Newborn Screening for Angelman Syndrome

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The average age of diagnosis for children with Angelman syndrome (AS) is estimated to be around 3 years of age in the United States. As exciting new therapeutics are being developed and tested for individuals with AS, the possibility of improving outcomes for patients and their families is a real possibility. However, our knowledge of brain and central nervous system development suggests that in order to maximize outcomes as a result of any therapeutic, very early implementation is likely to be best, requiring a focus on reducing the age of diagnosis in AS.

Newborn screening is a public health program designed to identify infants with conditions for which very early treatment has known and critical benefit. Screening is done on a blood spot taken from the newborn shortly after birth. The process for nominating a condition for NBS is formal and requires extensive evidence that the condition meets predetermined criteria. Therefore, even though AS does not currently meet the criteria for a nomination to the Recommended Universal Screening Panel (RUSP), it is not too early for the community to begin thinking and planning for the possibility of NBS for AS in the future.

This presentation will review the potential benefits of earlier identification of AS, describe the current criteria for the RUSP in the United States, and outline steps the AS community can be taking to prepare for a possible inclusion of AS on NBS panels. The presentation will also provide an overview of an ongoing voluntary expanded NBS study, called Early Check, which is currently being implemented in North Carolina. Early Check is designed to provide an opportunity to help prove benefit of newborn screening for conditions that do not meet eligibility for the RUSP. The possibly inclusion of AS on the Early Check panel is currently being considered; this presentation will provide an avenue for discussion of the benefits and potential challenges for new families receiving a diagnosis of AS in their newborn.