

# Criteria Work Group: Update on the Nominating and Review Process

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6-5-06 ACHDGDNC



# Nomination work group (10/05 - )

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

HRSA staff: Michele Puryear, Marie Mann



# 3 proposed steps for nominations

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- Step #1: The Nomination Form
- Step #2: Federal administrative review
- Step #3: Review by ACHDGDNC
  - A) External evidence-based review
  - B) Committee review



# Nomination Process - concepts

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



# Tasks

## - *Since 2-06 ACHDGDNC Meeting*

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- 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 (*Dr. Dougherty*)
  - ACHDGDNC



# Nomination Form:

## *Since last AC meeting*

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- 1) Made modifications to the NF
- 2) Piloted NF to consumers and health professionals
- 3) Solicited additional input from consumer health advocacy groups



# Suggestions for the submission process from the 2/06 *ACHDGDNC* meeting

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- 1) Keep the NF simple
- 2) Keep the process consistent with downstream review
- 3) Tracking number for submissions
- 4) Streamline process - funnel multiple applications on same or linked disorders



# Previous *ACHDGDNC* meeting (continued)

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- 5) Only the *ACHDGDNC* can reject an NF application, even with expedited review
- 6) Conflict-of-interest disclosure to NF and overall nominating process (*ACHDGDNC too*)
- 7) How to prioritize?
  - May want to start with those 84 disorders that were on the ACMG's original list
- 8) Consider economic impact of screening in the EBR step.



# Revised Nomination Form

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Based on comments received at  
the last *ACHDGDNC*



# HRSA/ACMG UNIFORM PANEL (DRAFT ~~01/21/06~~ 5/18/06)

## NOMINATION OF CONDITION - Fact Sheet

<b>Name of proponent</b>	<u>(Organization, if relevant)</u>	<b>Date</b>	
<b>Condition</b>			
<b>Type of disorder</b>			
<b>Screening method</b>			
<b>Treatment strategy</b>	<i>* <u>Note: Please reference each statement, listing references below</u></i>		

CONDITION	Comment	Gene	Locus	OMIM
<b>Incidence</b>	(Reference required; By pilot screening or clinical identification?)			
<b>Timing of clinical onset</b>	(Relevance of the timing of newborn screening to onset of clinical manifestations)			
<b>Severity of disease</b>	(Morbidity, disability, mortality) <span style="color: red;"><u>What spectrum of severity?</u></span>			

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

## Nomination of condition (page 2)

TREATMENT	Comment
<b>Modality</b>	(Drug(s), diet, replacement therapy, transplant, other)
<b>Urgency</b>	(How soon after birth treatment needs to be initiated to be effective)
<b>Efficacy</b>	(Extent of prevention of mortality, morbidity, disability) <u>Treatment limitations, such as difficulty with acceptance or compliance?</u>
<b>Availability</b>	(Any limits of availability)
<b>Risks</b>	(Potential medical or other ill effects from treatment)

**KEY REFERENCES (Specific citations - limit to 15)**

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**Submit nomination to:**

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

**Submission check list**

<b>Cover letter by proponent</b>
<b>Nomination form</b>
<b>Copy of references listed on this form</b>

**Contact information (proponent)**

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**REFERENCES (continued)**

12	
13	
14	
15	

# Responses to pilot NF

(as of 5/31/06)

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## CONSUMERS:

- Sharon Terry, MA - President, Genetic Alliance
- Jill Fisch – President, Save Babies Through Screening
  - pilot NF with Wilson’s Disease
    - Kimberly F. Symonds, Wilson's Disease Association
- Micki Gartzke – Director, Hunter’s Hope Foundation
  - pilot NF with Krabbe Disease

# Responses to pilot NF

(as of 5/31/06)

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## HEALTH PROFESSIONALS:

- Carol Greene, MD - U. Maryland
  - Glutaric Aciduria type 1
- Jennifer Puck, MD - UCSF
  - SCID (Severe Combined Immune Deficiency)
- Priya S. Kishnani, MD - Duke U.
  - Pompe Disease
- Jill Jareki, PhD - Research Director, Families of SMA
  - Spinal Muscular Atrophy

# Comments from the Genetic Alliance

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- Pilot NF with a largely well trained, savvy consumer committee
- Have strict general guidelines for inclusion:
  - A) Limits of condition for which onset is  $> 30$ ?  $40$ ?
  - B) Reject disorders for which there are no treatments
  - C) Define minimum incidence

\*Define “treatment”



# Genetic Alliance comments (continued)

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- \* GA offers to coordinate consumers to:
  - 1) Determine suitability for considering a new disorder
  - 2) Assist in completing Nomination Form
  - 3) Interpret decisions



# More from the Genetic Alliance

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\* Lingerling issue:

\* Define “*treatment*”



# Nomination Process: Next tasks

## ■ Steps 1-3:

- Step 1: Nomination Form ◀ — — — — —
  - COMPLETE PILOT PHASE:
    - Review current submissions
    - Additional feedback?
  - *ACHDGDNC* TO APPROVE NF
- Step 2: Federal administrative step
  - DEFINE PROCESS
- Step 3: *ACHDGDNC* review
  - DEFINE EBR PROCESS — — — — —

