ABSTRACT

Positive Effects of Hippotherapy on Children with Dravet Syndrome

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Hippotherapy has been used since the time of Hippocrates as a therapeutic treatment. Though not experimentally researched in Dravet Syndrome, there is reason to believe that hippotherapy would be beneficial to both the physical and emotional needs of these children. Severe Myoclonic Epilepsy of Infancy, or more commonly known as Dravet Syndrome, is a rare seizure disorder that effects roughly one in every thirty thousand children before the age of seven. It is an epileptic encephalopathy that is caused by a mutation in the SCNA1 gene. This mutation is the basis for the numerous, often uncontrollable seizures that these children face. As a side effect of these seizures, most children experience motor impairments, most notably ataxia. There are many drug and non-pharmacological treatments that have been shown effective to help reduce the number and severity of the seizures, but most are very expensive and still experimental. Based on proven results in patients with similar disorders such as multiple sclerosis that cause ataxia, it is concluded that hippotherapy would be effective but needs further experimental study. In addition to hippotherapy being cheaper than many alternatives, it would be effective in the following areas of a child with Dravet Syndrome's life such as quality of life, social skills, coordination, and balance.

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POSITIVE EFFECTS OF HIPPOTHERAPY ON CHILDREN WITH DRAVET SYNDROME

A Thesis Submitted to the Faculty of

Baylor University

In Partial Fulfillment of the Requirements for the

Honors Program

By

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Waco, Texas

May 2016

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CHAPTER ONE

Literature of the Disorder

Severe Myoclonic Epilepsy of Infancy (SMEI) or more commonly known as Dravet Syndrome was first described in 1978, but it was not declared a syndrome until 1989 (Dravet, 2011). It is estimated that between 1 in every 20000 and 1 in every 40000 children before the age of seven have Dravet Syndrome, and Dravet Syndrome is more commonly found in males than females at a 2:1 ratio (Bender, Morse, Scott, Holmes, & Lenck-Santini, 2012). In 64% of cases, there is a family history of epilepsy or febrile convulsions (Davis, 2012). More recently, geneticists have found that approximately 70-80% of children with Dravet Syndrome have a mutation of the SCN1A gene ("What is Dravet Syndrome?," n.d.).

Dravet Syndrome is classified as an epileptic encephalopathy, which according to the International League Against Epilepsy (ILAE) (2001) means that "the epileptiform abnormalities themselves are believed to contribute to the progressive disturbance in cerebral function" (Berg et al., 2010). However, in 2010, the ILAE further specified that, "the epileptic activity itself may contribute to severe cognitive and behavioral impairment above and beyond what might be expected from the underlying pathology alone, and that these can worsen over time" (Berg et al., 2010).

The syndrome generally appears as seizures during the first years of life in otherwise healthy infants (Dravet, 2011). Specifically, between three and eight months of

age, infants experience seizures as febrile seizures with clonic (jerking) or tonic clonic (jerking preceded by stiffening), general or unilateral side-alternating seizures, and sometimes prolonged up to epilepticus status where the patient does not regain consciousness between seizures ("What is Dravet Syndrome?," n.d.). After a few months, polymorphic seizures such as: myoclonic seizures, atypical absences, focal seizures, and episodes of obtundation status appeared (Dravet, 2011).

Obtaining a Dravet Syndrome diagnosis in a timely manner has proven to be extremely difficult for most families. According to a 2009 survey of parents of children with Dravet Syndrome, at least 50% of the respondents did not receive an official diagnosis for more than three years after their child experienced his/her first seizure (Skluzacek, Watts, Parsy, Wical, & Camfield, 2011). In addition, 68% of these families consulted with three or more neurologists before receiving the diagnosis of Dravet Syndrome (Skluzacek, Watts, Parsy, Wical, & Camfield, 2011). Children suspected to have Dravet Syndrome can undergo genetic testing to confirm the diagnosis, but the test is extremely expensive and most families already have high medical bills and medication debt ("What is Dravet Syndrome?," n.d.). If the family can afford genetic testing, diagnosing Dravet Syndrome is much easier. Seventy percent of Dravet Syndrome cases have one of over 600 mutations in the coding sequence of the SCN1A gene (Dravet, 2011). More than half of the Dravet Syndrome mutations are due to stop codons or deletions that cause loss of function (Catterall, 2012). This demonstrates that SCN1A haploinsufficiency is pathogenic (Bechi et al., 2012). Up to 80% of individuals with clinical features of Dravet Syndrome test positive for mutations of the SCN1A gene

("What is Dravet Syndrome?," n.d.). These mutations result in the dysfunction of voltage-gated sodium channels in neurons (Marini et al., 2011). The current hypothesis is that a deletion of the Nav1.1 channel reduces the excitability of GABAergic Purkinje neurons, which are the output pathway for information on movement, coordination, and balance from the cerebellar cortex (Fletcher et al., 1996; Raman and Bean, 1997; Grusser-Cornehls and Baurle, 2001; Suasbier et al., 2004). Degeneration of Purkinje neurons and abnormal expression of voltage-gated ion channels in them are associated with ataxia which is one of the major side effects of the disorder (Fletcher et al., 1996; Raman and Bean, 1997; Grusser-Cornehls and Baurle, 2001; Suasbier et al., 2004) Other major side effects include photosensitivity, delayed psychomotor development, clumsiness, intercritic myoclonus, and delayed language/speech development (Bureau & Bernardina, 2011).

Dravet Syndrome can be considered complete (SMEI) or borderline (SMEIB). Patients with Borderline Dravet Syndrome have all the same characteristics and side effects except that they will lack the myoclonic seizures (Dravet, 2011). This idea has become widely accepted and is part of the reason Dravet Syndrome has been coined, after Dr. Dravet who discovered it.

CHAPTER TWO

Treatments of Dravet Syndrome

As with any disorder, the earlier a patient can start anti-seizure medications, the better. Not only does medication reduce the risk of seizures in general, it also can help

reduce the severity of the seizures that do occur. This is very important from the standpoint of the side effects and general outcome of the seizures. The more severe and frequent the seizures are the more detrimental the effect on physical, social, and emotional development of the child. There are currently two popular drugs being used by Dravet Syndrome patients, Stripentol (STP) and Topiramate (Chiron & Dulac, 2011). Though both are very popular, Topiramate has not been as extensively studied as Stripentol in Dravet Syndrome and is more often used in pharmacoresistant cases of Dravet Syndrome (Chiron & Dulac, 2011). There are a multitude of other antiepileptic drugs that have been used in the past, but Dravet Syndrome is very different from most epileptic disorders (Catterall, 2012). In fact, some of the most common antiepileptic drugs can actually make the seizures of patients with Dravet Syndrome worse ("What is Dravet Syndrome?," n.d.).Sodium channel blocking anti-epileptic drugs should be avoided if the diagnosed condition is caused by haploinsufficiency of Nav..1 channels, as is the case with SMEI (Catterall, 2012). These drugs are known to exacerbate symptoms in patients with Dravet Syndrome instead of relieving them (Catterall, 2012). This same seizure-worsening effect is seen with Lamotrigine, Carbamazepine, Vigabatrin, high-dose intravenous Phenobarbital and/or Pentothal (Catterall, 2012). Generally, patients with Dravet Syndrome are placed on a treatment program or regimen that includes a combination of two or more drugs, nonpharmacological/alternative treatment, and multiple types of therapy (Korff et al., 2007).

CHAPTER THREE

Alternative Treatment Options

In addition to pharmacologic treatment, most children with Dravet Syndrome also undergo alternative therapies in conjunction with antibiotics (Korff et al., 2007). These therapies can help reduce the number of seizures as well as help counteract the damaging side effects of the seizures associated with Dravet Syndrome. Some of the more popular alternative treatment options include: intravenous immunoglobulin therapy, vagus nerve stimulation therapy, the ketogenic diet, cannabis, and hippotherapy ("What is Dravet Syndrome?," n.d.). While these are not all of the alternative therapy options, these are among the more popular options that families of children with Dravet Syndrome are currently seeking.

Intravenous Immunoglobulin Therapy (IVIg) is a form of treatment for epilepsy that has been used since 1977 ("Role of Inflammation in Epilepsy and Treatment With IVIg | Ice Epilepsy Alliance," n.d.). The use of this therapy is based on the principle that reducing inflammation reduces the risk of seizure ("Role of Inflammation in Epilepsy and Treatment With IVIg | Ice Epilepsy Alliance," n.d.). This has been scarcely used as a treatment option for children with Dravet Syndrome, but in some cases has reduced seizure frequency and severity. Another less commonly used treatment is vagus nerve stimulation (VNS). This therapy acts on the principle that if regular electrical pulses are sent to the brain, the regularity of pulses can help reduce seizure occurrence and severity (Caraballo, 2011). It is more commonly used in other forms of epilepsy, but it has been successful in some cases of children with Dravet Syndrome (Caraballo, 2011).

The ketogenic diet is perhaps the most popular alternative treatment. It consists of taking in three to four times as much fat as protein and carbohydrates combined (Freeman & Kelley, 1994; Kossoff et al., 2009). Generally, the ketogenic diet is advised for children between two and seven years of age due to the high fat content, but it has also been affective in adolescents and adults (Kossoff et al. 2009). Some patients have started using cannabis to help relax muscle tone and tension, but this is not a very popular option. Finally, hippotherapy is an alternative therapy that, while not commonly considered for children with Dravet Syndrome, may actually be a viable and effective option.

CHAPTER FOUR

Brief Discussion on the Different Types of EAAT

Hippotherapy, a therapy using a horse to help patients with neuromuscular dysfunction, is just one type of therapy classified under the umbrella term, equine assisted activities and therapies (EAAT). Equine assisted activities and therapies include two main groups: equine assisted activities (EAA) and equine assisted therapies (EAT) (Path International, 2016). Equine assisted activities include any activities at a center where the patient, instructors, volunteers, and equines are involved. This can include vaulting, grooming, ground activities, shows, parades, equine-assisted learning, or therapeutic riding (Path International, 2016). Interactive vaulting includes the patient or student performing different movements on and around a horse (Path International, 2016). These movements range from basic moves such as sitting on the horse without holding on to more difficult movements such as standing or kneeling on a horse (Path

International, 2016). Equine-assisted learning promotes life skill development for individual, professional, and educational goals through interaction with horses (Path International, 2016). The purpose of therapeutic riding is to positively contribute to individuals with special needs physically, emotionally, socially, and cognitively without having the presence or goals of a medical professional (Path International, 2016).

Equine assisted therapies are different from equine assisted activities because they include rehabilitative goals that are related to the patient's needs and set by a medical professional such as an occupational therapist, physical therapist, or speech therapist who must be present at the time of therapy (Path International, 2016). Different types of equine assisted therapy include equine-assisted psychotherapy, equine-facilitated psychotherapy, and hippotherapy (Path International, 2016). Equine-assisted psychotherapy and equine-facilitated psychotherapy are very similar in that they both involve experiential psychotherapies with either an individual or group and horses (Path International, 2016). The minor difference is that equine-facilitated psychotherapy places more emphasis on riding as the mental health professional uses more instruction when working with the horses (Path International, 2016). In equine-assisted psychotherapy, the therapist instructs during groundwork, so the patient does not have to actually mount and ride the horse (Path International, 2016). This keeps the primary focus on the client to solve problems and not the horse.

CHAPTER FIVE

Hippotherapy

Hippotherapy has long been described as physically beneficial to humans. One of the earliest written descriptions of this was when Hippocrates wrote a chapter on "Natural Exercise" and mentioned riding circa 400 BC (Macauley & Gutierrez, 2004). Much later, in the 1960s, the countries of Germany, Switzerland, and Austria all began to use the horse as an adjunct to physical therapy and called this endeavor "hippotherapy" which is the term used today ("History of Hippotherapy and AHA Inc.," 2010). Over the next forty years, scientists all over the world worked quickly to further investigate and develop the idea of hippotherapy. By 1995, a standardized curriculum had been developed and the American Hippotherapy Association was formed ("History of Hippotherapy and AHA Inc.," 2010).

The American Hippotherapy Association (AHA) defines hippotherapy as "a term that refers to the use of the movement of the horse as a strategy by physical therapists, occupational therapists, and speech-language pathologists to address impairments, functional limitations, and disabilities in patients with neuromusculoskeletal dysfunction" ("History of Hippotherapy and AHA Inc.," 2010). "This strategy is used as part of an integrated treatment program to achieve functional outcomes" (Wilson & Turner, 1997). Patients may be oriented facing forward, backward, sideways, or lying prone or supine depending on their condition and the goal of the therapy set by the therapist ("History of Hippotherapy and AHA Inc.," 2010).

Hippotherapy can be remarkably helpful for physical therapy. When using hippotherapy as a physical therapy, the primary focus is often the patient's postural and motor responses ("History of Hippotherapy and AHA Inc.," 2010). Positive effects can be

seen particularly in motor coordination, postural alignment, strength, muscle tone, and stiffness/flexibility (Wilson & Turner, 1997). This is accomplished because the rhythmic movement of a horse provides a sense of rhythm to the patient, which encourages muscle tone and correct postural alignment ("History of Hippotherapy and AHA Inc.," 2010). In addition, the rhythmic, three-dimensional movements of the pelvis of a horse and its footfalls help reduce the formation of abnormal muscle tone of the rider ("History of Hippotherapy and AHA Inc.," 2010). All these benefits are particularly helpful for patients with ataxia from various diseases and syndromes such as multiple sclerosis, epilepsy, and cerebral palsy.

CHAPTER SIX

Possible Use of Hippotherapy for Dravet Syndrome

With a growing body of evidence supporting the therapeutic effects of hippotherapy for patients with neuromuscular disorders such as cerebral palsy and muscular dystrophy and the similarities between these disorders and Dravet Syndrome, it is reasonable to consider hippotherapy as a possible treatment option for children with Dravet Syndrome. Hippotherapy would be particularly beneficial for children with Dravet Syndrome because they suffer multiple physical side effects of seizures in early life including poor coordination difficulties with balance, and ataxia, as well as, emotional side effects related to quality of life. Hippotherapy appears to benefit patients with other epileptic disorders both physically and emotionally.

Multiple Sclerosis, for example, has been extensively studied and there is strong evidence supporting the use of Hippotherapy as a therapeutic tool for patients with Multiple Sclerosis (Wootla et al., 2012). In the same way that patients with Multiple Sclerosis cannot properly transmit signals to the target muscle due to a hardened myelin sheath, patients with Dravet Syndrome cannot transmit neuromuscular signals due to the defected sodium channel (Wootla et al., 2012). Likewise, the interruption in signal transmission in patients with MS causes seizures, which leads to the same physical side effects as those seen in Dravet Syndrome. Both types of patients suffer difficulty in transmitting signals, and both patients' conditions are worsened by multiple seizures (Wootla et al., 2012). In a study conducted in Sweden, eleven patients with Multiple Sclerosis participated in hippotherapy to see if it affects balance, gait, spasticity, functional strength, coordination, pain, self-rated level of muscle tension (SRLMT), activities of daily living (ADL), and health-related quality of life (Hammer et al., 2005). Ten of the 11 patients reported significant improvement in one or more of the variables tested, especially in balance, muscle tension, and activities of daily living (Hammer et al., 2005). The results of this study support the potential therapeutic effects of hippotherapy for individuals with Dravet Syndrome, which presents with similar physical problems due to the abnormal muscle tone or lack of tone.

Hippotherapy appears to also be helpful for children with cerebral palsy. Cerebral palsy, like MS, is similar to Dravet Syndrome in that they both manifest themselves in infancy and early childhood and are characterized by seizures, ataxia, stiff or tight muscles, and lack of coordination (NINDS, 2010). In a ten patient study, Casady and

Nichols-Larson (2004) assessed changes in general functional development and gross motor skill in children with Cerebral palsy between the ages of 2 and 7 who participated in hippotherapy. The children received hippotherapy once a week for ten weeks. The researchers found positive significant differences between the pre and post test results, demonstrating that hippotherapy was successful for all ten children with cerebral palsy (Casady & Nichols-Larson, 2004).

CHAPTER SEVEN

Conclusions and Recommendations

Hippotherapy may be a beneficial therapy for children with Dravet Syndrome, specifically improving ataxia. Researchers have found that hippotherapy positively effects ataxia in patients with multiple disorders similar to Dravet Syndrome, such as mutiple sclerosis and cerebral palsy. However, to date, there is only anecdotal evidence to support the use of hippotherapy for children with Dravet syndrome. Rudd Kierstead, a board member at Blue Rider Stables, Inc. stated this about his son and his experience with hippotherapy:

> "Willem has Dravet's Syndrome and his weekly riding lesson is a barometer for how he is doing overall. His instructors are so in tune with him, they can tell when there has been a change in his medication. The feedback I get from his instructors regarding his neurological and physical status is helpful to me as a parent. My son does not engage his core muscles in his daily life and struggles to walk or even sit up straight.

> While he is riding, I see him using his trunk muscles in order to sit up straight and tall on his horse. There is this transformation when he gets on a horse. I am a numbers guy, and very clinical in my life-these lessons just work!" (Sierau, 2013)

Additional research on the use of hippotherapy as a therapeutic tool for children with Dravet's Syndrome is both justified and needed. Based on positive anecdotal evidence and the similarities between this syndrome and CP and MS, it is reasonable to assume that children with Dravet's Syndrome will benefit from Hippothearpy and that this type of therapy could eventually be considered a standard therapeutic tool, reimbursable by health insurance. Though there are some rehabilitation centers that volunteer their services for free, these are very rare because most facilities have limited resources. Research would hopefully provide a path to future funding. The major limitation to research would be finding enough patients who are willing to participate in the study in one geographic area because Dravet Syndrome is a fairly rare disorder. However, parental support and the critical need for research in this area may provide the supported needed to move forward.

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