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

DATE:

NAME OF NOMINATOR AND ORGANIZATION (include professional degrees)	INDICATE AFFILIATION (i.e., Health Professional, Subject Matter Expert, Researcher, Clinician, Advocate, etc.)

CO-SPONSORING ORGANIZATIONS (include professional degrees)	INDICATE AFFILIATION (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

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

SECTION I, PART B

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

TEST	STATEMENT
Screening test(s) to be used	Description of the high volume method, instrumentation and if available as part of multi-analyte platform.
Modality of Screening	(Dried blood spot, physical or physiologic assessment, other)

TEST	STATEMENT
Does the screening algorithm include a second tier test? If so, what type of test and availability?	(Dried blood spot, physical or physiologic assessment, other)
Clinical Validation	Location, duration, size, preliminary results of past/ongoing pilot study for clinical validation, positive predictive value, false positive rate, analytical specificity, sensitivity.
Analytical Validation	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.
Considerations of Screening and Diagnostic Testing	False positives, carrier detection, invasiveness of method, other.
Potential Secondary Findings	Detection or suggestion of other disorders.

SECTION II, PART B

CONFIRMATORY TESTING	STATEMENT
Clinical and Analytical Validity	Quantitative or qualitative? Include sensitivity, specificity, etc.
Type of test and/or sample matrix (blood, radiology, urine, tissue sample, biophysical test)	

CONFIRMATORY TESTING	STATEMENT
Is test FDA cleared/approved	Include availability information, sole source manufacturer, etc.
List all CLIA certified labs offering testing in the US	Link to GeneTests and Genetic Test Reference if applicable.

SECTION II, PART C

POPULATION-BASED PILOT STUDY	STATEMENT
Location of Prospective Pilot	
Number of Newborns Screened	
Number of Screen Positive Results	Positive by primary test vs. 2nd tier test if applicable.
False Positive Rate; False Negative Rate (if known)	False positive by primary test vs. 2nd tier test if applicable.
Number of Infants Confirmed with Diagnosis	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.**	
1	
2	
3	
4	
5	
6	
7	
8	
9	
10	
11	
12	
13	
14	
15	
16	
17	
18	
19	
20	

X	SUBMISSION CHECK LIST	SUBMIT NOMINATIONS ELECTRONICALLY TO:
☐	Cover letter by Nominator	Email: ACHDNC@hrsa.gov
☐	Nomination form	
☐	Conflict of Interest Forms filled out by Nominator and all Co-Sponsoring Organizations	Designated Federal Official Genetic Services Branch Division of Services for Children with Special Health Needs Maternal and Child Health Bureau Health Resources and Services Administration
☐	Copies of publications/articles used as references	

CONTACT INFORMATION FOR NOMINATOR: