

**CONSIDERING DISORDERS
FOR NBS:**
Criteria Work Group Report of the
Nominating and review process

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2-14-06 ACHDGDNC

Nomination work group (10/05-2/06)

- 6 ACHDGDNC members, appointed by chair (Dr. Howell):
 - Amy Brower
 - Coleen Boyle
 - Peter Coggins
 - Denise Dougherty
 - Nancy Green
 - Piero Rinaldo

HRSA staff: Michele Puryear, Marie Mann

3 proposed steps for nominations

- Step #1: The Nomination Form
- Step #2: Federal administrative review
- Step #3: Review by ACHDGDNC
 - A) Evidence-based review (EBR) - external
 - B) Committee review

Nomination Process - concepts

- Broad access to the nomination process
- Considered review
- Streamlined processes
- Transparency
- Consistent criteria throughout the nomination process, including EBR
- 3 broad areas identified:
 - Condition, Test, Treatment
 - References



Nomination Form (DRAFT)

Heading

Condition:

- Incidence

- Timing

- Severity

HRSA/ACMG UNIFORM PANEL (DRAFT 01/21/06)							
NOMINATION OF CONDITION - Fact Sheet							
Name of proponent					Date		
Condition							
Type of disorder							
Screening method							
Treatment strategy							
CONDITION	Comment	Gene		Locus		OMIM	
Incidence	(Reference required; By 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)						

Test:

- Screening

- Modality

- Clinical validation

- Lab performance

- Confirmatory

- Risks

TEST	Comment
Screening test(s) to be used	(High volume method, platform)
Modality of screening	(Dried blood spot, physical or physiologic assessment, other)
Clinical validation	(Location, duration, size, preliminary results of past/ongoing pilot study for clinical validation)
Laboratory performance metrics	(Sensitivity, specificity, detection rate, positive predictive value, false positive rate)
Confirmatory (diagnostic) testing	(Reliability, availability)
Risks	(False positives, carrier detection, invasiveness of method, other)

Nomination Form - DRAFT

Treatment:

- Modality
- Urgency
- Efficacy
- Availability
- Risks

Nomination of condition (page 2)	
TREATMENT	Comment
Modality	(Drug(s), diet, replacement therapy, transplant, other)
Urgency	(How soon after birth treatment needs to be initiated to be effective)
Efficacy	(Extent of prevention of mortality, morbidity, disability)
Availability	(Any limits of availability)
Risks	(Potential medical or other ill effects from treatment)

References and submission information

KEY REFERENCES (Specific citations - limit to 15)		Submit nomination to:	
1		Michele A. Lloyd-Puryear, M.D., Ph.D. Chief, Genetic Services Branch Division of Services for Children with Special Health Needs Maternal and Child Health Bureau 5600 Fishers Lane, Rm 18-A-19 Rockville, MD 20857 301-443-8604-fax 301-443-1080-phone	
2		Submission check list <input type="checkbox"/> Cover letter by proponent <input type="checkbox"/> Nomination form <input type="checkbox"/> Copy of references listed on this form	
3			
4			
5		Contact information (proponent)	
6		REFERENCES (continued) 12 13 14 15	
7			
8			
9			
10			
11			

Nomination Step #2: Federal administrative review of NF

- Administrative step - completion and clarification of NF
- Composition needs to be determined:
 - HRSA +/- other agencies
- Outcomes:
 - (a) Request additional information for the form; or
 - (b) Forward the NF to the AC for consideration.
- Would not reject a NF nor evaluate the quality of the data
- Transparent - Document all relevant correspondence for committee access

Step #3: ACHDGDNC Committee Review

- Not yet delineated
- Requires external expertise
- Transparency
- Options for ACHDGDNC:
 - Expedited review
 - Request more information from the Nominator(s)
 - EBR (Step #3A) prior to full committee consideration
 - Additional steps?



Next Tasks

- Step #1 - Finalize Nomination Form
 - Input from ACHDGDNC
 - Pilot to potential users
 - Modify and ratify by ACHDGDNC
- Step #2 - Clarify federal review
- Step #3 - Delineate processes:
 - EBR
 - ACHDGDNC, other?