

**A Process for the Addition
of New Conditions
to the NBS Uniform Panel:
A Tentative Outline**

**ACHDGDNC
Washington, DC
October 20, 2005**

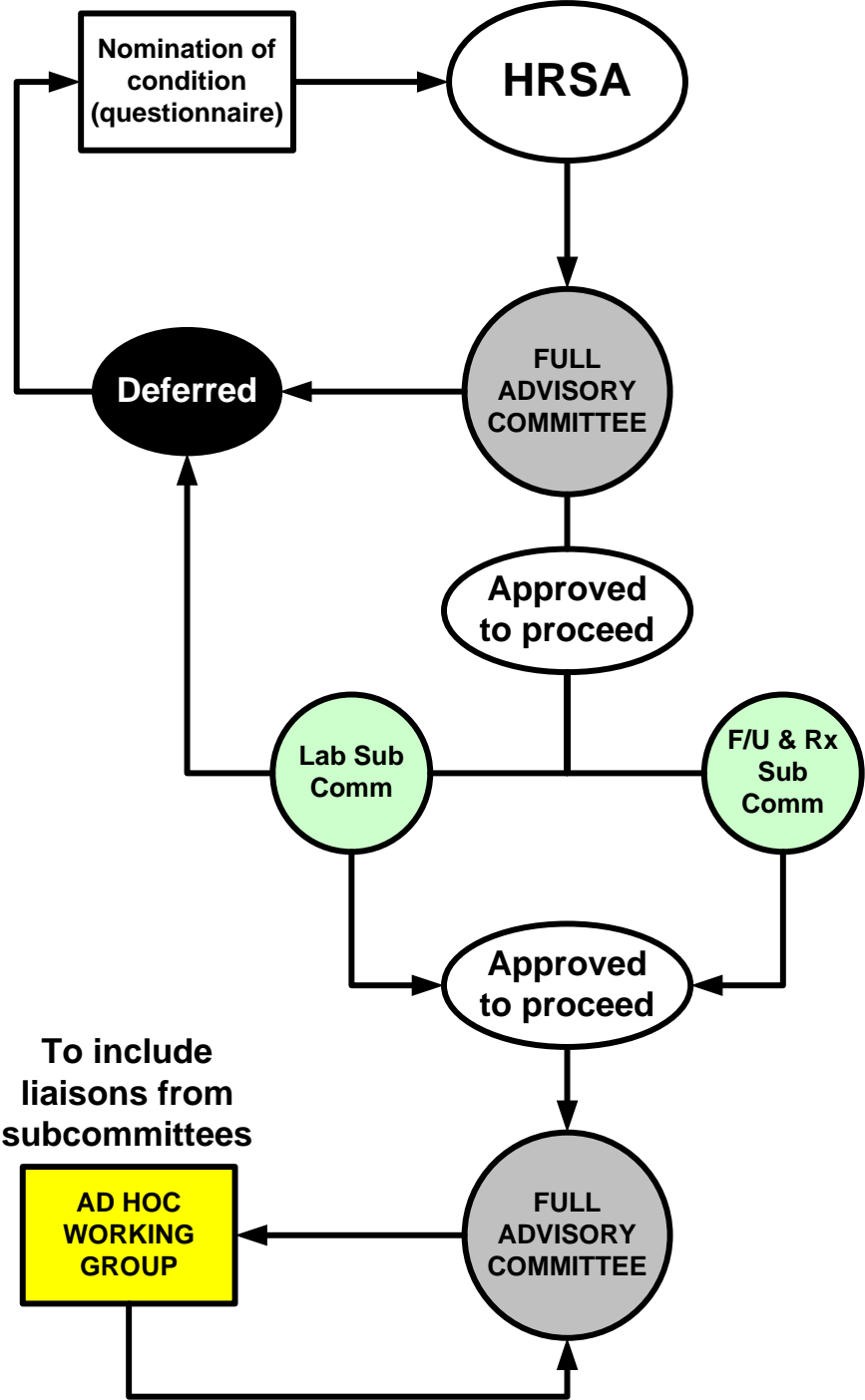
Assumptions

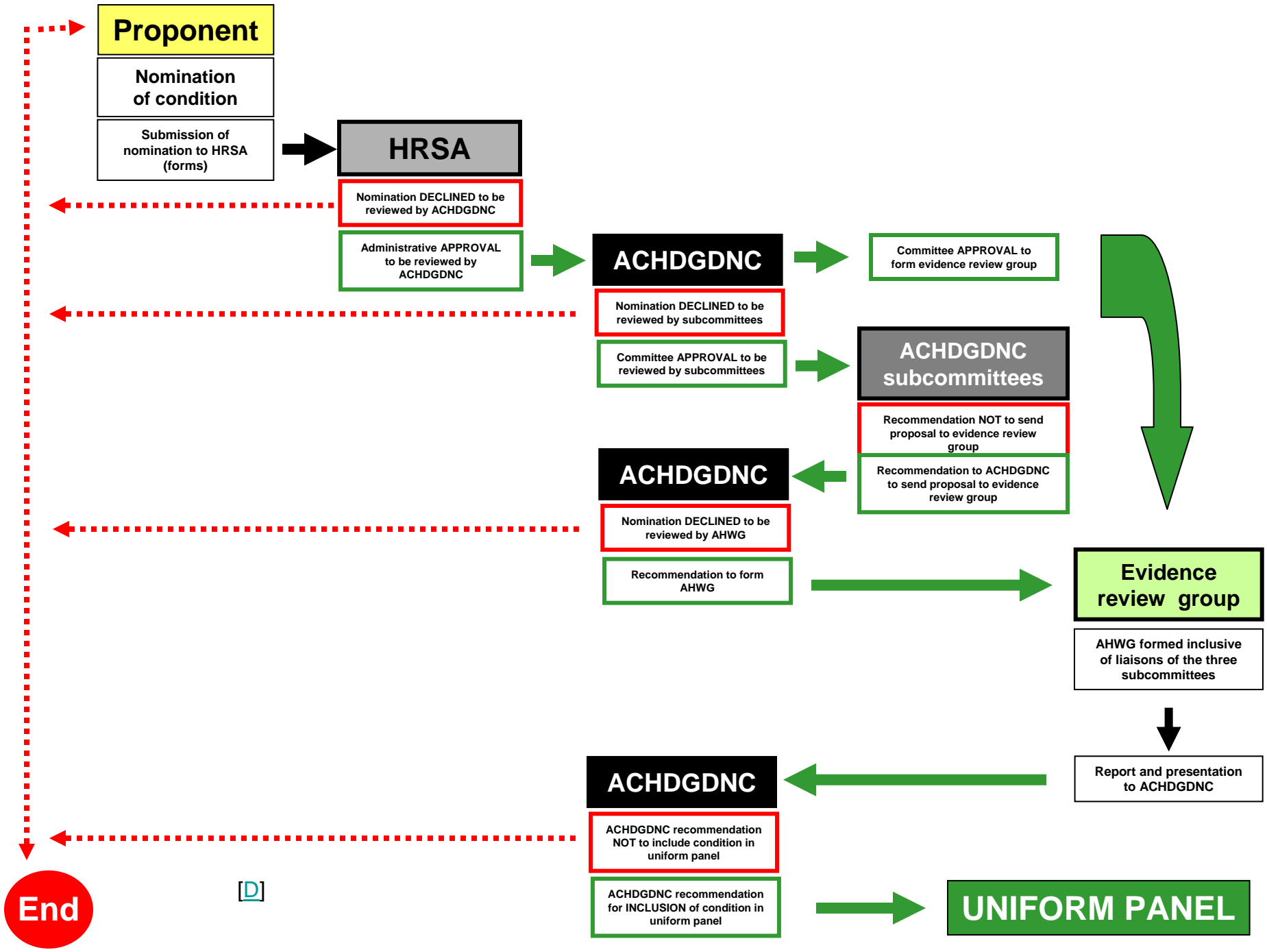
- **Relatively simple nomination process**
- **Assessment should have similarity to the one used by the expert group to establish the uniform panel (fairness and consistency)**
 - **Reliance on established criteria (ACMG, others)**
 - **Utilization of similar tools**
- **Approval through progressive 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**





End

[D]

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

<input type="checkbox"/> Provider of Screening Services	<input type="checkbox"/> Clinician, scientist
<input type="checkbox"/> Representative of professional organization	<input type="checkbox"/> Industry, for-profit organization
<input type="checkbox"/> Representative of patient support organization	<input type="checkbox"/> Patient, family member, advocate

This nomination form includes:

- 1 This cover page
- 2 A list of CRITERIA and SCORES
- 3 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)
- 4 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 scores provided
A BLANK means ZERO**

Submit form by mail 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-13
 Rockville, MD 20857
 301-443-8604-fax
 301-443-1080-phone

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CRITERIA	CATEGORIES	SCORE
Incidence of condition	>1.5,000	100
	>1.25,000	75
	>1.50,000	50
	>1.75,000	25
	<1,100,000	0
Sign & Symptoms clinically identifiable in the first 48 hours	Never	100
	<25% of cases	75
	<50% of cases	50
	<75% of cases	25
	Always	0
Burden of disease (Natural Hx if untreated)	Profound	100
	Severe	75
	Moderate	50
	Mild	25
	Minimal	0
Does a sensitive AND specific screening test currently exist?	YES	200
	NO	0
Test characteristics (Yes = apply score; No = zero)	Double in neonatal blood spots OR by a simple, in-nursery physical method	100
	High throughput (>200/day/FTE)	50
	Overall analytical cost <1\$ per test per condition	50
	Multiple analytes relevant to one condition are detected in same run	50
	Other conditions identified by same analytes	50
Availability of treatment	Multiple conditions detected by same test (multiple platform)	200
	Treatment exists and is widely available in most communities	50
	Treatment exists but availability is limited	25
Cost of treatment	No treatment available or necessary	0
	Inexpensive	50
	Expensive (>\$50,000/patient/year)	0
Potential efficacy of existing treatment	To prevent ALL negative consequences	200
	To prevent MOST negative consequences	100
	To prevent SOME negative consequences	50
	Treatment efficacy not proven	0
Benefits of early intervention (INDIVIDUAL OUTCOME)	Clear scientific evidence that early intervention resulting from screening optimizes outcome	200
	Some scientific evidence that early intervention resulting from screening optimizes outcome	100
	No scientific evidence that early intervention resulting from screening optimizes outcome	0
Benefits of early identification (FAMILY & SOCIETY)	Early identification provides clear benefits to family and society (education, understanding prevalence and natural history, cost effectiveness)	100
	Early identification provides some benefits to family and society	50
	No evidence of benefits	0
Early diagnosis and treatment prevent mortality	YES	100
	NO	0
Availability of diagnostic confirmation	Providers of diagnostic confirmation are widely available	100
	Limited availability of providers of diagnostic confirmation	50
	Diagnostic confirmation is available only in a few centers	0
Acute management	Providers of acute management are widely available	100
	Limited availability of providers of acute management	50
	Acute management is available only in a few centers	0
Simplicity of therapy	Management at the primary care or family level	200
	Requires periodic involvement of a specialist	100
	Requires regular involvement of a specialist	0

Max score 2100

Score Card (2)

UNIFORM PANEL CONDITIONS		DEFINITION	INCIDENCE	PREVALENCE	PHYSICIAN	HOSPITAL	DIAGNOSTIC
Reference Conditions			1718	1883	1642	1633	
YOUR SCORE							
MEDICAL HISTORY (MHA)							
GENETIC TESTING (GTT)							
TREATMENT (T)							
COST OF TREATMENT (COT)							
BENEFIT OF EARLY IDENTIFICATION (BEI)							
BENEFIT OF EARLY IDENTIFICATION (BEI) - FAMILY & SOCIETY							
EARLY DIAGNOSIS AND TREATMENT (EDAT)							
DIAGNOSTIC CONFIRMATION (DC)							
CLINICAL MANAGEMENT (CM)							
SIMPLICITY OF THERAPY (ST)							

Scoring of
MCAD
CH
PKU
SCA
CAH
(optional)

CONDITION(S) UNDER EVALUATION		CRITERIA	WEIGHT	SCORE
Proponent:				
Date:				
Criteria				
Incidence of condition	>10,000	100		
	>100,000	75		
	>100,000	50		
	>1,000,000	25		
	>1,000,000	0		
Signs & Symptoms clinically identifiable in the first 48 hours	Severe	100		
	>50% of cases	75		
	>50% of cases	50		
	>75% of cases	25		
	Always	0		
Burden of disease if untreated	Profound	100		
	Severe	75		
Historical prevalence	Severe	100		
	Mild	75		
Cost of treatment	High	100		
	Low	75		
Potential for early intervention	High	100		
	Low	75		
Benefits of early identification (FAMILY & SOCIETY)	High	100		
	Low	75		
Diagnostic confirmation	Procedures of diagnostic confirmation are widely available	100		
	Limited availability of procedures of diagnostic confirmation	75		
Clinical management	Procedures of clinical management are widely available	100		
	Limited availability of procedures of clinical management	75		
Simplicity of therapy	Requires periodic treatment of a specialist	100		
	Requires regular treatment of a specialist	75		

Once a condition has been identified, scoring by committee and subcommittee members, and ad hoc working group members could be added to the evidence used during deliberation

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Nomination Requirements Fact Sheet

HRSA/ACMG UNIFORM PANEL NOMINATION OF CONDITION - Fact Sheet			
Proponent		Date	10/18/06
Condition			
Type of disorder			
Ethnicity			
Screening method			
CONDITION	Comment	Gene <input type="text"/>	Locus <input type="text"/> OMM <input type="text"/>
Incidence	(include source of data)		
Phenotype at birth	(include typical age)		
Burden if untreated			
TEST	Comment		
Screening test & PILOT STUDY	Location, duration, size, and preliminary results of past/pending pilot study must be provided		
Doable in DBS or by physical method	(describe major characteristics)		
Estimated daily throughput	(Cases/PT/Day or cases/treatment/Day, as applicable)		
Cost of test	(actual cost and estimated testing fee)		
Multiple analytes/markers	(If applicable)		
Secondary targets (other conditions)	(If applicable) [D]		
Multiplex platform	(If applicable)		

Condition

Test

Nomination of condition (page 2)													
TREATMENT	Comment												
Availability & cost													
Efficacy of treatment													
Benefits of early intervention													
Benefits of early identification													
Prevention of mortality													
Confirmation of diagnosis													
Acute management													
Simplicity of therapy													
REFERENCES AND WEB SITES	Submit form to HRSA at the address listed below												
<table border="1" style="width: 100%; border-collapse: collapse;"> <tr><td style="width: 20px;">1</td><td></td></tr> <tr><td>2</td><td></td></tr> <tr><td>3</td><td></td></tr> <tr><td>4</td><td></td></tr> <tr><td>5</td><td></td></tr> <tr><td>6</td><td></td></tr> </table>	1		2		3		4		5		6		<p style="font-size: 1.2em; color: red; border: 1px solid red; padding: 5px; display: inline-block;">References</p> <p style="font-size: 0.8em;">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 301-443-8504-fax 301-443-1080-phone</p> <p style="text-align: center; font-weight: bold; font-size: 0.8em;">Submission check list</p> <p style="font-size: 2em; color: red; border: 1px solid red; padding: 5px; display: inline-block;">Checklist</p> <p style="font-size: 0.8em;">Copy of references listed on this form</p>
1													
2													
3													
4													
5													
6													

Treatment

References

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)**