

Consensus Statement in Support of Duchenne Newborn Screening

We, as experts in the fields of genetics, pediatrics, neurology, and public health, hereby endorse the implementation of Duchenne muscular dystrophy (DMD) newborn screening as an essential component of comprehensive healthcare for infants. DMD is a severe and progressive genetic disorder that affects approximately 1 in 5,000 live male births, making it one of the most common and devastating childhood neuromuscular diseases.

After careful consideration of the available scientific evidence, we acknowledge the following key points supporting the implementation of Duchenne Newborn Screening:

Timely Intervention and Treatment Initiatives:

Duchenne is characterized by a progressive loss of muscle function, with symptoms typically appearing in early childhood. Timely diagnosis through newborn screening provides a critical window for intervention and initiation of therapeutic strategies before irreversible muscle damage occurs.

Timely treatment, including early intervention services that address delays in speech, motor, and cognitive development, as well as emerging genetic and pharmacological therapies, have shown promise in slowing disease progression and improving outcomes for individuals with Duchenne. Newborn screening facilitates timely access to these interventions, maximizing their potential benefits.

Additionally, families of babies identified with Duchenne through newborn screening benefit from genetic counseling resources, as well as opportunities for emotional and psychological support, allowing for informed decision-making and proactive healthcare planning.

Reduced Diagnostic Delays and Avoidable Diagnostic Costs:

Newborn screening eliminates the diagnostic odyssey that many families currently face, reducing the time from symptom onset to diagnosis. While medical costs are unavoidable in Duchenne, the avoidable costs attributable to delayed diagnosis represent the burden on patients and families searching for a diagnosis. Delayed diagnosis also results in healthcare dollars that could be better spent on treatment and supportive therapies that improve patient quality of life and may even increase workforce productivity for families. Recently, a study of the avoidable costs in the Duchenne diagnostic odyssey were \$211, 229 per family and averaged 20 days of missed work per year for a parent.

Public Health Impact:

Implementing Duchenne Newborn Screening contributes to the broader public health goal of early detection and intervention for genetic conditions. It aligns with the principles of preventive medicine and represents a proactive approach to managing a significant healthcare challenge.

Ethical Considerations:

The benefits of early diagnosis and intervention, coupled with the minimal invasiveness of newborn screening, support the ethical justification for implementing Duchenne Newborn Screening. Informed consent and appropriate counseling must be integral components of the screening process, ensuring respect for autonomy and shared decision-making.

Based on the accumulating evidence, we strongly advocate for the incorporation of DMD into newborn screening programs. Timely identification of affected individuals is a crucial step towards eliminating unnecessary and avoidable diagnostic odyssey, improving patient outcomes, and enhancing the quality of life for children with Duchenne. We call upon policymakers, healthcare providers, and advocacy groups to work collaboratively in implementing and optimizing Duchenne Newborn Screening programs to ensure their effectiveness and accessibility on a national scale.

Signed,

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