Overview of Metabolic Disorders

WITH WIC FOCUS

Becky J Whittemore, FNP-BC
MN, MPH
• Describe general metabolic disorders and the resulting nutritional needs
• Explain appropriate guidance of WIC foods for specific metabolic disorders
• Tell how to coordinate care for shared patients
• Discuss the outcome of discontinuation of metabolic formulas in the WIC formulary
• Identify additional training or tools required
What are Metabolic Disorders?

- Genetic Disorders that affect the metabolism of food
  - Food that is not broken down properly may produce chemicals that build up in various parts of the body, causing medical problems and learning problems
  - Missing or defective enzymes (proteins) necessary to metabolize food
- Inherited disorders
  - Each parent is a “carrier” of a non-working trait that is passed to the child
- Prompt and proper treatment can prevent or lessen symptoms
### Types of Metabolic Disorders

#### Protein Disorders
- **Amino Acids**
  - Phenylketonuria
  - Maple Syrup Urine Disease
- **Organic Acids**
  - Methylmalonic Aciduria
  - Propionic Aciduria
- **Urea Cycle**
  - Citrullinemia
  - Argininosuccinic Aciduria

#### Carbohydrate Disorders
- Galactosemia
- Glycogen Storage Disease

#### Fatty Acid Disorders
- Medium Chain Acyl CoA Dehydrogenase Deficiency
- Long Chain Acyl CoA Dehydrogenase Deficiency
- Very Long Chain Acyl CoA Dehydrogenase Deficiency
Medical management

- Typically identified as positive newborn screen
  - Referred to metabolic physician on call
    - Notify Primary Care Provider
    - Recommend intervention
    - Infant and family notified and diagnostic testing completed
Current Treatment Strategies for Metabolic Disorders

Accumulation of toxic substance?

*Restrict amount available*

Absence of important product?

*Supplement product or co-factor*

Both?

*Combine approaches*
Goals of Medical Nutrition Strategy

- Three fold approach
  - Acute/emergency management
  - Long term management
- Maintain biochemical balance
- Careful monitoring to ensure adequate nutrition (protein and calories) for growth and development
- Support social and emotional development
Amino Acid Disorder

Early diagnosis is critical for success

- Phenylketonuria (PKU)
  - Excess phenylalanine as mutation in phenylalanine hydroxalase
  - Deficiency of tyrosine
  - Deficiency of neurotransmitters dopamine and norepinephrine
Amino Acid Disorders’ Treatment: “Diet for Life”

- **Infants**
  - PHE-free formula
  - Supplemented with breast milk or regular infant formula

- **Children**
  - PHE-free formula, bars and low protein medical foods
  - Supplemented with milk or when older with low PHE natural foods
Amino Acid Disorders’ Treatment: “Diet for Life”

- Prevent phenylalanine accumulation
- Provide enough phenylalanine for normal growth using phe-free metabolic formula plus dietary restrictions and/or low protein products
- Supplement the tyrosine
- Provide adequate calories, protein, fats and carbohydrates, vitamins and minerals
Early diagnosis is critical for success

- **Arginosuccinic Aciduria**
  - *Excess arginosuccinic acid*
  - *Excess ammonia*
Urea Cycle Disorders’ Treatment:
Based on Individual Nutritional Needs

- Infants
  - Low protein formula
  - Supplemented with breast milk or regular infant formula
  - Supplement arginine and citrulline

- Children
  - Liver transplant—no need for protein restriction
  - NO liver transplant in mild cases slowly increasing protein tolerance
Urea Cycle Disorders’ Treatment

- Prevent nitrogen accumulation
- Provide enough protein and calories for normal growth using low protein metabolic formula plus dietary restrictions and/or low protein products
- Supplement arginine/citrulline
- Emergency treatment intervention (letters)
- Liver Transplant
Organic Acid Disorders

Early diagnosis is critical

- Propionic Aciduria
  - Excess 3OH Propionate
  - Excess Glycine
Disorders of Carbohydrate Metabolism– “Diet for Life”

Early diagnosis is critical for success

- Galactosemia
  - Excess galactose 1 Phosphate
Classical Galactosemia
Defective gene: GALT

Normal Liver Cells
- Galactose (from diet)
  - Galactose-1-phosphate
    - UDP-Galactose
    - Further metabolism

Mutated Liver Cells
- Galactose (from diet)
  - Galactose-1-phosphate
  - Galactose-1-P accumulates depleting the liver of phosphate needed to make ATP for cell energy needs.
  - In some patients galactose is converted to galactitol in lens cells leading to the formation of cataracts.

Lens Cells
- Galactose (not processed)
- Galactitol (dulcitol)

Galactose is derived from the diet by splitting lactose into galactose and glucose. The liver metabolizes galactose as a fuel or to make glucose.
Galactosemia’ Treatment: "Diet for Life"

- **Infants**
  - Soy based infant or elemental formula
  - Stop all breastfeeding and regular infant formula
  - Limit intake of galactose

- **Children**
  - Limit intake of galactose
  - New guidelines
    - Extra Sharp Cheese
    - Legumes
Glycogen Storage Disease

Early diagnosis is critical for success

- Glycogen Storage Disease
  - Stores excessive amount glycogen in liver
  - Hypoglycemic
Glycogen Storage Disease’ Treatment

- Prevent accumulation of too much glucose, uric acid
- Provide enough protein and calories for normal growth while limiting glucose, fructose intake
- Supplement corn starch or Glycosade
- Continues night time feeding
- Emergency treatment intervention (letters)
- Liver Transplant in some disorders
Fatty Acid Oxidation Disorders

Early diagnosis is critical for success

- Medium Chain Acyl Co A Dehydrogenase Deficiency
  - Deficiency of enzyme to breakdown Medium Chain Fats
MCAD Disease’ Treatment

- Prevent catabolic state
- Provide enough protein and calories for normal growth
- FREQUENT feedings (3 months-every 3 hours, 6 months-every 6 hours)
- Emergency treatment intervention (letters)
CHANGES in WIC and METABOLIC CLINIC INTERFACE

- WIC no longer provider of special Metabolic Formulas
- WIC continues to see families and provide guidance
- WIC continues to provide food vouchers for other foods
- If limited diet, Metabolic RDs complete form and send to Nurse Practitioner to review and sign
Shared Patients Coordination

- **Two Dietitians:**
  - Sandy Van Calcar (503) 494-5500
  - Joyanna Hansen (503) 494-4263

- **Nurse Practitioner**
  - Becky Whittemore (503) 494-2776
    - whittemb@ohsu.edu
  - GENERAL NUMBER (503) 494-7859
Next Steps ...

- Coordination of Care Needs
- Learning Needs
  - Specific Disorders
  - Specific Diet Discussion
- Other
Resources


- *An Introduction to Metabolic Disorders* at [www.emdn-mitonet.co.uk/PDF/12-metabolic.pdf](http://www.emdn-mitonet.co.uk/PDF/12-metabolic.pdf)

We don’t want anything falling through the cracks
Do not get frustrated…
Call us if you need assistance