Presented on behalf of GMDI

- Role of Genetic Metabolic Dietitians in Newborn Screening (NBS) Long-term Follow up
- Activities in the field of genetic metabolic nutrition
- Current workforce and challenges
- Future needs and plans
The mission of GMDI is to provide standards of excellence and leadership in nutrition therapy for genetic metabolic disorders through clinical practice, education, advocacy, and research.
<table>
<thead>
<tr>
<th>Metabolic Disorders</th>
<th>Hematology</th>
<th>Others</th>
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<tbody>
<tr>
<td>Organic Acidurias</td>
<td>Fatty Acid Oxidation</td>
<td>Amino Acids</td>
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<td>Propionic acidemia</td>
<td>Carnitine uptake defect/carnitine</td>
<td>Classic Phenylketonuria</td>
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<td>Methylmalonic academia (MUT)</td>
<td>transport</td>
<td>Maple Syrup urine</td>
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<td>Methylmalonic academia (Cbl A, B)</td>
<td>Medium-chain acyl-CoA dehydrogenase</td>
<td>Homocystinuria</td>
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<td>Isovaleric acidemia</td>
<td>Very long-chain acyl-CoA dehydrogenase</td>
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<td>3-Hydroxy 3-methylglutaricaciduria</td>
<td>Long-chain L-3-hydroxyacyl-CoA</td>
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<td>3-Methylcrotonyl-CoA carboxylase</td>
<td>dehydrogenase</td>
<td>Tyrosinemia 1</td>
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<td>Holocarboxylase synthase def</td>
<td>Trifunctional protein deficiency</td>
<td>Arginosuccinate aciduria</td>
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<td>ß-Ketothiolase deficiency</td>
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<td>Citrullinemia I</td>
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<td>Glutaric acidemia I</td>
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<td>Conditions in bold are treated with medical foods and/or single amino acids, amino acid mixtures, vitamins, or other cofactors</td>
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<td>Therrell BL, MGM 113 2014</td>
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<td>Biotinidase deficiency</td>
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<td>Congenital adrenal</td>
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<td>hyperplasia</td>
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<td>Congenital hypothyroid</td>
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<td>Cystic fibrosis</td>
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<td>Classic Galactosemia</td>
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<td>Pompe</td>
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<td>Hearing loss</td>
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<td>immunodeficiency</td>
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<td>MPS</td>
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ACHDNC defined the goal of LTFU as assuring the best possible outcome for individuals with disorders identified through newborn screening.
Successful management of NBS-identified disorders requires:

**Immediate initiation of treatment**
- **Efficient communication**: NBS coordinator, PCP, family, genetics team
- **Access to treatment**: Expert RDN, coordination of specialty formula

**Lifelong diet intervention**
- Ongoing **care coordination** (not necessarily reimbursable)
- **Evidence based interventions**: To ensure access to medical food, medications and lifelong care
- **Quality improvement**: Systems evaluations
- **Knowledge generation**: Collecting and documenting data for clinical trials and registries

**Trained workforce**
Registered Dietitian Nutritionist (RDN) with
- Specialized training in genetic metabolic disorders
- Established networks
  - DME/pharmacy
  - Community providers
  - Schools

**Chronic and critical phases, e.g., pregnancy, illness, hospitalizations**
Trained Workforce in Nutrition & Genetics: Paving the way to Precision Nutrition in IMDs

- Medical foods
  - specialized formulas

- Low Protein Modified Foods

- Regular foods/
  - Intact protein
Roles and Responsibilities

**Clinical and Public Health**
- Outpatient, community health, inpatient
- Newborn screening coordinators
- Participation with regional genetic networks
- State newborn screening advisory boards

**Research**
- Clinical trials and patient registries
- Independent researchers
- Industry-sponsored investigator-initiated protocols

**Education**
- Patient and family post diagnosis
- Parent organizations
- Academia and medicine

**Industry**
- Sales and marketing
- Medical science liaisons, educators
- Researchers

**Government**
- FDA
- NIH
Where do we work?

• University medical center (56%)
• Public hospital/medical facility (20%)
• Private hospital/medical facility (12%)
• Industry (20% list serv members)

How are we funded?

• Hospitals (fee for service, salaried)
• State health departments
• Newborn screening contracts
• Fees for multidisciplinary team visits
Challenges

• Increase trained workforce in this expanding field
• Unmet needs for patient education and nutrition follow-up
• Overburdened care coordination
• Need for clarity in the roles of genetic metabolic dietitians; developing core competencies
• Retaining and promoting skilled dietitians
• Enhancing leadership opportunities
• Independent reimbursement

Underpaid and overworked!
Workforce Issues

- Lack of qualified RDNs
  - Patient care ratio: 133:1
  - Disparity between earnings and responsibilities (30%)
    - Inadequate reimbursement for MNT
    - Mean salary
      - RDN (AND 2019): $68,600
      - Metabolic RDN (GMDI 2020): $70,000
      - Genetic counselor (NSGC 2020): $94,957
  - No standard certification/credential

- Uneven geographic representation
- Limited diversity within the workforce
- Inadequate reimbursement for MNT services
- Time spent on prior authorizations and advocacy for treatments vs. patient care (from survey >5 hours per week)
Where do we go from here?

• Support nutrition services and MNT to individuals with genetic metabolic disorders
  - Enhance and diversify the nutrition workforce
  - Increase telemedicine technical support and funding to increase access to services

• Add access to genetic metabolic dietitian and medical foods as quality indicators for NBS programs

• Include nutrition data in LTFU patient registries to generate knowledge and inform practice

• Offer funding/grant opportunities to support training programs and educational activities to prepare current and future workforce
  - ECHO nutrition
  - Web-based curricula
  - Face-to-face workshops
  - Post Master’s/graduate fellowships
Thank you!

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GMDI – Genetic Metabolic Dietitians International
https://www.gmdi.org/