

**SACHDNC Form for Nomination of a Condition for Inclusion in the Uniform Screening Panel**

<b>DATE</b>	
<b>NAME OF NOMINATOR AND ORGANIZATION</b> (include professional degrees)	<b>INDICATE AFFILIATION</b> (i.e., Health Professional, Subject Matter Expert, Researcher, Clinician, Advocate, etc.)
<b>CO-SPONSORING ORGANIZATIONS</b> (include professional degrees)	<b>INDICATE AFFILIATION</b> (i.e., Health Professional, Subject Matter Expert, Researcher, Clinician, Advocate, etc.)

*\*Note: Please reference each statement/answer with the corresponding reference number listed in Section III – Key References.*

**SECTION I – CONDITION INFORMATION AND TREATMENT**

**SECTION I, PART A**

<b>CONDITION</b>	<b>STATEMENT</b>
<b>Nominated Condition</b>	
<b>Type of Disorder</b>	
<b>Screening Method</b>	
<b>Gene</b>	
<b>Locus</b>	Include ClinVar link if applicable.
<b>OMIM or other names for condition</b>	Include Genetics Home Reference link if applicable.
<b>Case Definition</b>	
<b>Incidence</b>	Determined by what method(s): pilot screening or clinical identification?
<b>Timing of Clinical Onset</b>	Relevance of the timing of newborn screening to onset of clinical manifestations.
<b>Severity of Disease</b>	Morbidity, disability, mortality, spectrum of severity.

**SECTION I, PART B**

<b>TREATMENT</b>	<b>STATEMENT</b>
<b>Modality</b>	Drug(s), diet, replacement therapy, transplant, other. Include information re regulatory status of treatment.
<b>Urgency</b>	How soon after birth must treatment be initiated to be effective?
<b>Efficacy (Benefits)</b>	Extent of prevention of mortality, morbidity, disability. Treatment limitations, such as difficulty with acceptance or adherence.
<b>Availability</b>	Limits of availability?
<b>Potential Harms of Treatment</b>	Potential medical or other ill effects from treatment

**SECTION II – EVIDENCE-BASED INFORMATION**

**For a nominated condition to be considered there are 3 core requirements:**

1. Validation of the laboratory test (see Section II, Part A)
2. Widely available confirmatory testing with a sensitive and specific diagnostic test (see Section II, Part B)
3. A prospective population based pilot study (see Section II, Part C)

**SECTION II, PART A**

<b>TEST</b>	<b>STATEMENT</b>
<b>Screening test(s) to be used</b>	Description of the high volume method, instrumentation and if available as part of multi-analyte platform.
<b>Modality of Screening</b>	(Dried blood spot, physical or physiologic assessment, other)
<b>Does the screening algorithm include a second tier test? If so, what type of test and availability?</b>	(Dried blood spot, physical or physiologic assessment, other)

TEST	STATEMENT
<b>Clinical Validation</b>	Location, duration, size, preliminary results of past/ongoing pilot study for clinical validation, positive predictive value, false positive rate, analytical specificity, sensitivity.
<b>Analytical Validation</b>	Limit of detection/quantitation, detection rate, reportable range of test results, reference range. Include regulatory status of test, information about reference samples and controls required for testing and availability of or potential for external quality assurance system, e.g., QC and PT for both screening and confirmatory tests.
<b>Considerations of Screening and Diagnostic Testing</b>	False positives, carrier detection, invasiveness of method, other.
<b>Potential Secondary Findings</b>	Detection or suggestion of other disorders.

**SECTION II, PART B**

CONFIRMATORY TESTING	STATEMENT
<b>Clinical and Analytical Validity</b>	Quantitative or qualitative? Include sensitivity, specificity, etc.
<b>Type of test and/or sample matrix (blood, radiology, urine, tissue sample, biophysical test)</b>	
<b>Is test FDA cleared/approved</b>	Include availability information, sole source manufacturer, etc.
<b>List all CLIA certified labs offering testing in the US</b>	Link to GeneTests and Genetic Test Reference if applicable.

**SECTION II, PART C**

<b>POPULATION-BASED PILOT STUDY</b>	<b>STATEMENT</b>
<b>Location of Prospective Pilot</b>	
<b>Number of Newborns Screened</b>	
<b>Number of Screen Positive Results</b>	Positive by primary test vs. 2 <sup>nd</sup> tier test if applicable.
<b>False Positive Rate; False Negative Rate (if known)</b>	False positive by primary test vs. 2 <sup>nd</sup> tier test if applicable.
<b>Number of Infants Confirmed with Diagnosis</b>	How is diagnosis confirmed [clinical, biochemical, molecular]?

### **SECTION III – KEY REFERENCES**

#### **LIST OF REFERENCES**

**Limited to 20 references from scientific journals to support statements in Sections I-IV. For sources based on un/non-published data, references may be written statements from clinicians, researchers, and/or investigators.**

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<b>SUBMISSION CHECK LIST</b>		<b>SUBMIT NOMINATIONS ELECTRONICALLY TO:</b> Sara Copeland, MD SACHDNC Designated Federal Officer Genetics Services Branch Division of Services for Children with Special Health Needs, Maternal and Child Health Bureau, HRSA 5600 Fishers Lane, Room 18A-19, Rockville, MD 20857 Email: <a href="mailto:Screening@hrsa.hhs.gov">Screening@hrsa.hhs.gov</a> Fax: 301-480-1312 Phone: 301-443-1080
<input type="checkbox"/>	Cover letter by Nominator	
<input type="checkbox"/>	Nomination form	
<input type="checkbox"/>	Conflict of Interest Forms filled out by Nominator and all Co-Sponsoring Organizations	
<input type="checkbox"/>	Copies of publications/articles used as references	
<b>CONTACT INFORMATION FOR NOMINATOR:</b>		