A Review of Spinal Muscular Atrophy

Heather Gilbreath is a 2006 graduate of the University of Texas Southwestern Medical Center Physician Assistant Program where she now serves as a Clinical Assistant Professor. She is employed at Children’s Health Dallas as a Manager within Advanced Practice Services. Clinically, Heather has cared for pediatric patients with neuromuscular disorders for over 10 years. Serving in a subspecialty population has allowed Heather to participate in multiple clinical trials, become a member of a national task force for the early identification of neuromuscular diseases and help develop a proxy motor outcome measurement for young children with neuromuscular diseases through the NIH. Other accolades include receiving the 2013 Children’s Medical Center Advanced Practice Professional Award for education and advocacy.

Spinal muscular atrophy (SMA) is one of the many diseases recognized each August in an effort to spread national awareness and education. As a physician assistant who has specialized in seeing patients with neuromuscular diseases for over 12 years, I feel that early identification of patients suffering from SMA is crucial; now more than ever given that 2 individual treatments options have been approved by the FDA.

Introduction

Spinal muscular atrophy (SMA) is an autosomal recessive disorder characterized by degeneration of spinal cord motor neurons, resulting in progressive muscular atrophy and weakness. This disease can take away the ability for those affected to walk, eat and ultimately breathe. Of note, this disorder is reported to be the leading genetic cause of infant death.

SMA is caused by mutations within the survival motor neuron gene 1 (SMN1), which is responsible for the production of the SMN protein essential to motor neurons. In SMA, insufficient levels of the SMN protein lead to degeneration of the lower motor neurons, leading to debilitating and often fatal muscle weakness.
Clinical Presentation

Three pediatric forms of the SMA exist, with a fourth type that develops in adults. SMA type 1, also known as Wernig-Hoffman disease or infantile-onset SMA, is the most severe form of the disease and accounts for approximately 50% of patients. Occasionally, there is a history of decreased intrauterine movements during pregnancy. Symptom onset is prior to 6 months of age and includes severe hypotonia, substantial muscle weakness, swallowing and feeding difficulties and impaired breathing.

SMA type 2 develops between the ages of 7 and 18 months of age. The first sign is often a delay or regression of motor milestones. Patients suffer from significant hypotonia as well as muscle weakness with the lower extremities more affected than the upper extremities. Individuals with type 2 SMA can typically sit unsupported when placed but are unable to walk independently.

SMA type 3, also known as Kugelburg-Welander disease, is typically diagnosed after 18 months of age. Individuals are initially able to walk but suffer from significant weakness leading to an increasing limitation of mobility as they grow. Most individuals eventually require the use of a wheelchair. Scoliosis can also develop.

SMA type 4 is very rare. The onset of the disease is usually in adulthood and leads to mild motor impairment. While symptoms can begin as early as the age of eighteen years old, they typically do not begin until after age 35.

Physical Examination

The physical examination in any patient with SMA has common findings including hypotonia, muscle weakness and muscle wasting but with varying degrees based on the type of SMA. Additional common findings include diminished to absent reflexes and the presence of tongue fasciculations. Infants are oftentimes noted to be lying in a frog-legged position, demonstrate severe head lag and can easily slip through the hands of a provider when held with vertical suspension. Clinicians should also look for altered breathing patterns, including paradoxical breathing. A bell-shaped chest develops with time in direct proportion to respiratory weakness. Scoliosis as well as joint contractures also develop within the clinical course.

Progression/Prognosis

Progression/prognosis varies from patient to patient and is dependent upon the clinical presentation and type of SMA. Common complications as a result of varying degrees of muscle weakness include an impaired cough, respiratory insufficiency, dysphagia, gastroparesis, constipation and evolving orthopedic issues including scoliosis. Cognitive development is typically not affected.
Treatment Options

Care guidelines have been developed addressing the need for multidisciplinary care including a neuromuscular specialist, pulmonologist, orthopedist, and gastroenterologist in efforts to provide quality care. Furthermore, patients benefit from physical, occupational and speech therapy services.

In December 2016, the FDA approved nusinersen (Spinraza) to treat children (including newborns) and adults with SMA. Nusinersen is an antisense oligonucleotide (ASO) designed to treat SMA caused by mutations in chromosome 5q that lead to SMN protein deficiency. Nusinersen was shown to increase production of full-length SMN protein.

In May 2019, the FDA also approved onasemnogene abeparvovec-xioi (Zolgensma), gene therapy using a genetically engineered virus to deliver a healthy copy of the SMN1 gene to target motor neurons aiming to subsequently restore levels of function SMN protein for patients less than 2 years of age.

Although no cure remains, treatment options like the medications described above have demonstrated improved strength ultimately altering the landscape of SMA. With treatments now available, the federal government has now added SMA to its official list of recommendations for newborn screening. Despite this, many states have not yet added statewide screening for SMA. Therefore, it is of utmost importance that pediatric providers easily recognize the clinical signs and symptoms allowing for early recognition and early treatment.

References

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SPAP Member Spotlight
Jessica Saint-Paul, PA, MPH, MCHES

Jessica Saint-Paul is a California Licensed Primary Care Physician Assistant and Public Health Practitioner. She served at Watts Healthcare Corporation as the Clinic Manager and primary clinician for ten area feeder schools providing primary care, health education and family planning services at their School-Based Health Clinic. Jessica is a strong advocate for adolescent health and medicine. She is the Executive Director for C.H.A.I.N. Reaction, Inc. and serves as the Medical Specialist Consultant for Essential Access Health. In addition, she is Adjunct Faculty with the Los Angeles Community College District in the disciplines of Public Health and Health Occupations. She earned a Master of Public Health degree from UCLA and a Bachelor of Science degree and Primary Care Physician Assistant Certificate from Howard University. She is currently a Fellow completing her doctoral degree in Medical Science at the University of Lynchburg. Most recently, she won the 2019 SPAP PA of the Year award for her hard work and dedication towards the profession.

How did your career as a PA start?

I learned about the PA profession at Howard University while researching scholarship opportunities for students interested in health careers. I saw a Supervising Physician Assistant position at a federally qualified health center in Watts, California. At first glance, I was concerned the opportunity would not be extended to me as a new PA graduate. In the meantime, I served at a Family Medicine/Urgent Care Industrial Clinic while providing health assessments for foster care youth in residential group homes. My position included on-call responsibilities during nights and weekends. I sharpened my urgent care skills and increased my scope of practice. After a year, I was offered the opportunity to serve as the Supervising Physician Assistant at the school-based health clinic and my responsibilities included working at the main site and their satellite clinic in the internal medicine, pediatrics and OB/GYN departments.

What does your average day at work look like?

The most rewarding aspect as a PA in Pediatrics in School-Based health care, is an understanding that you will encounter non-traditional days. I have administrative and clinical responsibilities that start with reviewing charts and labs. Patient encounters range from sports physical, acute injuries, vaccinations, diabetes management and family planning visits. Pursuing my doctoral degree has shifted my work schedule. I am often seen reading peer review journals which I utilize to provide technical assistance using evidence-based strategies to agencies providing family planning services. In addition, I teach distance education courses and work on developing systems to improve student
success. The best part of my day is convening with Peer Health Advocates to create innovative programs to increase awareness of school-based services.

**How did you first hear about SPAP?**

I learned about SPAP back in 2010 after being accepted as a Speaker for SPAP’s 2nd Annual CME conference in Texas. This was the first time in my PA career that I had the opportunity to collaborate with PAs who share the same passion for Pediatrics! SPAP topics were all relevant to improving my practice in Pediatrics. This was such a great complement to my state PA organization and AAPA conferences.

**Why pediatrics?**

The adolescent period is a time where healthy behaviors can be established and maintained. As a PA in Pediatrics and Public Health, I desired a position that would allow me to make significant contributions to community health through primary prevention. I believe our youth are the most deserving and impressionable patients we can serve in medicine. Adolescents are in varies stages of knowledge, attitudes and beliefs in regard to their healthcare. Providing youth and adolescents anticipatory guidance early in life can assist them and their families in making healthier lifestyle choices. It is mutually rewarding to be part of your patient’s growth from childhood to adolescence.

**What is your favorite part of being a PA?**

I appreciate the flexibility of our PA profession. As PAs, we are trained in Primary Care. I have had the opportunity to serve adolescents as a PA in medicine, education and residential care settings. In addition, due to the complexities and disparities in our health care system, PAs will always be needed to increase access to affordable quality health care.

**Any advice for new PAs or PA students?**

I would like to encourage PA students to consider serving in a School-Based Health Center (SBHC). SBHCs allow your patients to access comprehensive care in a safe and trusted environment. You will be exposed to well-child care and manage patients with acute and chronic conditions. At SBHCs you have the best setting to improve patient care and compliance. Lastly, I would advise PA colleagues to make a commitment to continuous learning and quality improvement. We are contributing to shaping the lives our pediatric patients. Get involved in leadership to prepare for Optimal Team Practice so we can advance our profession and improve patient care outcomes.