



Office of
Dietary Supplements
National Institutes of Health



NCATS
Catalyzing
Innovation

NUTRITION AND DIETARY SUPPLEMENT INTERVENTIONS for Inborn Errors of Metabolism (NDSI-IEM)



An Initiative to Build an Evidence-Based Research Framework

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National Institutes of Health

Secretary's Advisory Committee on Heritable
Disorders in Newborns and Children

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NIH . . . Turning Discovery into Health

Robust Data are Lacking

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- On the safety and effectiveness of many of the treatments used for IEM
 - ▣ Cochrane Review on the efficacy of tyrosine supplementation for PKU (2010, Webster and Wildgoose)
 - Three trials met inclusion criteria with results on 56 subjects
 - No recommendations could be made regarding use of tyrosine in routine clinical practice
 - ▣ AHRQ Comparative Effectiveness Review, Adjuvant Treatment for PKU (February 2012)
 - Key Q1--the relationship between Phe levels and IQ
 - 16 studies met review criteria
 - Study quality: 1 = good, 4 = fair, 11 = poor

Needs and Opportunities in IEM Research

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Traditional research and development is changing



Utilize available resources from multiple private and public sector partners

Sample size, recruitment, geographical distribution, and ethical issues



Many rare diseases and IEM will not go to phase 3 trials; small pilot or phase 2 studies

Utilize novel clinical trial designs, but . . .



Work with FDA review divisions for proposed study design and assessment of results

Bridge the gap between scientific discovery and clinical application



NCATS and other NIH translational research programs

NDSI-IEM Initiative

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Nutrition and Dietary Supplement Interventions for Inborn Errors of Metabolism

- 2008—Inception from discussions in the Office of Dietary Supplements and the Office of Rare Diseases Research
- 2010—Initiative launched
- January 2011--a meeting of Federal partners was convened
- December 2011--stakeholder workshop was held
- January 2012--work began on specific activities

NDSI-IEM Mission

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- Identify gaps in the understanding of the safety and effectiveness of nutrition and dietary supplement interventions used to treat persons with IEM.
- Through collaboration and partnerships among a wide range of stakeholders, develop and implement a framework to conduct evidence-based research.

NDSI-IEM Stakeholder Workshop

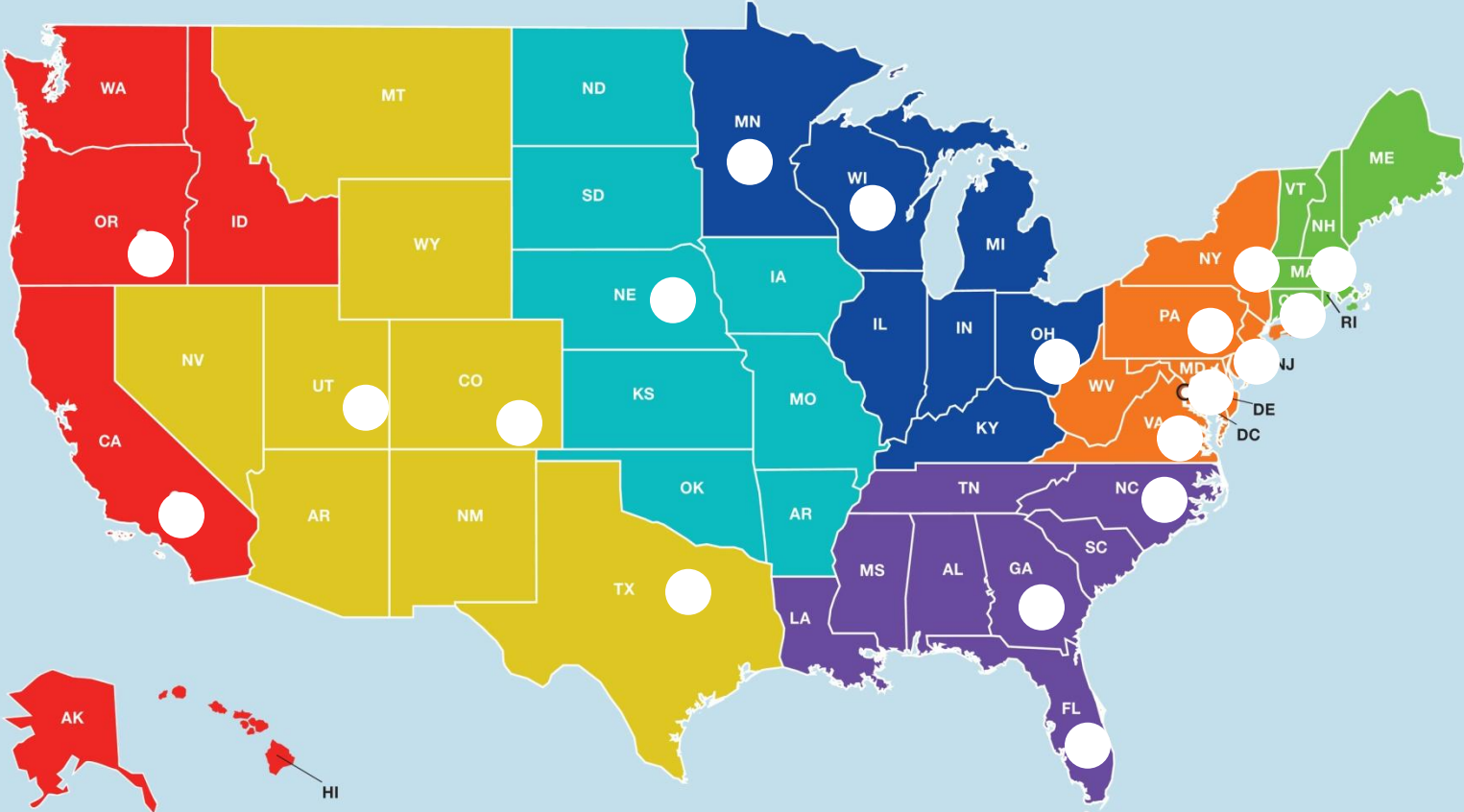
December 6-7, 2011

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Participants

- Advocacy organizations
- Clinical/research/academia
- Genetic and NBS Services Regional Collaboratives
- Professional associations
- Industry representatives
- Federal partners (NIH, FDA, HRSA, and AHRQ)

Geographic Distribution of Participants by State Within the Genetic and Newborn Screening Services Regional Collaboratives



- KEY**
- Region 1: New England Regional Genetics Group, Inc.
 - Region 2: The New York/Mid-Atlantic Region
 - Region 3: Southeast NBS and Genetics Collaborative
 - Region 4: The Region 4 Genetics Collaborative
 - Region 5: Heartland Genetics and Newborn Screening Collaborative
 - Region 6: Mountain States Genetics Regional Collaborative Center
 - Region 7: Western States Genetic Services Collaborative
 - States represented with participants
 - Countries represented with participants: Australia, France, Switzerland, and United Kingdom

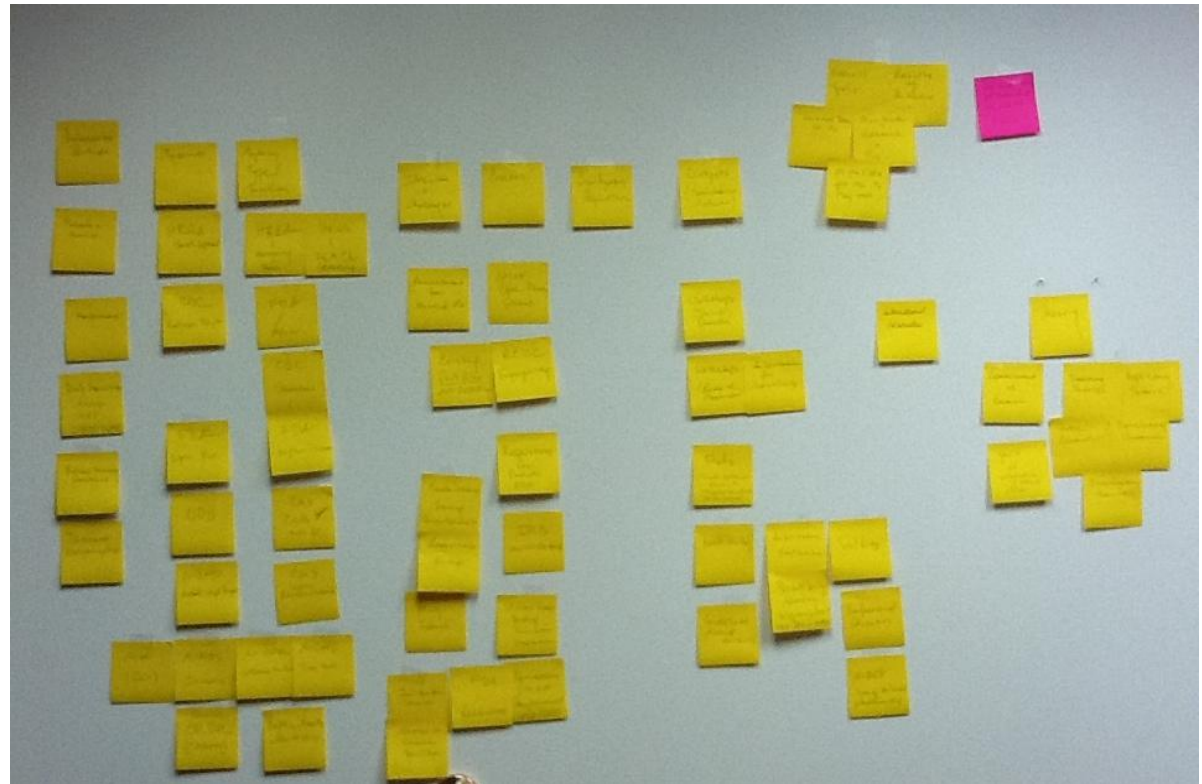
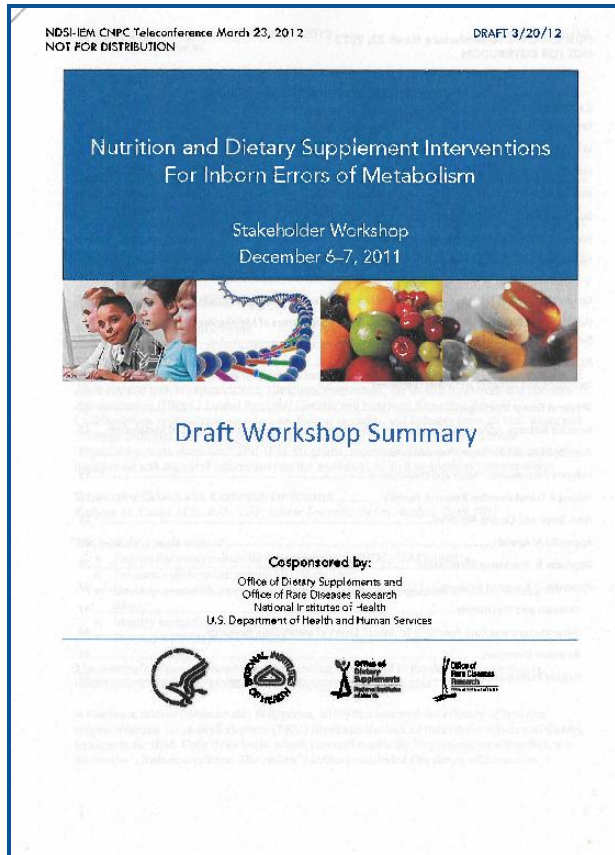
Setting a Transformative Research Agenda--Action Plan

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- Define short and long-term priorities
 - What can be done in 1 yr, 5 yrs, 10 yrs?
- Define a preferred sequence of events based on resources
 - What is most critical to address?
- Mechanisms needed
 - Formal working groups; in-depth strategic planning process; other?

Stakeholder Workshop Outcomes

- Twenty-one hours of recorded talks and discussion
- An 80-page summary document



Activities Underway

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- Core Planning Group
 - Review short-term through long-term activities
 - Agree and prioritize
- Website update and enhancements
- Prepare workshop proceedings for publication
 - Writing group formed and have begun their work
- Develop a list of diseases and treatments
- Survey existing research, expertise, resources, and needs
 - ODS in collaboration with SIMD and GMDI
- Create a web portal to organize available resources (for researchers, clinicians, and patients/families)

Future Activities

- Build a framework for any disease and treatment
- Collaboration between NIH, FDA, and clinical/research teams to foster cooperation and facilitate successful grant applications
- International collaboration using existing mechanisms, i.e., the ISSIEM and IRDRC
- Plan to conduct natural history studies
- Education/mentoring plan (especially for new researchers)
- Standardize database information and language
- Build registries or utilize existing ones

NDSI-IEM Website Development

- Password protected site for sharing of information among those involved in the initiative
 - <http://www.team-share.net/NDSI-IEM-Initiative/>
 - Login and password: IEM/IEM
- Public access pages on the ODS website

NUTRITION AND DIETARY SUPPLEMENT INTERVENTIONS for Inborn Errors of Metabolism (NDSI-IEM)



An Initiative to Build an Evidence-Based Research Framework

OVERVIEW 

CORE PLANNING
GROUP 

WORKING GROUPS 

ACTIVITIES 

NDSI-IEM MEETINGS 

RESOURCES 

NEWS & UPDATES 

CONTACT US 

Overview

Recognizing the lack of robust evidence for the safety and efficacy of the nutrition and dietary supplement interventions used in inborn errors of metabolism (IEM), this initiative was established in 2009 to build an evidence-based research framework.

Mission

To identify gaps in our understanding of the safety and effectiveness of nutrition and dietary supplement interventions used to treat persons with inborn errors of metabolism. Through collaboration and partnerships among a wide range of stakeholders, develop and implement a framework to conduct evidence-based research.

Guiding Principles of NDSI-IEM

- To serve patients throughout the lifecycle
- To partner and collaborate among all involved and interested parties
- To harmonize activities across public and private entities
- To educate all those who have an impact on the well-being of persons with IEM
- To think creatively and consider new approaches to solving problems



Office of
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Office of
Rare Diseases
Research
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Overview

Working Group

NDSI-IEM Meetings

Resources

News & Updates

Contact Us



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