A Process for the Addition of New Conditions to the NBS Uniform Panel: A <u>Tentative</u> Outline

ACHDGDNC Washington, DC October 20, 2005

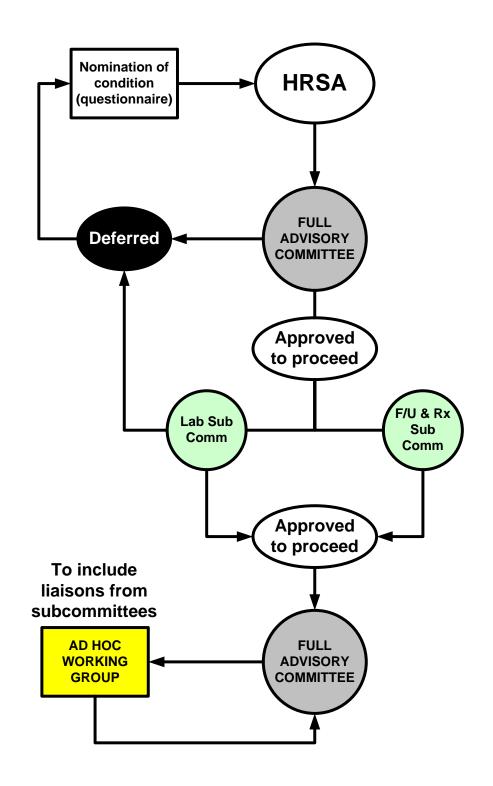
Assumptions

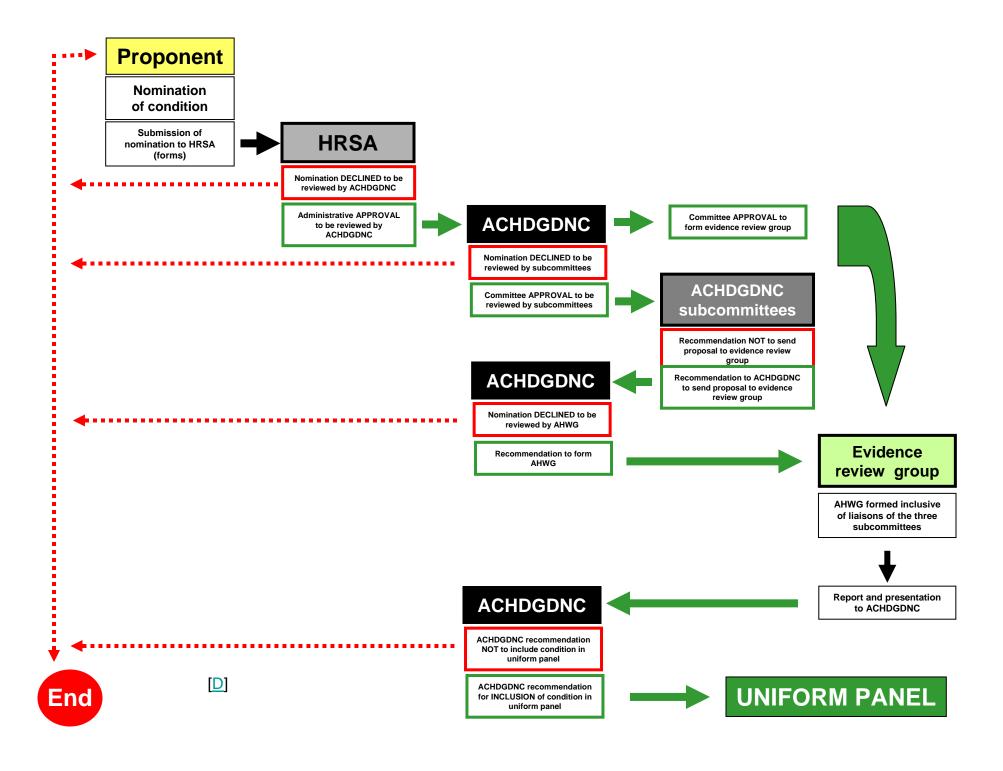
- Relatively simple nomination process
- Assessment should have similarity to the one used by the expert group to establish the uniform panel (fairness and consistency)
 - Reliance on established criteria (ACMG, others)
 - Utilization of similar tools
- Approval through progressive steps
- Formation of ad hoc working group with representation of each subcommittee
- Final recommendation for inclusion/deferral rests with the full committee

ACHDGDNC April 21, 2005

Laboratory Subcommittee

"Loopy" process, no clear beginning or end point





Proponent

Nomination of condition

Submission of nomination to HRSA (forms)

Proponent

- Provider of NBS services
- Representative of professional organization
- Representative of patient support group
- Clinician, scientist
- Industry, for-profit organization
- Patient, family member, advocate
- Other, not listed above

Nomination Requirements (April 2005)

- Questionnaire (unspecified)
- "Forms" relative to
 - Screening test
 - Treatment options

Nomination Requirements (October 2005)

- Cover letter
- Score card (UP criteria)
- Fact sheet
- References (with limit)

Nomination Requirements Score Card

HRSA/ACMG UNIFORM PANEL

NOMINATION OF CONDITION - Score Card

This form is to be filled as a requirement for the evaluation by HRSA, ACHDGDNC, its subcommittees, and ad hoc working groups of conditions not included in the 2005 HRSA/ACMG uniform panel

Proponent:		
NAME	Phone	
AFFILIATION	Fax	
DATE	E-mail	
ADDRESS		

	CHECK THE CATEGORY THAT BEST APPLIES TO YOU					
Γ	Provider of Screening Services	Г	Clinician, solentist			
Г	Representative of professional organization	Г	Industry, for-profit organization			
Γ	Representative of patient support organization	Г	Patient, family member, advocate			

This nomination form includes:

- This cover page
- 2 A list of CRITERIA and SCORES
- A worksheet listing NBS REFERENCE CONDITIONS. Scoring these conditions is encouraged to self-assess how the proponent's scores compare with the scores derived from the HRSA/ACMG survey (listed at the top)
- A blank worksheets where to list and score the condition(s) being proposed for inclusion in the uniform panel

When applicable, please provide the name of the deficient enzyme and the OMIM number in addition to the common name of the disorder

For each criterion, enter one of the acores provided A BLANK means ZERO

Submit form by mall or fax to HRSA at the address listed below

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



CRITERIA	CATEGORIES	SCORE
	>1.5,000	100
	>1:25,000	75
Incidence of condition	>1:50,000	50
	>1:75,000	25
	<1:100,000	0
	Never	100
Sign & Symptoms clinically	<25% of cases	75
Identifiable in the first 48 hours	<50% of cases	50
identinable in the mot 40 nours	<75% of cases	25
	Always	0
Burden of disease	Profound	100
Duluell of disease	Severe	75
	Moderate	50
(Natural Hx if untreated)	Mid	25
	Minimal	0
Does a sensitive AND specific screening test	YES	200
currently exist?	NO NO	0
	Doable in neonatal blood spots OR by a simple, in-nursery physical method	100
	High throughput (>200/day/FTE)	50
Test characteristics	Overall analytical cost <1\$ per test per condition	50
(Yes = apply score; No = zero)	Multiple analytes relevant to one condition are detected in same run	50
(,	Other conditions identified by same analytes	50
	Multiple conditions detected by same test (multiplex platform)	200
	Treatment exists and is widely available in most communities	50
Availability of treatment	Treatment exists but availability is limited	25
,	No treatment available or necessary	
a - 1 - 1 1 1	Inexpensive	50
Cost of treatment	Expensive (>\$50,000/patient/year)	
	To prevent ALL negative consequences	200
Potential efficacy of existing	To prevent MOST negative consequences	100
treatment	To prevent SOME negative consequences	50
	Treatment efficacy not proven	0
	Clear scientific evidence that early intervention resulting from screening optimizes outcome	200
Benefits of early intervention	Some admittide evidence that early intervention resulting from screening optimizes outcome	100
(INDIVIDUAL OUTCOME)	No adentific evidence that early intervention resulting from screening optimizes outcome	
Benefits of early identification	Early identification provides clear benefits to family and society (education, understanding prevalence and natural history, cost effectiveness)	100
(FAMILY & SOCIETY)	Early identification provides some benefits to family and society	50
	No evidence of benefits	
Early diagnosis and treatment	YES	100
prevent mortality	NO NO	0
Availability of diamonts	Providers of diagnostic confirmation are widely available	100
Availability of diagnostic	Limited availability of providers of diagnostic confirmation	50
confirmation	Diagnostic confirmation is available only in a few centers	
	Providers of acute management are widely available	100
Acute management	Limited availability of providers of acute management.	50
•	Acute management is available only in a few centers	
	Management at the primary care or family level	200
Simplicity of therapy	Requires periodic involvement of a specialist	100
	Requires regular involvement of a specialist	

Score Card (2)

UNIFORM PANEL				Hemoglobin 8	21 hydrocylaan			
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CONDITION(S) UNDER EVALUATION							
Proponent:							
Date:							
Criteria		27.00	•				
	H 5,000	100					
Incidence of	F128,000	79					
condition	>100,000	66 26					
	120	-					
	None	100					
Signs & Symptoms	-CEPIC of cases	79					
clinically identifiable	-SIPS of saves	-					
in the first 48 hours	-CTPN of sames	25					
Burden of disease if	Minays Professional	100					
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Nomination Requirements Fact Sheet

HR\$A/ACMG UNIFORM PANEL NOMINATION OF CONDITION - Fact Sheet Date Proponent Condition Type of disorder Screening method OMIM Gene Locus Incidence **Condition** include typical ag Phenotype at birth Burden If untreated .ocation, duration, size, and preliminary results of past/ongoing pilot study must be provided) Screening test & PILOT STUDY (describe major characteristics) Doable in DBS or by physical method Cases/FTE/day or cases/instrument/day, as applicable) Estimated dally throughput (actual cost and estimated testing fee) Cost of test (Fapplicable) Multiple **Test** analytes/ markers (Fapplicable) Secondary targets (other \square conditions) (Fapplicable) Multiplex platform

	Nomination of condition (page 2)
TREATMENT	Comment
Availability & cost	
Efficacy of treatment	
Benefits of early Intervention	
Benefits of early Identification	Treatment
Prevention of mortality	
Confirmation of diagnosis	
Acute management	
Simplicity of therapy	
	Michele A. Lioyd-Punyear, M.D., Ph.D. Chief, Genetic Services Franch on of Services for Children with Special Health Needs Maternal and Child Health Bureau S600 Fishers Lane. Rm 18-A-19 Rockville, MD 20857
3	301-443-9604-fax 301-443-1080-phone
4	Checklist

To Be Determined

- How to get the word out (call for proposals)
- Decision making process of full committee and subcommittees (coordination)
 - How to resolve disagreement between subcommittees
- Ad hoc working groups
 - Selection process
 - Size
 - Timeline
 - Selection of subcommittee liaisons
- HRSA implementation process of any recommendation to include additional conditions (rolling additions, annually)