ACHDNC
Advisory Committee on Heritable Disorders in Newborns and Children

**Summary of Nomination Requirements and Key Considerations**

**Three Core Requirements for a nominated condition:**

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

**Key Considerations:**

1. Is the condition medically serious?
2. Is the case definition and the spectrum of the disorder well described, to help predict the phenotypic range of those children who will be identified based on population-based screening?
3. Are there prospective pilot data* (U.S. and/or international) from population-based assessment available for this disorder?
4. Does the screening test have established analytic validation?
5. Are the characteristics of the screening test(s) reasonable for the newborn screening system (among other aspects, a low rate of false negatives)?
6. Are those who are most likely to benefit from treatment identifiable (especially if the treatment is onerous or risky)?
7. Is there a widely available and CLIA and/or FDA approved confirmatory test/diagnostic process?
8. Are there defined treatment protocols, FDA approved drugs (if applicable) and treatment available?

***Pilot Study Requirements:**

1. Data should be available from pilot studies involving population-based screening of identifiable newborns.

a. The study should evaluate the newborn screening process from collection through diagnosis and identify at least one screen-positive newborn with confirmation of presence of the condition under consideration.

b. The population included in the pilot study, and the screening protocol used, should be similar to the U.S. population and to state newborn screening programs with respect to known prevalence of the condition, and the timing and approach to screening. The screening modality used in the pilot study should be comparable to the method proposed in the application.

2. Data should be available on the analytical validation of one or more screening modalities proposed for use in population-based screening in newborns.
3. Data should be available on the net benefits of clinical interventions following early detection compared to clinical diagnosis.