Newborn Screening for Spinal Muscular Atrophy

Testimony of Cure SMA
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Good afternoon. Dr. Bocchini and members of the Advisory Committee, thank you for the opportunity to testify today. My name is Shannon Zerzan. I am the mother of a son with Spinal Muscular Atrophy, the leading genetic cause of death for infants. Since our son's diagnosis we have worked closely with Cure SMA to raise awareness and funds to support their mission of a world without SMA. Cure SMA supports and directs comprehensive research that drives breakthroughs in treatment and care, and provides families the support they need.

On behalf of Cure SMA, my family, and thousands of other families affected by SMA, I am here to comment regarding the committee's nomination and evaluation process for candidate conditions on the recommended uniform newborn screening panel.

Newborn screening is an issue of paramount importance to the SMA community. SMA families, as well as investigators and clinicians believe that newborn screening holds great promise for ensuring access to a treatment and helping to move toward a cure for this deadly disease.

Over the last decade, there have been significant advances in the development and approval of a treatment for SMA. In fact, earlier this month we were pleased to hear that a partnership between two pharmaceutical companies has resulted in the closing of the phase 3 clinical trials of a treatment for infantile-onset SMA. We are at an exciting precipice with the potential for seeing an approved treatment for SMA with the filing of a New Drug Application to the Food & Drug Administration likely later this year.

Both human natural history data and animal model data suggest that early drug intervention allows for the greatest efficacy in SMA. Natural history data indicates that there's a small opportunity for intervention to obtain the best results in the most common and severe form of SMA type I.

Preliminary data in mouse models also indicate that pre-symptomatic drug intervention is more effective than post-symptomatic, with the results being remarkably consistent. Tests have demonstrated the best results when drugs are given as early as possible. In the most severe mouse model of SMA the efficacy of drug treatment has been shown to diminish substantially after the first week of life.

Diagnostic delay is very common in SMA. It can take weeks, months, and in milder forms of the disease even years to accurately diagnose. Most parents of children born with SMA leave the hospital with a healthy baby. Everything seems fine, until it isn't. One day your baby is moving and eating and breathing as he or she should, and then things change. One study has shown that infants with SMA type I demonstrate normal motor neuron innervation during the pre-symptomatic phase of the disease, but suffer rapid and severe loss of motor units during the first three months of life. This can result in the loss of more than 90% of motor units by six months of age.

Studies also have shown that proactive treatment such as respiratory and nutritional support administered in the first few weeks-to-months of life, prolongs survival and improves quality of life of an infant with SMA type I. Thus far, such interventions are typically only available in response to medical crises. These newborns should never have to wait to reach crisis. Newborn screening for SMA can change this.

Pre-symptomatic intervention and drug treatment is not possible without pre-symptomatic diagnosis. It is of the utmost importance that SMA be added to the recommended uniform screening panel to ensure patients and families are made aware of the disease through newborn screening, told of the need for treatment, and can obtain treatment at the earliest possible moment.

In conclusion, the SMA community strongly urges the Advisory Committee to take up consideration of the forthcoming SMA screening nomination with concerted focus on the availability of a treatment for SMA in the very near future, the success of the technology in screening for SMA, and the demonstrated benefits of early intervention. I thank the Committee for the opportunity to address you today and appreciate your consideration of our views.