Committee Business

Section I, Part A. Condition

Condition	Statement
Nominated Condition	
Type of Disorder	
Screening Method	
Gene	**If applicable, if not N/A**
~~**Enzyme**~~ **Critical biomarker**	**If applicable, if not N/A**
Locus	Include ClinVar link if applicable.
OMIM or other names for condition	Include Genetics Home Reference link if applicable.
Case Definition	Include the specific case definition for the screening target.
Incidence	**Include U.S. incidence estimate and citation**. Determined by what method(s): pilot screening or clinical identification?
Timing of	Relevance of the timing of newborn screening to onset of clinical manifestations **for phenotypes that would be detected**.

Committee Business

SECTION I, PART A. CONDITION

CONDITION	STATEMENT
Clinical Onset	
Severity of Disease	Morbidity, disability, mortality, spectrum of severity. **Include U.S. distribution/prevalence of known phenotypes if ~~applicable~~ known.**

Committee Business

SECTION II, PART A. VALIDATION OF THE LABORATORY TEST

TEST	STATEMENT
Screening Test(s), Platform, and Procedures	Description of the high volume method **(number of samples run in high-throughput?), instrumentation (e.g., tandem mass spectrometry, digital microfluidics, other) and if available as part of multi-analyte platform. Disposables - Lab-based analysis or off-the-shelf (OTS) kits? If OTS kits, FDA ~~approved~~ cleared or authorized (provide FDA submission number if applicable)? Vendors/suppliers if known?**

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SECTION II, PART A. VALIDATION OF THE LABORATORY TEST

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., quality control (QC) and proficiency testing (PT) for both screening and confirmatory tests. **Has the CDC's Newborn Screening and Molecular Biology Branch (https://www.cdc.gov/nceh/dls/nsmbb.html) been contacted regarding these and are other validation measures currently pending or available?**

Committee Business

SECTION II, PART B. CONFIRMATORY TESTING AND SHORT-TERM FOLLOW-UP/DIAGNOSIS

Regulatory Status of Confirmatory Testing	Is test FDA ~~cleared/approved~~ cleared or authorized? If so, include ~~year/reference~~ FDA submission number. Describe availability of confirmatory testing, information, sole source manufacturer, specialized testing centers, etc.