Newborn Screening Review Abstract for World Muscle Society

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Title: A FIVE YEAR REVIEW OF NEWBORN SCREENING FOR SPINAL MUSCULAR ATROPHY IN THE STATE OF UTAH: LESSONS LEARNED

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Spinal muscular atrophy (SMA) is an autosomal recessive genetic disease characterized by degeneration and loss of alpha motor neurons in the spinal cord anterior horn, resulting in progressive symmetrical weakness, atrophy of the proximal voluntary muscles, and infant death. It is estimated that more than 95% of SMA patients present with homozygous deletion of the *SMN1* gene.

Clinical symptoms manifest in the first weeks to months of life in the most severe cases. With multiple available therapies preventing symptom development and slowing disease progression, newborn screening for SMA is essential to identify at-risk individuals. Utah was first to begin statewide newborn screening for SMA in the US.

From 2018 to 2023, a total of 239,844 infants were screened. 13 babies screened positive and were confirmed to have SMA. One of the identified patients was diagnosed prenatally and was already known to our programs. An additional case was determined to be a false positive. We are not aware of any false-negative cases. All patients were seen promptly with genetic diagnosis confirmed within 1 week of the initial clinical visit. Patients were treated with nusinersen or onasemnogene abeparvovec. Treated patients with 2 copies of *SMN2* are meeting important developmental milestones inconsistent with the natural history of type 1 SMA. Patients with 3-4 copies of *SMN2* follow normal developmental timelines.

Newborn screening is an effective tool for early identification and treatment of patients with SMA. Treatment before symptom onset results in a dramatic shift in natural history of patients with SMA, with most patients meeting appropriate developmental milestones. Identification of patients with two copies of *SMN2* identified through newborn screen constitutes a neurogenetic emergency. Due to complexities of follow up, a multidisciplinary team, including close communication with the newborn screening program, is required to facilitate diagnosis and treatment in a timely manner.