a RYR1-related disease experience significantly more difficulties functioning across multiple domains (i.e., social interaction and home management). As these are different domains measured in the current study, the current study finds that younger individuals do not experience elevated risk for functioning and adaptive skill difficulties. As such, we can conclude that it is likely the difficulties in functioning/adaptive domains may apply to adulthood rather than younger developing children/adolescents as they can rely on caregivers and family for support in those areas.

Overall, most children with a RYR1-related disease experience a normal amount of risk for developing behavioral and emotional problems. There is clear evidence of this with consideration that over 85% of children in the sample experience normal externalizing risk, adaptive skills risk, and overall behavioral and emotional risk. Upon closer investigation, a much

smaller majority (65%) of children/adolescents experience a normal level of risk for developing internalizing problems (anxiety, depression, somatization). As such, we can conclude that simply having a RYR1-related disease does not warrant concern for developing behavioral and emotional problems, however, families, health care practitioners, and school staff should monitor the internalizing behaviors of individuals with a RYR1-related disease. As evident in the current sample, the largest amount of children/adolescents with elevated risk (35%) for developing or having problems, was in internalizing behaviors.

Limitations

The main limitation of this study relates to the limited number of participants who engaged in the study. Although this was to be expected due to the rarity of this disease, it is still worth noting. Additionally, some participants ended the survey before finishing, thus limiting the number of responses. Moreover, this study allowed international participants who reside outside the United States. Therefore, the survey sections that addressed United States Federal special education services could not be completed by the international caregivers, and thus limited responses regarding special education in the RDEISS. The diversity of the sample is also worth noting as a majority of the sample was white, while only 13% of the population made up other ethnicities (i.e., Hispanic, Latinx, or Spanish; Native American or Alaska Native; Middle Eastern; Black/African American; Asian).

Conclusion

In conclusion, the current study depicts the classroom difficulties experienced by children/adolescents with a RYR1-related disease. That is, primarily, children/adolescents experience difficulties with handwriting and written expression. This study also investigated the frequency of special education identification among children with a RYR1-related disease.

Findings suggest that Orthopedic Impairment and Other Health Impairment are the primary IDEA disability categories in which children with a RYR1-related disease are typically identified. Further, the current research assessed which types of school-based services (accommodations and modifications) are provided for students with a RYR1-related disease. Findings suggest that physical therapy, adaptive physical education, speech-language pathology services, and orientation mobility services are the primary modifications to instruction provided for children/adolescents with a RYR1-related disease in school. Extended time to move between classes, seating accommodations, use of the elevator, and class proximity/class schedule adjustments were the primary school-based accommodations provided for individuals with a RYR1-related disease. The current research further measured caregiver satisfaction with their child's school-based services. Overall, a majority of caregivers of a child/adolescent with a RYR1-related disease report being satisfied with the school-based services provided, written, and implemented as part of their child's IEP or Section 504 Plan. Of those who were not satisfied with their child's school-based services, communication difficulties, lack of understanding of their child's disability and needs, school staff refusing to provide services as they cannot "see" their child's disease impact on their educational function, and difficulty around lunch/mealtime were the primary common themes pulled for reasons they were not satisfied. Lastly, this study investigates the amount of risk children/adolescents have of developing or currently having behavioral and emotional problems. Overall, a majority of children with a RYR1-related disease experience a normal amount of risk for behavioral, emotional, and adaptive skill difficulties. However, a lesser majority exhibit a normal amount of internalizing behavioral (anxiety, depression, somatization) risk. Thus, supporting the need for close monitoring of internalizing symptoms as a child/adolescent with a RYR1-related disease develops.

As noted above, the frequency of students with chronic health conditions in schools is increasing. The CDC Health Schools (2021) reports that in the United States, more than 40% of school-aged children have at least one chronic health condition (i.e., asthma, obesity, other physical conditions, and behavior/learning problems). School-based professionals are regularly encountering children with chronic health conditions in the school setting. Notably, students with such chronic health conditions often require accommodations, assessment, intervention, and intensive collaboration with medical providers, families, and school staff to effectively care for these students (Dempsey, 2020). The current study continues to provide evidence of the importance of parent, school, and medical professional communication, and active monitoring (academic, behavioral, emotional, and social) of students with chronic health conditions. School staff such as school psychologists and other mental health professionals (social workers, counselors, etc.) in the schools are in a unique position to coordinate school-based services for students with complex medical conditions. As medical healthcare professionals have extensive knowledge in addressing medical issues regarding children and their chronic illness; mental health professionals (i.e., school psychologists) understand the complex nature of cognitive, academic, behavioral, emotional, and social factors of children with chronic/complex health-related conditions (Perfect et al., 2023). Thus, School psychologists play an essential role in fostering successful and positive educational experiences for children with chronic health conditions such as a RYR1-related disease.

The current research answered the research questions depicted in Chapter 1 in their entirety and hopes to inform families of children with a RYR1-related disease, health care practitioners, and school staff of what the educational experiences of these children/adolescents look like and how to best treat, care and support them as they develop and grow. Future research

should look to investigate this topic further with a larger and more diverse sample size. Additionally, future researchers may consider evaluating group differences in the educational experiences of these children compared to a healthy population.

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Appendix A

RYR1-Related Disease Educational Impact and Satisfaction Survey (RDEISS)

INCLUSION CRITERIA

1. Are you the primary caregiver of a child currently in grade K-12 diagnosed with a RYR1-related disease?
    - **a. Yes**
    - b. No

Parent Demographics (Bitterman et al., 2008; *Adapted*)

1. What is your (the caregiver) gender identity?
    a. Female
    b. Male
    c. Non-Binary
    d. Prefer not to specify

2. What is your (the caregiver) race identity?
    a. Asian
    b. Black/African American
    c. Middle Eastern
    d. Native American or Alaska Native
    e. Native Hawaiian or Other Pacific Islander
    f. Hispanic, Latinx, or Spanish origin
    g. White
    h. Biracial or Multicultural
    i. Race not listed/Other
    j. Unknown

3. What is your household income?
    a. $20,000 or less
    b. $20,001-$40,000
    c. More than $40,000

4. What is your highest education level?
    a. Less than a high school diploma
    b. High school diploma or GED
    c. Some college
    d. Four-year degree or higher

5. Is there another caregiver in the household?
    a. Yes
    b. No

6. (*If yes to 5*) What is the highest education level of the other caregiver?
    a. Less than high school diploma
    b. High school diploma or GED
    c. Some college
    d. Four-year degree or higher
    e. N/A – single caregiver

Child Demographics (Maura A. Miglioretti, 2019; *Adapted* & National Association of Independent Schools: Student Demographic Survey, 2016; *Adapted*).

Instructions: If you have 2 or more children with a RYR1-related disease, please complete this survey on your oldest child. With your oldest child in mind, please continue to complete the remainder of the survey.

7. Does your child attend school in the United States? (*If no, only fill out to question 25)*
    a. Yes
    b. No
8. What is this child's grade level for the current academic year? (Select from list)
    a. (Select from dropdown menu, range K-12)

9. Does this child attend school full time or part-time?
    b. Full time - 5 or more full days a week
    c. Part-time - less than 5 full days a week

10. What type of school does this child currently attend?
    a. Public
    b. Private/parochial (not a private school specifically for children with disabilities)
    c. Private school specifically for children with disabilities
    d. Charter
    e. Cyber
    f. Homeschooled
    g. Other

11. What is this child's race?
    a. Asian
    b. Black/African American
    c. Middle Eastern
    d. Native American or Alaska Native
    e. Native Hawaiian or Other Pacific Islander
    f. Hispanic, Latinx, or Spanish origin
    g. White
    h. Biracial or Multicultural
    i. Race not listed/Other
    j. Unknown

12. What is your child's gender identity?
    a. Female
    b. Male
    c. Non-Binary
    d. Prefer not to specify

13. What is your child's current age in years?
    a. (Select age from dropdown menu, range <3-21)

14. Was your child ever retained (held back) in school?
    a. Yes
    b. No

15. What was your child's age in years when they first exhibited RYR1-related disease symptoms?
    a. (Select from dropdown menu, range <3 years-21)

16. How old was your child (in years) when they were diagnosed with a RYR1-relateded disease?
    a. (Select from dropdown menu, range <3 years-21)

17. What grade was your child enrolled in when they first exhibited RYR1-related disease symptoms?
    a. (Select from dropdown menu, range prek-12)

18. My child has been diagnosed with (select all that apply):
    e. Central Core Disease (CCD)
    f. Multi-minicore Disease (MmD)
    g. Centronuclear Myopathy (CNM)
    h. Congenital Fiber Type Disproportion (CFTD)
    i. Malignant Hyperthermia (MH)

19. Was your child previously diagnosed for another disease that was later better explained under a RYR1-related disease?
    a. Yes
    b. No

20. My child's RYR1-related disease primary symptoms include (indicate all that apply):
    j. Weakness of the eye muscles
    k. Weakness of the facial muscles
    l. Weakness of the muscles closest to the torso (proximal muscle weakness)
    m. Muscle cramping and/or muscle pain

n. Decrease tolerance to exercise and heat
o. Breathing problems
p. Difficulties walking/climbing stairs
q. Contractures
r. Hip dislocation
s. Spinal deformities (scoliosis, kyphosis, lordosis)
t. Difficulty eating and swallowing
u. Difficulty speaking

21. My child's other symptoms of their RYR1-related disease include (indicate all that apply)
    a. Anxiety
    b. Depression
    c. Irritability, aggression, or severely oppositional behaviors
    d. Deterioration of school performance
    e. Sensory or motor difficulties
    f. Somatic symptoms (includes sleep disturbances, wetting/bed wedding, urinary frequency)
    g. OCD
    h. Social difficulties

22. My child has also been diagnosed with (indicate all that apply)
    a. Attention deficit hyperactivity disorder
    b. Autism spectrum disorder
    c. Major depressive disorder
    d. Generalized anxiety disorder
    e. Separation anxiety disorder
    f. Oppositional defiant disorder
    g. Specific learning disorder
    h. Other developmental or psychiatric disorder

23. Did your child receive early intervention services in schools prior to kindergarten?
    a. Yes
    b. No

24. What is your perception of your child's symptom severity? (See definitions for details)
    - Mild – low symptom severity; very few symptoms; small amount of impaired daily functioning; requiring minimal assistance.
    - Moderate – modest symptom severity; some symptoms, medium amount of impaired daily functioning, requiring assistance.
    - Severe – severe symptom severity; many symptoms, large amount of impaired daily functioning; requiring significant assistance.
        a. Mild
        b. Moderate

c. Severe

Educational Information (Maura A. Miglioretti, 2019; Adapted)

25. *(If b to 7, skip to exit message after 25 is answered)* What difficulties does your child currently experience **at school**? (Indicate all that apply)
    a. Paying attention
    b. Hyperactivity
    c. Impulsivity
    d. Handwriting
    e. Social difficulties
    f. Organization
    g. Aggression
    h. Anxious
    i. Withdrawn/depressed
    j. Sensory sensitivity
    k. Memory problems
    l. Poor work completion
    m. Poor test performance
    n. Defiant/uncooperative
    o. Oral expression
    p. Listening comprehension
    q. Reading problems
        - Basic reading
        - Fluency
        - Reading comprehension
    r. Math problems
        - Calculation
        - Math reasoning
    s. Spelling
    t. Written expression
    w. Poor peer relationships
    x. No problems.

26. Has your child had a Section 504 Plan?
    a. Yes
    b. No

27. Has your child had formal special education services (i.e., Individualized Education program – IEP) under the Individuals with Disabilities Education Act (IDEA)?
    a. Yes
    b. No

28. *(If a to 27)* As your child was found eligible for special education services and an IEP was written, the IEP team identified which disability category/categories?
    a. Autism
    b. Specific Learning Disability – Reading, Writing, Math
    c. Blind
    d. Visually impaired
    e. Deaf
    f. Hearing impaired
    g. Other health impairment
    h. Emotional disturbance
    i. Multiple disabilities
    j. Traumatic brain injury
    k. Speech/Language impairment
    l. Intellectual disability
    m. Orthopedic impairment
    n. Don't know/aren't sure).

29. *(If a to 26 or 27)* What services are provided for your child as part of their IEP or 504 plan?
    a. Reading
    b. Writing
    c. Math
    d. Positive behavior support plan
    e. Related services including:
        - Audiology
        - Counseling
        - Early identification and assessment
        - Interpreting services (deaf/blind)
        - Medical services
        - Orientation mobility services
        - Parent counseling/training
        - Physical therapy
        - Psychological services
        - Recreation/therapeutic recreation
        - Rehabilitation counseling/vocational rehabilitation
        - School health/school nursing services
        - Speech/language pathology services
        - Transportation services
    f. Assistive technology
    g. Adaptive P.E.
    h. Accommodations: (Select all that apply)
        - Seating accommodations
        - Class schedule adjustments
        - Class proximity adjustments (i.e., closer to bathroom and/or cafeteria and all classes on ground level, etc.)

- Writing accommodations (i.e., provided notes, provided a scribe for writing)
- Use of ramps/railings
- Use of elevator
- Extended time for lunch
- Extended time for make-up work
- Extended time to move between classes
- Two sets of books
- Two sets of class materials
- Time to rest during the day

30. Have you ever withdrawn your child from their school due to symptoms of a RYR1-related disease?
    a. Yes
    b. No

(Qualtrics logic: (If a to 25 or 26)

School Service Satisfaction (Slade et al., 2018; adapted)

31. Overall, how satisfied are you with the services currently outlined in your child's IEP or 504 plan?
    - Not at all Satisfied
    - Not Satisfied
    - Satisfied
    - Very Satisfied

32. How much agreement is there, in your opinion, between the content of your child's IEP or 504 plan document and services actually being provided?
    - No Agreement
    - Little Agreement
    - Good Agreement
    - Very Good Agreement

33. In practice, how satisfied are you with the school related services actually being provided to your child?
    - Not at all Satisfied
    - Not Satisfied
    - Satisfied
    - Very Satisfied

34. If you have **low satisfaction** with your child's school services, briefly explain why below.

    _____

Behavioral and Emotional Screening System-BESS (Kamphaus, R. W., & Reynolds, C. R. (2007). 1-29 questions - **Parent = 3-18 years old – Child/Adolescent K-12**

35. Gets along well with others.
36. Is easily upset.
37. Has a short attention span.
38. Gets into trouble.
39. Sets realistic goals.
40. Worries about things that cannot be changed.
41. Disobeys.
42. Says, "I hate myself."
43. Tracks down information when needed.
44. Is easily frustrated.
45. Is good at getting people to work together.
46. Breaks the rules.
47. Is nervous.
48. Defies people in authority.
49. Seems lonely.
50. Is overly aggressive.
51. Adjusts well to changes in routine.
52. Deceives others.
53. Says, "Nobody likes me."
54. Organizes chores or other tasks well.
55. Has trouble concentrating.
56. Gives good suggestions for solving problems.
57. Is negative about things.
58. Disrupts other children's activities.
59. Tries to bring out the best in other people.
60. Acts out of control.
61. Complains about health.
62. Responds appropriately when asked a question.
63. Loses temper too easily

Exit Message

64. If you have another child with a RYR1-related disease and would like to complete the survey again with that child in mind. Please select the options below.

    a. LINK to complete survey again.
    b. No