







Newborn Screening Technical Advisory Committee (TAC)

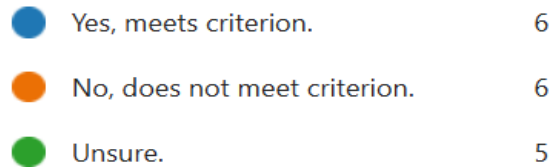
Meeting to Review Branch-Chain Ketoacid Dehydrogenase Kinase (BCKDK) Deficiency for the Newborn Screening Panel

TAC Member Voting Summaries and Comments

The following is a compilation of comments from TAC members provided when voting on each individual criteria, and an overall recommendation. Comments have been summarized and are organized by each criterion and then overall comments provided.

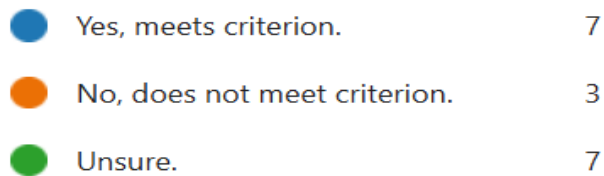
Criteria	Major themes
<p>1. Available Screening Technology</p> <p> Yes, meets criterion. 6</p> <p> No, does not meet criterion. 7</p> <p> Unsure. 4</p> 	<ul style="list-style-type: none"> Tests and technology are available for measuring BCA serum levels, but their performance, sensitivity, and specificity are unclear. While the upper limits of normal BCA levels are defined, lower limits can be estimated from population norms, and tandem mass spectrometry is already used to directly measure BCA plasma levels.

2. Diagnostic Testing and Treatment Available





- There is very limited evidence available for this disorder, making it unclear whether diagnostic criteria are met.
- The data on prevalence, long-term outcomes, false positives/negatives, and treatment effectiveness is insufficient, and the small sample size makes it difficult to verify the disorder's validity.

3. Prevention Potential and Medical Rationale



- There is a lack of sufficient data on the prevalence, long-term outcomes with early treatment, and the number of patients in the literature, making it difficult to assess the relevant criteria.

<p>4. Public Health Rationale</p> <ul style="list-style-type: none"> ● Yes, meets criterion. 2 ● No, does not meet criterion. 12 ● Unsure. 3 	<ul style="list-style-type: none"> Not enough information to assess this criterion. Rarity gives pause, but true prevalence is unknown.
<p>5. Cost Benefit / Cost Effectiveness</p> <ul style="list-style-type: none"> ● Yes, meets criterion. 0 ● No, does not meet criterion. 13 ● Unsure. 4 	<ul style="list-style-type: none"> There is insufficient data available to evaluate the condition, including the lack of BCA testing, limited prevalence information, and only 21 patients reported in the literature. Screening is not being conducted, and there are concerns about unintended consequences for conditions on the newborn screening panel.

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