Muscular Dystrophy Association

August 23, 2018

Dr. Paul Pitel
Chair, Genetics and Newborn Screening Advisory Council
Bureau of Laboratories
Porter Auditorium
1217 N Pearl Street
Jacksonville, Florida 32231

Dear Dr. Pitel,

The Muscular Dystrophy Association (MDA) is committed to transforming the lives of individuals affected by neuromuscular disease through innovations in science and innovations in care. As an umbrella organization representing more than 40 different disorders, MDA works to promote early screening, diagnosis, and treatment for multiple diseases, including Pompe disease (acid maltase deficiency). Robust newborn screening programs are essential to this work.

MDA appreciates this opportunity to comment on the Genetics and Newborn Screening Advisory Council's decision of whether to include Pompe in Florida's newborn screening panel. MDA advocated for the inclusion of Pompe disease on the Recommended Uniform Screening Panel (RUSP) when it was considered and added in 2013, and we will continue to advocate for its inclusion on state newborn screening panels. We are proud to work collaboratively with the clinical, research, and advocate community on screening efforts around a number of different disorders and look forward to facilitating the addition of even more neuromuscular diseases to the RUSP as they are ready to meet the rigorous evidence review standards set out by the body. With Pompe currently on the RUSP, there is now greater opportunity than ever to ensure that life saving and changing therapies and care are available to newborns in Florida.

Currently, most patients and their families learn of a neuromuscular disease like Pompe after signs of disease have already begun to manifest. Following the onset of symptoms, a number of different tests may be performed, usually over a period of time, as doctors work to determine a diagnosis. This process often is referred to as the "diagnostic odyssey" and, as the term implies, can be incredibly time-consuming and emotionally draining for families during a child's first few months and/or years of life. Critically, it can also mean that a window of opportunity to earlier treat the disease may be missed. Because newborn screening identifies disorders early, and before symptoms may appear, it eliminates the diagnostic odyssey in connection with the disorders it covers. Importantly, newborn screening ensures that every child can then begin to receive the right treatment for their condition and can be connected with expert clinical care and important supports and programs as soon as possible to drive the best possible health outcomes. Such testing can also provide families with a genetic diagnosis — information that often is required to determine whether their child is eligible to participate in promising clinical trials. MDA's engagement in the clinical care of infants identified in newborn screening is paramount.

Pompe is a metabolic muscle disorder, a group of diseases that interferes with the processing of food (in this case, carbohydrates) for energy production. It is a rare, heritable disorder that causes slowly progressive weakness, especially of the respiratory muscles and those of the hips, upper legs, shoulders and upper arms. Treatment for Pompe disease exists but, as is the case with all disorders present at birth, early detection is key. In 2006, the U.S. Food and Drug Administration granted approval for the use of Myozyme as a treatment for Pompe disease. The drug was developed by Genzyme Corporation, with support from MDA. Prior to the development of treatment, the infantile form of Pompe disease was often fatal within the first year of life.

MDA has long supported the early detection and treatment of Pompe disease and other neuromuscular disorders, and we urge the Council to add Pompe to Florida's newborn screening panel. If you have any questions, please contact me at advocacy@mdausa.org.

Sincerely,

Brittany Johnson Hernandez
Director of Advocacy

cc Dusty Edwards, BSN, RN
Program Director, Florida Newborn Screening Follow-Up Program