Oral Defense Announcement
University of Missouri – St. Louis Graduate School

An oral examination in defense of the dissertation for the degree
Doctorate in Nursing Practice with an emphasis area in Pediatric Nurse Practitioner

Natalie L. Goedeker
B.S. Nursing, Saint Louis University, 2012
M.S. Nursing, Saint Louis University, 2016

Predicting Outcomes of Early Treated Infants with Spinal Muscular Atrophy

Date: July 12, 2023
Time: 09:00-09:45
Place: Seton 106

Abstract
Introduction:
While prompt identification and treatment of infants with spinal muscular atrophy (SMA) can ameliorate outcomes, variability in outcomes persists. The aim of this study was to assess real-world management practices and outcomes of early-identified and treated infants with SMA.

Methods:
We performed retrospective chart review on infants with SMA treated at <=6 weeks of age at ninee medical centers between January 1, 2019 and June 1, 2023.

Results:
Sixty-five patients, 36 with two SMN2 copies, 28 with three SMN2 copies, and one with four SMN2 copies, were included. Forty-six (71%) received initial treatment with onasemnogene abeparvovec, and 19 (29%) with nusinersen. Twenty-seven (41.5%), most with two SMN2 copies, had SMA symptoms by initial treatment. Infants with two SMN2 copies had greater frequency of gross motor delays than those with >=three SMN2 copies. No children required permanent ventilation or total nutrition via tube feeds, but seven required nocturnal non-invasive ventilation and seven utilized both oral and tube feedings. Thirty-one received sequential or combination treatments, including fifteen who added treatment with nusinersen or risdiplam due to SMA symptoms or suboptimal outcome following onasemnogene abeparvovec.

Discussion:
Early identification and prompt treatment of infants with SMA can improve outcomes, as indicated in our cohort of children treated <=six weeks, all of whom are alive and without permanent ventilation. However, disease involvement persisted, particularly in infants with two SMN2 copies. Further studies on this cohort investigating feasibility, safety, and efficacy of earlier and prenatal treatment, early combination therapies, and additive non-SMN treatments are warranted.

Defense of Dissertation Committee
Charity Galgani, DNP, APRN, WHNP-BC, Committee Chair
Elise Schaller, DNP, MHA, APRN, PNP-BC
Craig Zaidman, MD