Overview of New Advisory Committee on Heritable Disorders in Newborns and Children Consumer-Friendly Resources

February 10, 2022

UNC SCHOOL OF MEDICINE
DEPARTMENT OF PEDIATRICS

UNC HEALTH CARE
Children's

Background

Consumer-Friendly Resources

- Committee's review of its nomination, evidence-based review and decision-making processes.
- Stakeholder feedback
- Expert opinion
- Previous nominators

Consumer-Friendly Resources

- Seven new/updated pages on the ACHDNC website
  - Nominate a Condition Page
    - Fillable PDF nomination form
  - Condition Nomination Review Process Page
  - Nominate a Condition FAQs Page
  - Key Questions Considered by the Committee Page
  - Sample Questions Addressed in an Evidence-Based Review Page
  - Committee Approach to Evaluating the Condition Review Report Page
  - ACHDNC History

Nominate a Condition

Advisory Committees on Heritable Disorders in Newborns and Children

- Home
- About
- Meetings
- Reports
- Letters
- Resources
- Recommended Uniform Screening Panel (RUSP)
- Nominate a Condition
- Previously Nominated Conditions
- Newborn Screening Timeliness Goals

Nominate a Condition

As of January 1, 2022, all conditions nominated for inclusion on the Recommended Uniform Screening Panel must use the Committee's updated condition Nomination Form, found in the Nomination Package Components section of this page.

On This Page

Additional Resources

- Condition Nomination Review Process
- Nominate a Condition FAQs
- Key Questions Considered by the Committee
- Sample Questions Addressed in an Evidence-Based Review
- Committee Approach to Evaluating the Condition Review Report (Decision Matrix)

ACHDNC Form for Nomination of a Condition for Inclusion in the Uniform Screening Panel

Date:

Nomination Team	
NAME OF NOMINATOR AND ORGANIZATION (include professional degree)	INDICATE AFFILIATION (i.e., Health Professional, Subject Matter Expert, Researcher, Clinician, Advocate, etc.)
CO-SPONSORING ORGANIZATIONS (include professional degrees)	INDICATE AFFILIATION (i.e., Health Professional, Subject Matter Expert, Researcher, Clinician, Advocate, etc.)

**Note: Please reference each statement/answer with the corresponding reference number listed in Section III - Key References.*

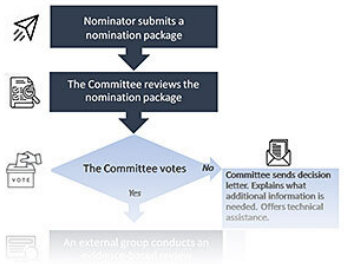
SECTION I - CONDITION INFORMATION AND TREATMENT

SECTION I, PART A. CONDITION

CONDITION	STATEMENT
Nominated Condition	
Type of Disorder	
Screening Method	
Gene	If applicable, if not N/A
Critical Biomarker	If applicable, if not N/A
Locus	Include ClinVar link if applicable.

Condition Nomination Review Process

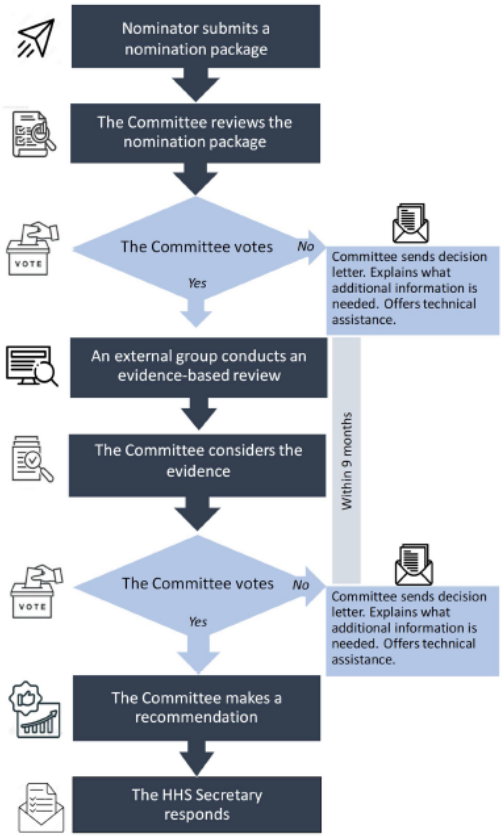
The ACHDNC Nomination, Review, and Decision-Making Process



Download the flowchart graphic (PDF, 883 kb)

- Nominator submits a nomination package
- Committee reviews the nomination package
- The Committee votes
- External group conducts an evidence-based review
- The Committee considers the evidence
- The Committee votes
- The Committee makes a recommendation
- The HHS Secretary responds

Diagram of Steps for Adding a Condition to the RUSP



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Nominate a Condition FAQs

Examples:

Q: How long does that process take to get a condition added to the RUSP?

A: For conditions that have been added to the RUSP using this process, the time from when a nomination is *first* presented to the Committee, to when the Secretary of Health and Human Services adds the condition to the RUSP has ranged from 1 year and 9 months (21 months) to 10 years (120 months). Most have been around 3 to 4 years.

Many condition nomination packages have had to be resubmitted to provide the Committee with all the information needed to consider whether the nomination is ready for full review.

Q: What happens if a nomination is not accepted or is deemed incomplete or not ready for Committee review?

A: Before a nomination is accepted, HRSA conducts an administrative review of the nomination package and form. If the nomination package or form is missing any information, the Committee's Designated Federal Official (DFO) will return it to the nominator, identifying the components needing further information. It is important to reach out to the HRSA DFO early and often during development of the nomination package. They are available to answer questions about the process.

Key Questions Considered by the Committee

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Key Questions Considered by the Committee

The nomination package answers key questions about the nominated condition, the screening process, and treatment. The Committee considers each of these questions during review:

1. *Condition Seriousness.* Is the nominated condition medically serious?
2. *Case Definition.* Are the condition's case definition and spectrum well described? Can they predict the phenotype or range of symptoms in newborns and children who will be identified through population-based screening?
3. *Analytic Validity.* Is the condition's screening process valid and reasonable for the newborn screening system? Is it sensitive enough to not miss any newborns who have the condition (i.e., have a low rate of false-positives)?
4. *Clinical Utility.* Is the screening process clinically useful? Is it specific enough to find babies who have the condition, especially those most likely to benefit from treatment (especially if treatment is involved or risky)?
5. *Treatments.* Are treatment protocols well-defined? Are U.S. Food and Drug Administration-approved drugs (if applicable) and treatments available?
6. *Prospective Pilot Data.* Are there data about how well population-based screening works to find newborns with the condition?

Date Last Reviewed: January 2022

Sample Questions Addressed in an Evidence-Based Review

Benefits and Harms of Screening and Diagnosis (Not Related to Treatment)

This topic reviews benefits and harms, not related to treatment, that could result from newborn screening and early diagnosis. Many benefits and harms affect both the newborn and family.

Key Questions under this topic include:

- What are the harms of wrongly classifying a baby without the condition as high-risk?
- What are the harms of wrongly classifying a baby with the condition as low-risk?
- What are the harms/benefits of diagnosing newborns who do not have the condition with condition-related gene changes?
- What are the harms/benefits of diagnosing newborns found from newborn screening with the condition?

Treatment and Long-Term Follow-Up Care

This topic reviews current treatment practices and guidelines. It covers treatment types, details, and duration and whether treatment changes based on age or symptoms.

Key Questions under this topic include:

- What are the treatment indications for the condition?
- Do treatment and long-term follow-up guidelines exist?
- Are there recommended treatments for the condition?
- Are there clinical experts who can oversee treatment and long-term follow-up care?

Committee Approach to Evaluating the Condition Review Report

<table>
<tr><th rowspan="2">NET BENEFIT/ CERTAINTY</th><th colspan="3" rowspan="2"></th><th colspan="3">READINESS</th><th colspan="2" rowspan="2">FEASIBILITY</th></tr>
<tr><th>Ready</th><th>Developmental</th><th>Unprepared</th></tr>
<tr><td rowspan="3">SIGNIFICANT Benefit</td><td rowspan="3">Certainty</td><td>HIGH</td><td>A1
Screening for the condition has a high certainty of significant net benefits, screening has high or moderate feasibility. Most public health departments are ready to screen.</td><td>A2
Screening for the condition has a high certainty of significant net benefits and screening has high or moderate feasibility. Public health departments have only developmental readiness.</td><td>A3
Screening for the condition has a high certainty of significant net benefits and screening has high or moderate feasibility. Public health departments are unprepared for screening.</td><td rowspan="2">Feasibility</td><td>HIGH or MODERATE</td></tr>
<tr><td>HIGH</td><td colspan="3">A4
There is high certainty that screening would have a significant benefit; however, most health departments have low feasibility of implementing population screening.</td><td>LOW</td></tr>
<tr><td>MOD</td><td colspan="3">B 1-4
There is moderate certainty that screening would have a significant benefit.</td><td>----</td></tr>
<tr><td>Small to ZERO Benefit</td><td rowspan="3">Certainty</td><td rowspan="2">MOD/HIGH</td><td colspan="3">C 1-4
There is high or moderate certainty that adoption of screening for the targeted condition would have a small to zero net benefit.</td><td>----</td></tr>
<tr><td>NEG Benefit</td><td colspan="3">D 1-4
There is high or moderate certainty that adoption of screening for the targeted condition would have a negative net benefit.</td><td>----</td></tr>
<tr><td>---</td><td>LOW</td><td colspan="3">L 1-4
There is low certainty regarding the potential net benefit from screening.</td><td>----</td></tr>
</table>

Download a PDF of the Decision Matrix (PDF - 254 KB)

- Principles for Making Recommendations
- Assessing Strength of Evidence at the Key Question Level
- Assessing the Magnitude of Net Benefit

Questions?