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# Nutrition Management Guidelines for Inborn Errors of Metabolism

## *Outlining Approach to Gathering Evidence and Building Consensus*

SOUTHEAST



NBS & GENETICS  
COLLABORATIVE

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# Statement of Conflict of Interest

**This speaker has no conflicts  
of interest to declare**

## Part I

- **Importance of Evidence-based Practice**
- **Steps in Evidence/consensus Analysis Process**

## Part II

- **Implementation Phase (MSUD & PROP and FAO)**
- **Evidence Based Nutrition Practice Guidelines for IEM**
- **Question formulation**
- **Literature search**
- **Pilot Delphi Survey**



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# Vision Based on NEED

**Online Resource with the best available research  
on Nutrition Management of IEM in a  
practitioner –friendly format**



# WHY interest in EBP ?

<b>1960s &amp; before</b>	<b>clinical judgment/ "Art"</b>
<b>1970s</b>	<b>wide variations in practice patterns</b>
<b>1980s</b>	<b>landmark studies showed inappropriate procedures</b>
<b>1990s</b>	<b>Increasing complexity of clinical decisions</b>

**First published use of term "evidence-based" describing clinical guidelines appeared in *JAMA* in 1990**

*Courtesy: Cummins ADA*

# Evidence Based Practice (EBP)

## Most Common Definition

*“The conscientious, explicit, and judicious use of current best evidence in making decisions about the care of individual patients” (Sackett 1996, BMJ)*

## How to achieve:

***“A set of principles and methods intended to ensure that to the greatest extent possible, medical decisions, guidelines, and other types of policies are based on and consistent with good evidence of effectiveness and benefit.”***

(Eddy 2005, Health Affairs)



# Why Evidence-based?

- ✓ **Improve quality of healthcare**
- ✓ **Decrease wide variations in practice**
- ✓ **Reduce the gap between what is known from research...and what happens in real life**
- ✓ **Take advantage of biomedical knowledge**

# Domains of Systems to Evaluate Research

Quality

Quantity

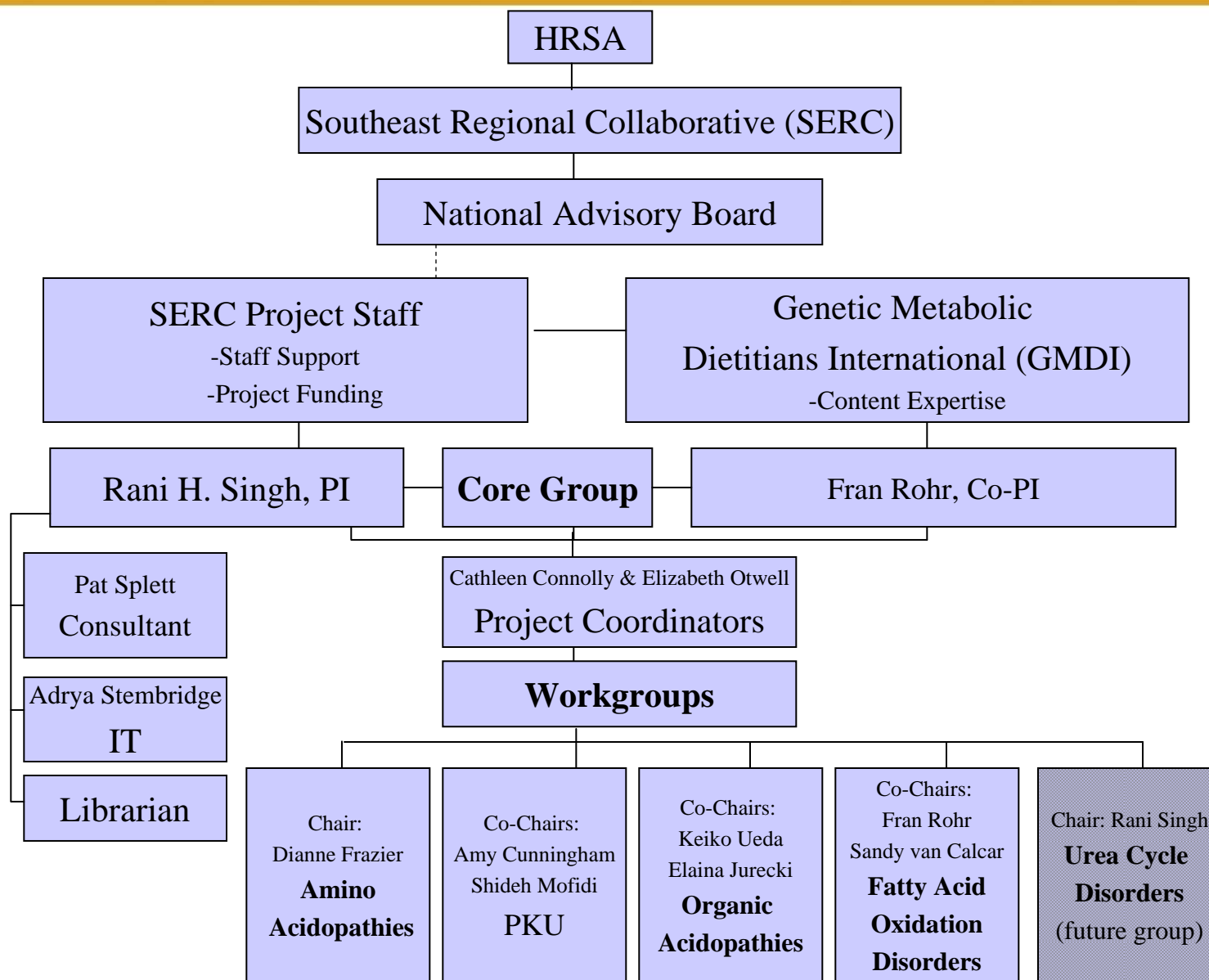
Consistency

Strength of  
evidence /  
Grading system

**Agency for Healthcare Research & Quality 2002 -  
[www.ahrq.gov](http://www.ahrq.gov)**

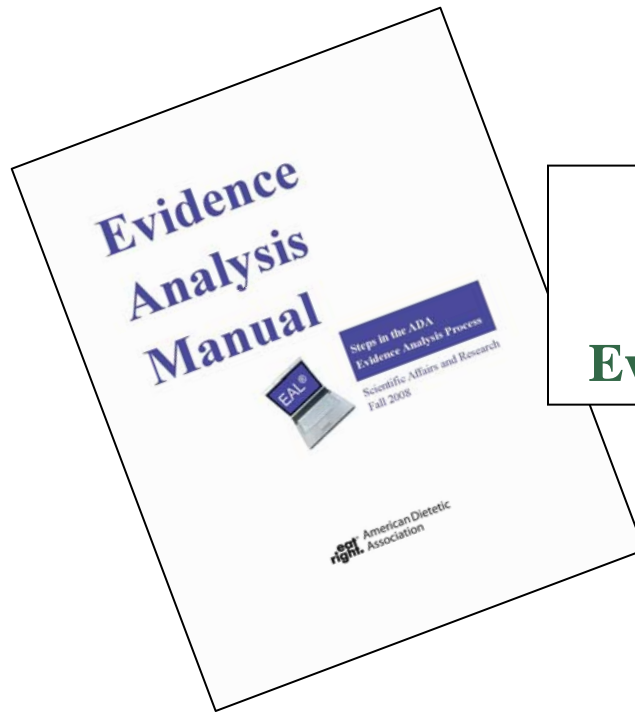


# Project Organization: Pilot 2007 & Formal Organization 2008





# Guideline Development Process



 American Dietetic  
Association

**Evidence Analysis Library**

- Evidence Based: Systematic search and review of scientific and practice literature
- ADA analyst training completed by core group



# Evidence-Analysis Process Steps

Select topic & appoint  
expert work group

1. Define questions  
and determine inclusion/  
exclusion criteria

5. Develop conclusion  
statement & assign grade

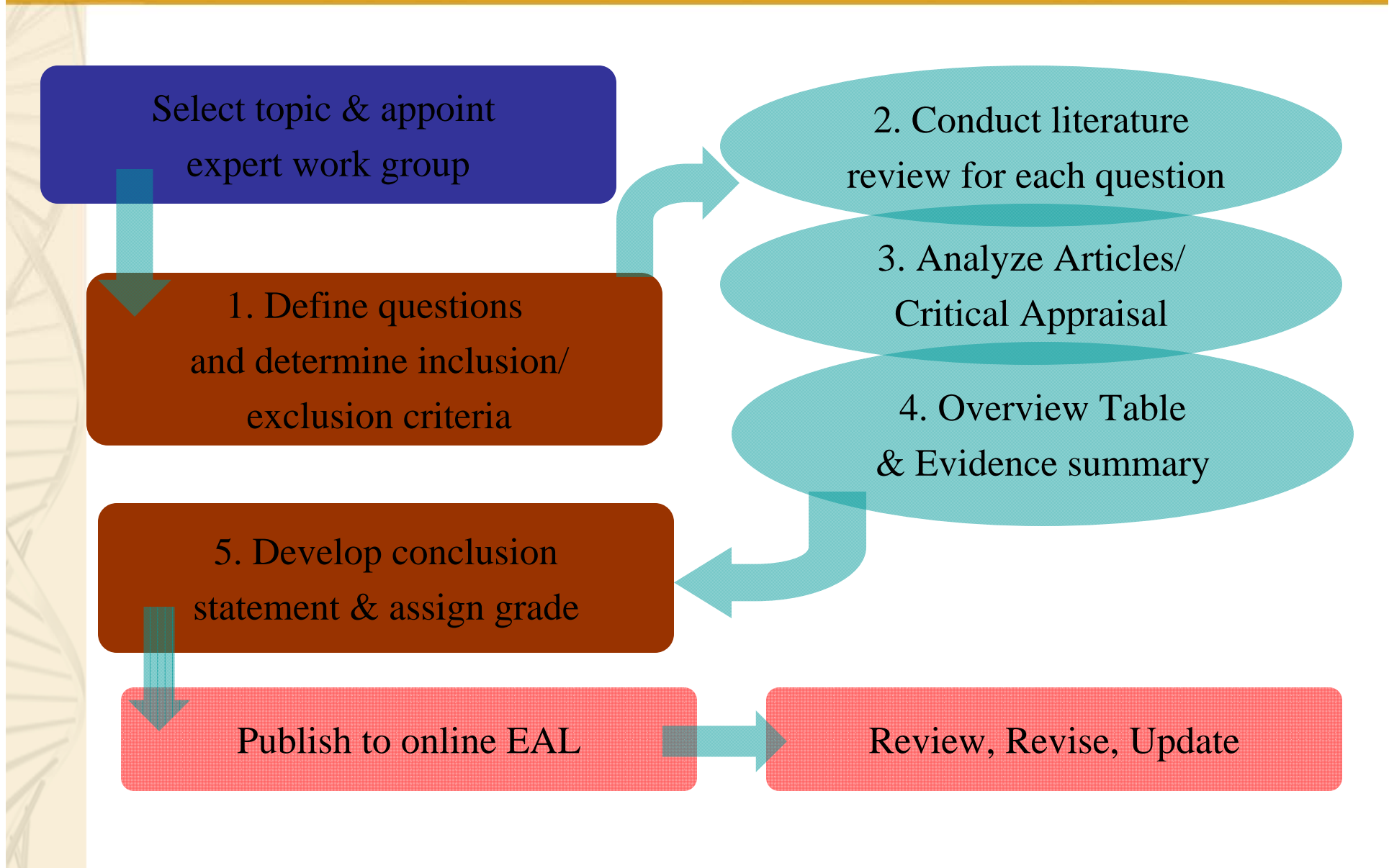
Publish to online EAL

2. Conduct literature  
review for each question

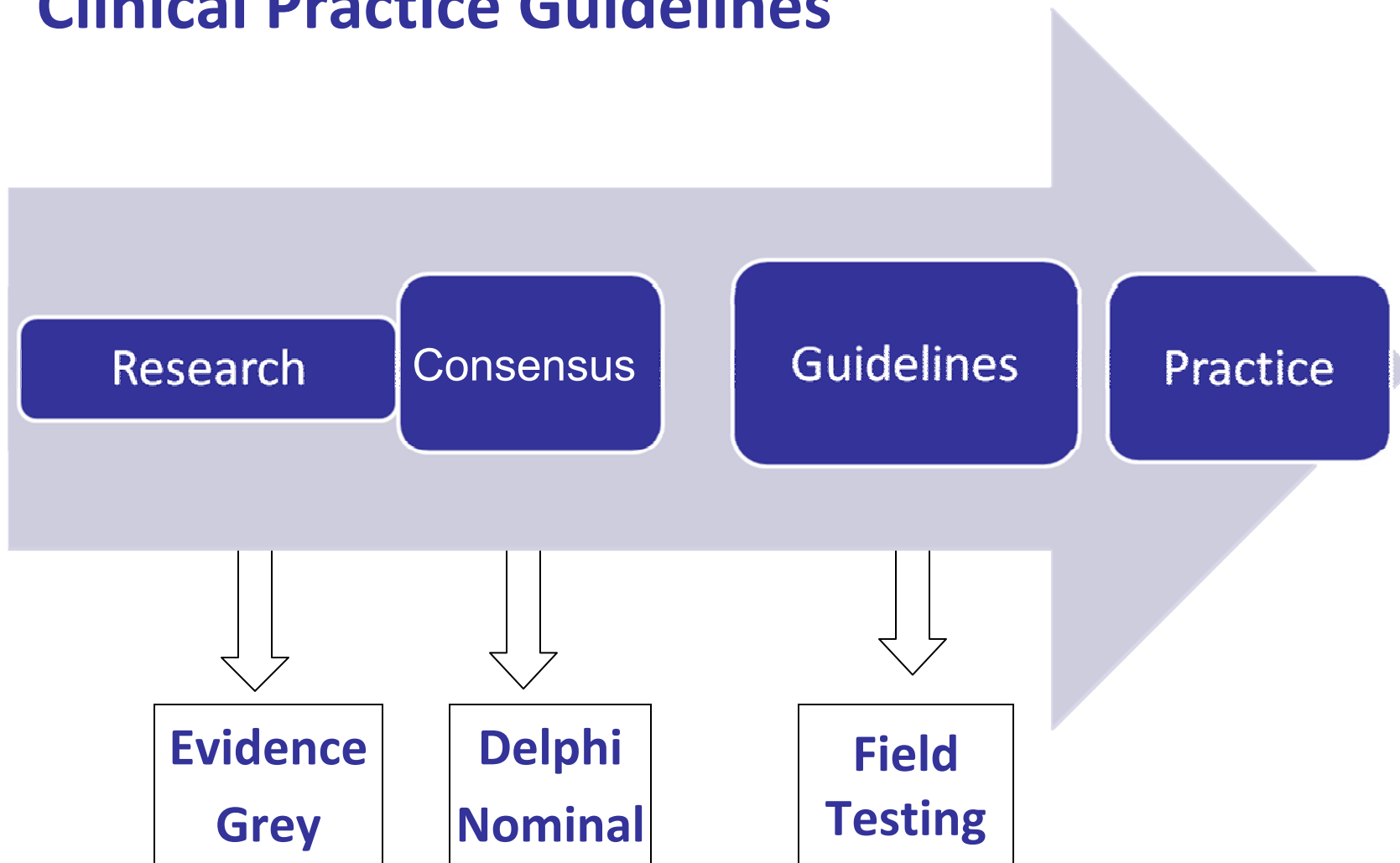
3. Analyze Articles/  
Critical Appraisal

4. Overview Table  
& Evidence summary

Review, Revise, Update



## Clinical Practice Guidelines

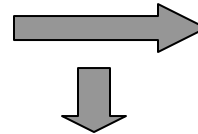




# Nutrition Guideline Development Process

## Evidence Analysis

- Systematic Review of published literature
- Collection/review of clinical protocols, presentations



## Delphi Process –

### Round 1

- Survey of physicians and dietitians from each HRSA Genetics Region

## Summary

- Recommendations for nutrition management based on literature review
- Identification of variations in practice based on Delphi survey



## Nominal Group Technique

- Face-to-Face meeting of expert dietitians, physicians, researchers, Consumers
- Discuss areas of practice variation, vote, discuss recommendations, vote again



## Delphi Process - Round 2

- Survey of physicians and dietitians from each HRSA Genetics Region regarding recommendations and variations in practice identified by nominal group technique



## Nutrition Guidelines

- Recommendations for nutrition management documenting areas of consensus and non-consensus



## Review Process

- External Review
- Field Testing

# Propionic Acidemia Key Topics

What are the nutrient requirements by age and nutrient?

How is the AA□restricted diet initiated in symptomatic patients?

Should all patients be challenged with biotin/supplemented with biotin?

Should all patients be supplemented with carnitine?

What diet should sick patients follow?

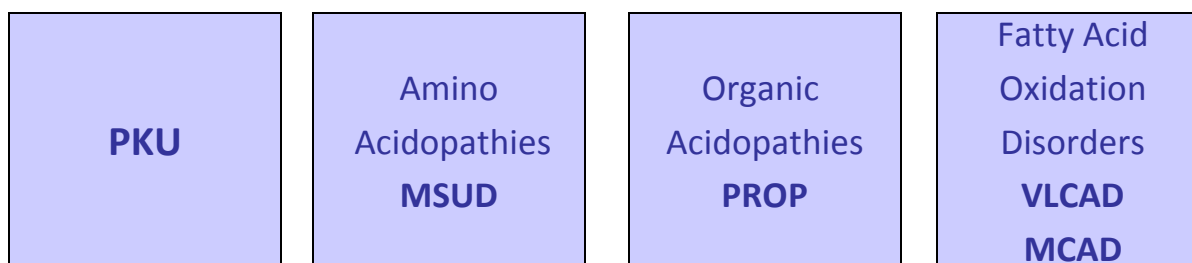
What are the monitoring guidelines?

Are dietary recommendations altered for menstrual cycle, pregnancy, lactation, and post□delivery?

What are dietary recommendations for special circumstances: pancreatitis, cardiomyopathy, pre□and post□ liver transplantation

# Workgroups

- **First 5 disorders for guideline development**

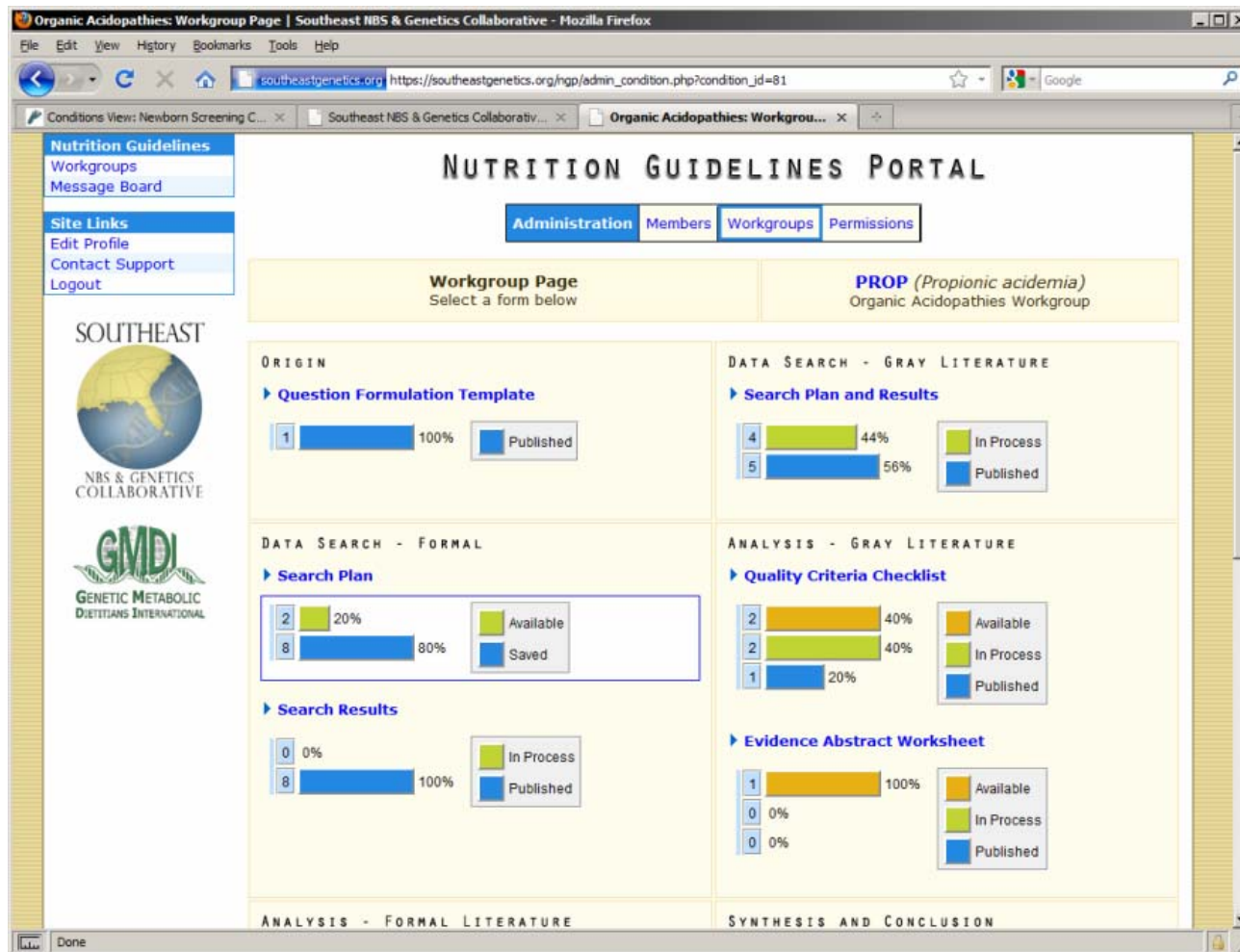


- **Priorities:**

- Disease incidence
- GMDI membership survey results
- Body of literature available
- Expertise of workgroup members

# Nutrition Guidelines Portal

- Allows work from multiple locations
- Based on a series of questions (5 disorders)
- Literature uploaded & abstracted by analysts
- Integrated message board



The screenshot displays the Nutrition Guidelines Portal interface within a Mozilla Firefox browser window. The browser's address bar shows the URL: [https://southeastgenetics.org/ngp/admin\\_condition.php?condition\\_id=81](https://southeastgenetics.org/ngp/admin_condition.php?condition_id=81). The page title is "Organic Acidopathies: Workgroup Page | Southeast NBS & Genetics Collaborative - Mozilla Firefox".

The main content area is titled "NUTRITION GUIDELINES PORTAL" and includes navigation tabs for "Administration", "Members", "Workgroups", and "Permissions". The current view is "Workgroup Page" for the "PROP (Propionic acidemia) Organic Acidopathies Workgroup".

The interface is divided into several sections:

- ORIGIN**: A "Question Formulation Template" section showing a progress bar for item 1 at 100% and a "Published" button.
- DATA SEARCH - GRAY LITERATURE**: A "Search Plan and Results" section showing progress bars for items 4 (44%) and 5 (56%), with "In Process" and "Published" status indicators.
- DATA SEARCH - FORMAL**: A "Search Plan" section showing progress bars for items 2 (20%) and 8 (80%), with "Available" and "Saved" status indicators. Below it is a "Search Results" section showing progress bars for items 0 (0%) and 8 (100%), with "In Process" and "Published" status indicators.
- ANALYSIS - GRAY LITERATURE**: A "Quality Criteria Checklist" section showing progress bars for items 2 (40%), 2 (40%), and 1 (20%), with "Available", "In Process", and "Published" status indicators. Below it is an "Evidence Abstract Worksheet" section showing progress bars for items 1 (100%), 0 (0%), and 0 (0%), with "Available", "In Process", and "Published" status indicators.
- ANALYSIS - FORMAL LITERATURE**: A section at the bottom left.
- SYNTHESIS AND CONCLUSION**: A section at the bottom right.

The left sidebar contains navigation links for "Nutrition Guidelines", "Workgroups", "Message Board", "Site Links", "Edit Profile", "Contact Support", and "Logout". Logos for "SOUTHEAST NBS & GENETICS COLLABORATIVE" and "GMDI GENETIC METABOLIC DIETITIANS INTERNATIONAL" are also present.





# Guideline Template

- **Background**
- **Biochemical Basis**
- **Nutrition Assessment**
- **Nutrition Problem Identification**
- **Nutrition Management/Intervention**
- **Education**
- **Monitoring and Evaluation**
- **Barriers to Implementation**
- **References**

# Nutrition Guidelines Review and Dissemination

- **Field testing**
  - Guidelines tested in management of patients
- **Dissemination**
  - **Web-based**
    - [www.gmdi.org](http://www.gmdi.org)
    - [www.southeastgenetics.org](http://www.southeastgenetics.org)
  - **Links**
    - External: OMIM, ACMG ACT Sheets, Star-G
    - Internal: food lists, formula information, DRIs
  - **Dynamic: periodic updates**



## Expected Project Outcomes

- Nutrition Guidelines for management of metabolic disorders for which there is little published scientific evidence
- Increased consistency of genetics care across centers, states and regions:
  - Less uncertainty and variability in patient management
  - Consistent documentation of care and tracking of outcomes
  - Sharing of current clinical data and experience to guide practice
  - Identification of priority questions for collaborative research

# Future Direction

- **Phase 2: Include tool kits for families**
- **Utilize the infrastructure to expand to general management guidelines.**

# Acknowledgements

- **GMDI**

**Fran Rohr, MS, RD – Project Co-PI & GMDI President  
Nutrition Guideline Workgroup Chairs and Members**

- **SERC**

**Coordinators and Contributors:**

**Cathleen Connolly, Elizabeth Otwell,  
Pat Splett, Adrya Stembridge**

- **HRSA funding support**

