Overview of Metabolic Disorders

WITH WIC FOCUS

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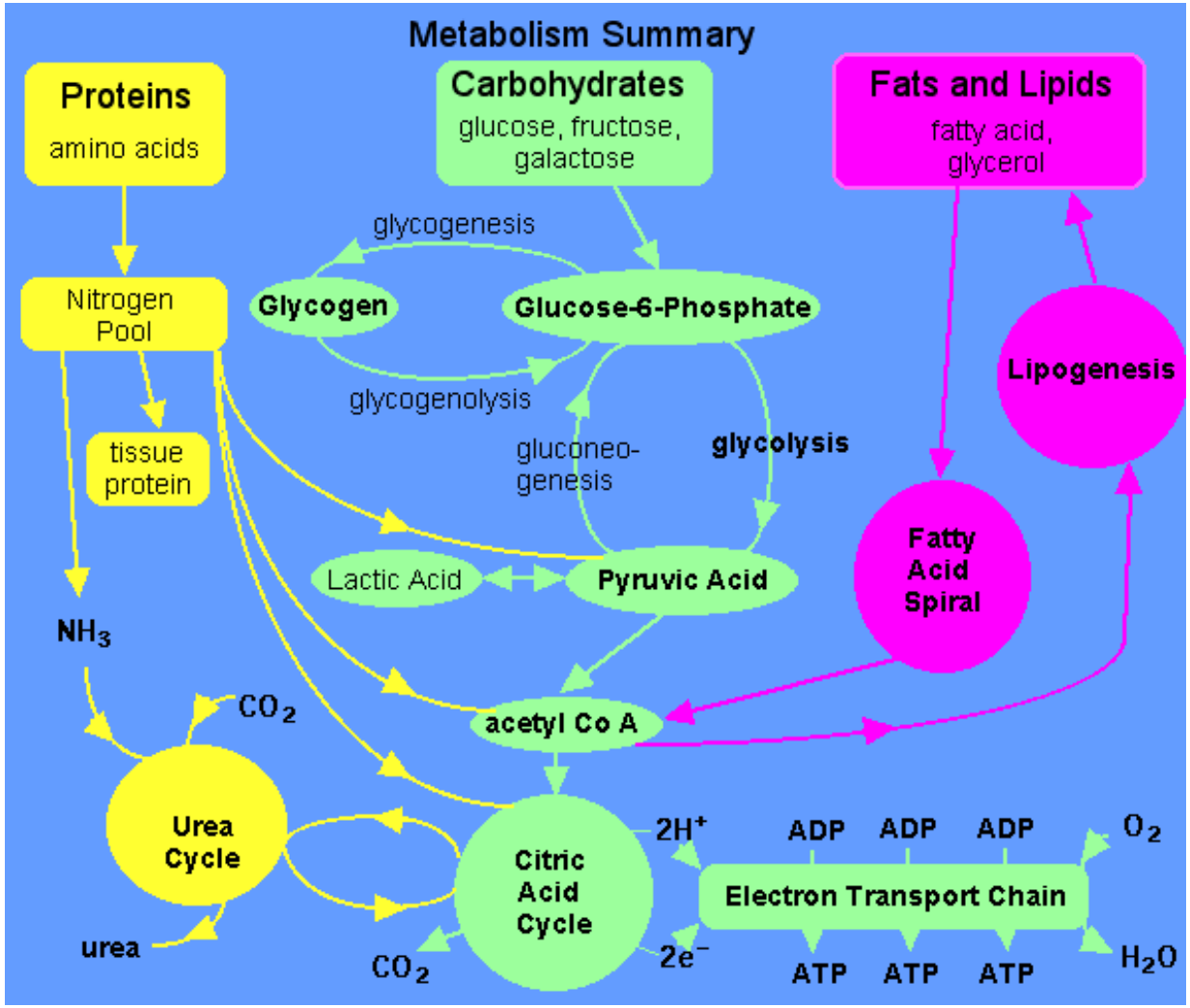
Learning Objectives

- 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

Don't need to memorize to understand


Metabolism Summary
Proteins
amino acids
Carbohydrates
glucose, fructose, galactose
Fats and Lipids
fatty acid, glycerol
glycogenesis
Nitrogen Pool
Glycogen
Glucose-6-Phosphate
Lipogenesis
glycogenolysis
tissue protein
gluconeo-genesis
glycolysis
Fatty Acid Spiral
Lactic Acid
Pyruvic Acid
NH3
CO2
acetyl Co A
Urea Cycle
Citric Acid Cycle
2H+
ADP ADP ADP
O2
Electron Transport Chain
urea
CO2
2e−
ATP ATP ATP
H2O

Types of Metabolic Disorders

Protein Disorders

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

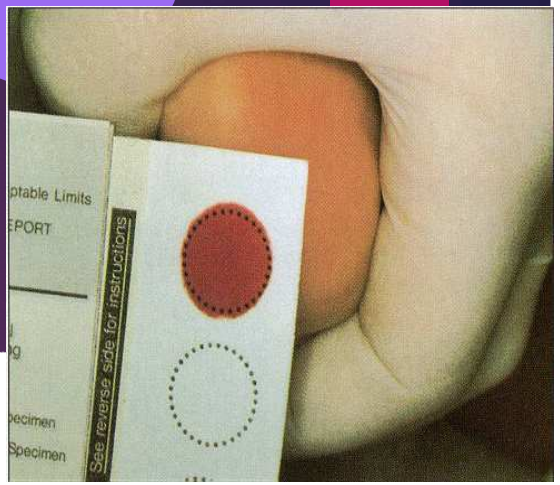
Carbohydrate Disorders

- **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

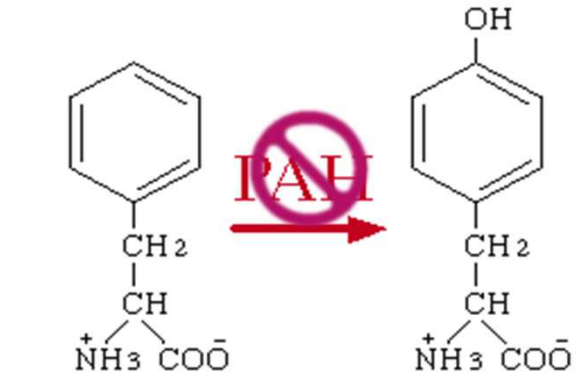
Goals of Medical Nutrition Strategy

- **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***



L-Phenylalanine L-Tyrosine

Amino Acid Disorders' Treatment: "Diet for Life"

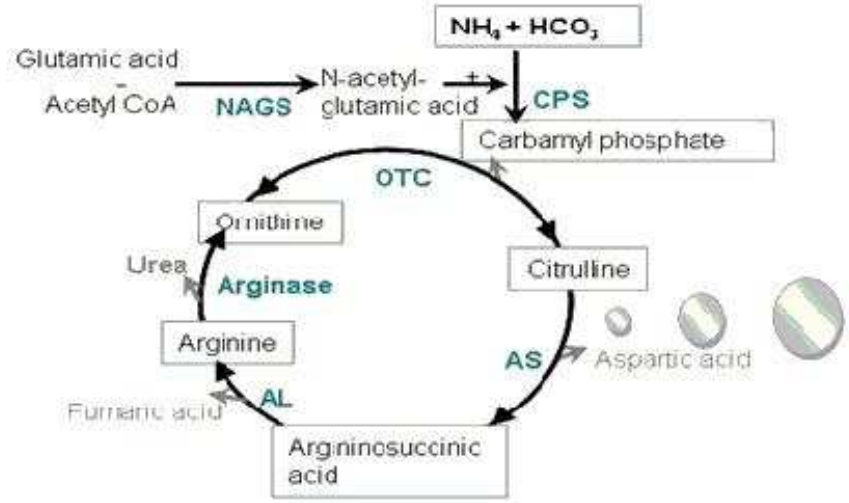
- **Infants**
  - **PHE-free formula**
  - **Supplemented with breast milk or regular infant formula**

- **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**

Urea Cycle Disorders



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**

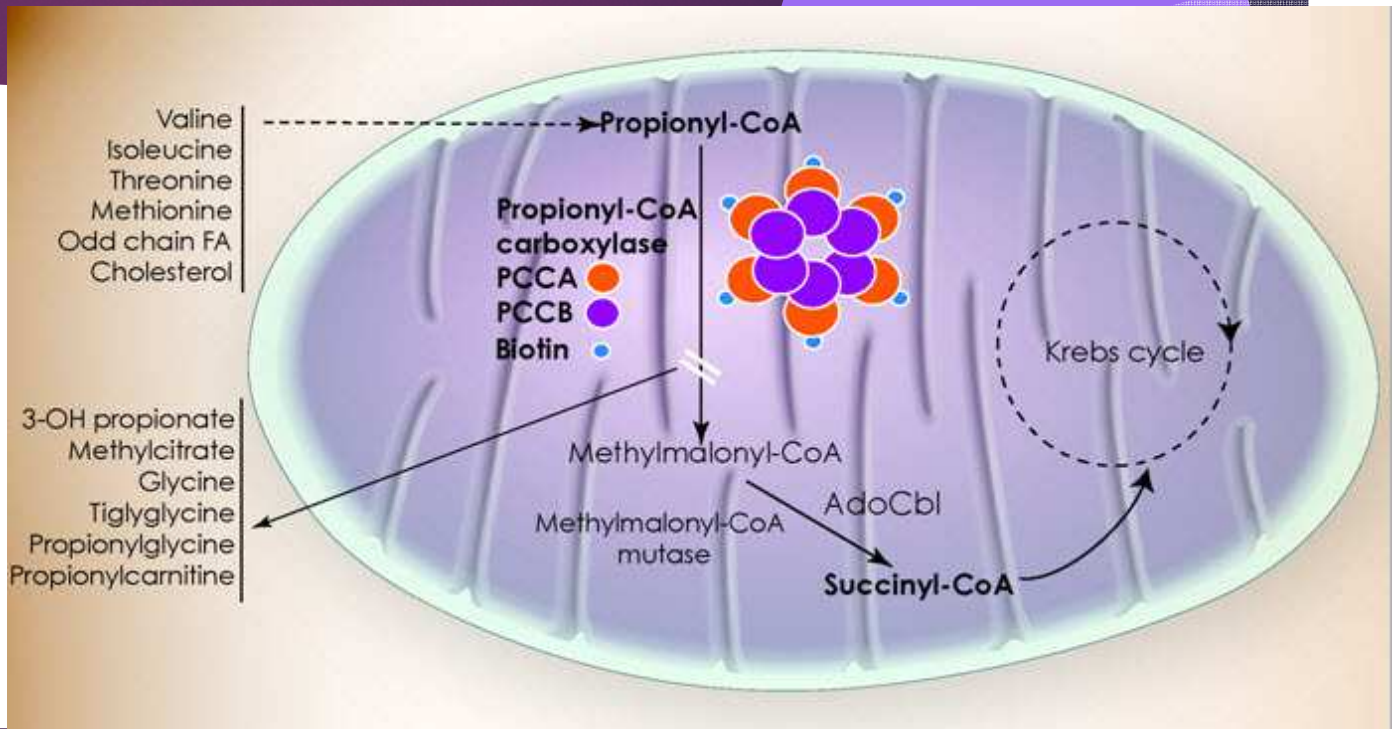
Urea Cycle Disorders' Treatment

- **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

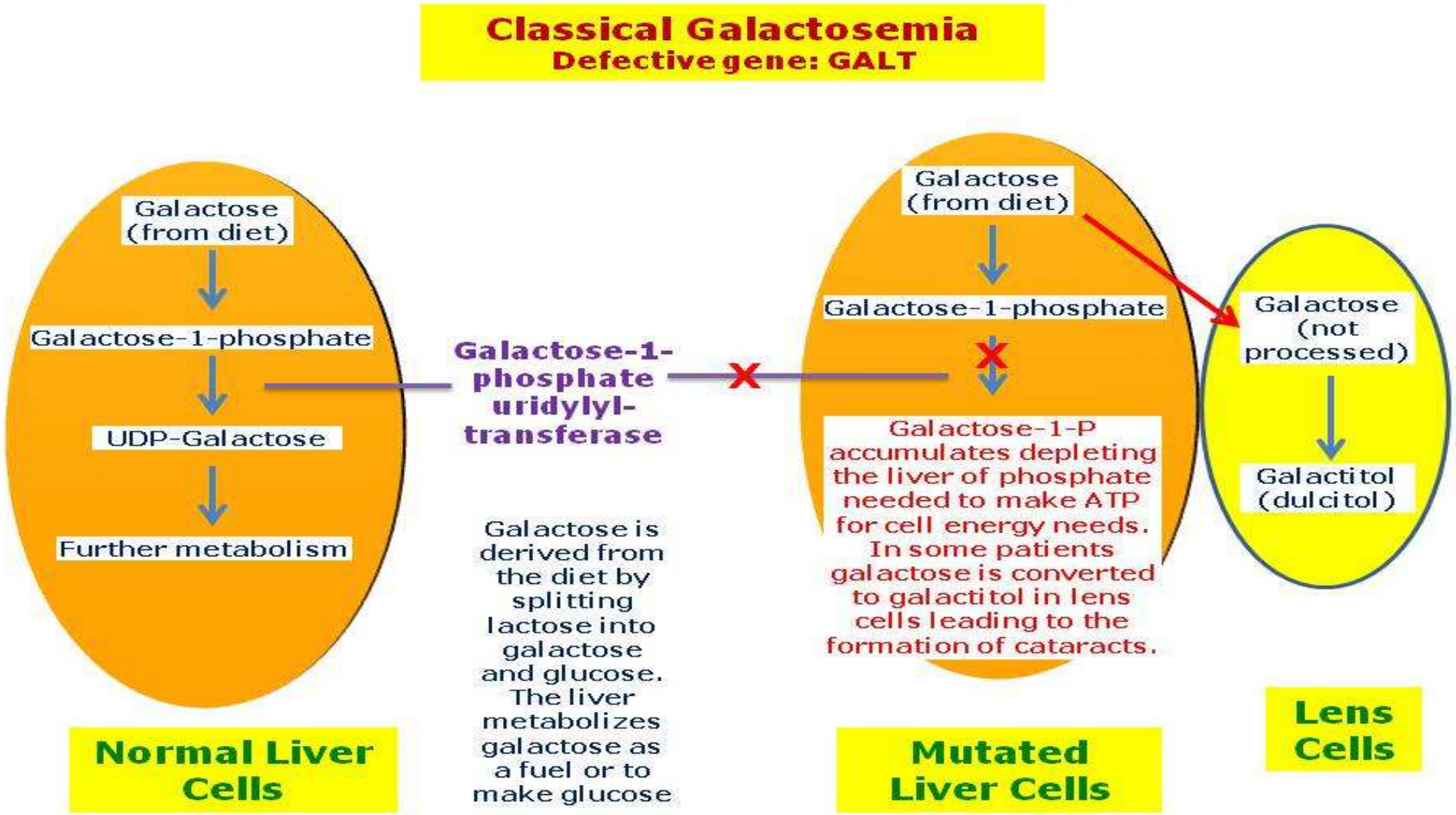
- **Propionic Aciduria**
  - ***Excess 3OH Propinate***
  - ***Excess Glycine***



Disorders of Carbohydrate Metabolism– "Diet for Life"

Early diagnosis is critical for success

- **Galactosemia**
  - Excess galactose 1 Phosphate

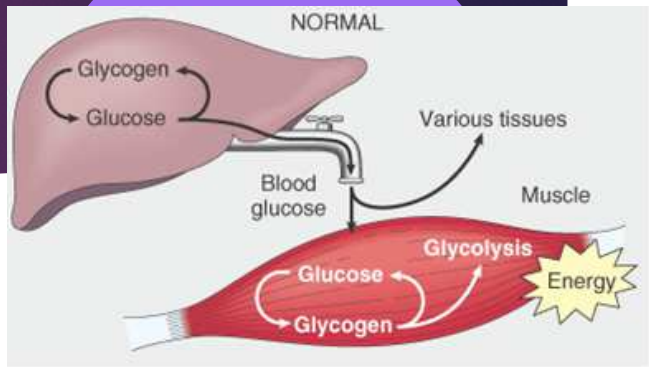

Classical Galactosemia
Defective gene: GALT
Galactose (from diet)
Galactose-1-phosphate
UDP-Galactose
Further metabolism
Galactose-1-phosphate uridylyl-transferase
Galactose is derived from the diet by splitting lactose into galactose and glucose. The liver metabolizes galactose as a fuel or to make glucose
Normal 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.
Mutated Liver Cells
Galactose (not processed)
Galactitol (dulcitol)
Lens Cells

Galatosemia' Treatment: " Diet for Life"

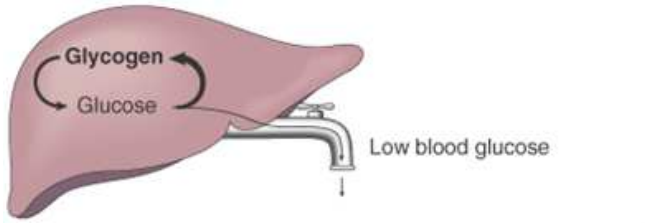
- **Infants**
  - **Soy based infant or elemental formula**
  - **Stop all breastfeeding and regular infant formula formula**
  - **Limit intake of galactose**

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

Glycogen Storage Disease



GLYCOGEN STORAGE DISEASE—HEPATIC TYPE



GLYCOGEN STORAGE DISEASE—MYOPATHIC TYPE

Early diagnosis is critical for success

- **Glycogen Storage Disease**
  - ***Stores excessive amount glycogen in liver***
  - ***Hypoglycemic***

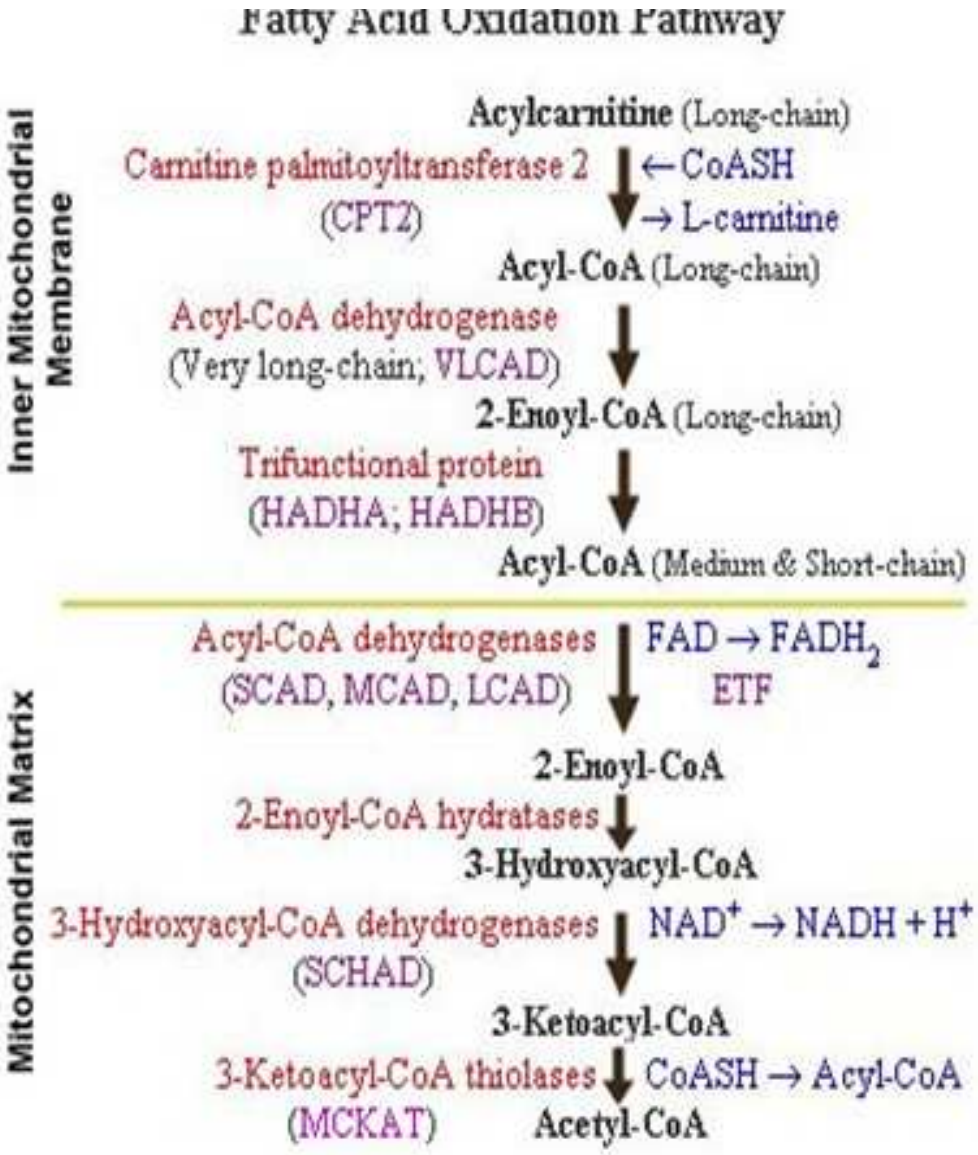
Glycogen Storage Disease' Treatment

- **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 I

Early diagnosis is critical for succes

- **Medium Chain Acyl Co A Dehydrogenase Defi**
  - ***Deficiency of enzyme to breakdown Medium C***



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

- Rice, G. M., & Steiner, R. 2016. Inborn Errors of Metabolism (Metabolic Disorders). *Pediatrics in Review*, (*37*), 3-17.
- *An Introduction to Metabolic Disorders* at www.emdn-mitonet.co.uk/PDF/12-metabolic.pdf
- Saudubray, J. M., van den Berghe, G., & Walter, J. H. (Eds.). 2012. *Inborn Metabolic Diseases,* 5th Ed., Heidelberg, Germany: Springer.

We don’t want anything falling through the cracks

THANK YOU

Do not get frustrated…
Call us if you need assistance