Diet in Metabolic Disorders
Introduction

• It’s a genetic disease

• Often, the body is missing an enzyme that is needed to process a certain type of amino acid

• As a result, these acids can build up in the body causing health problems

• Many metabolic disorders need special dietary therapy. People with metabolic disorders need ongoing counseling and monitoring by a team of physicians, nurses, genetic counselors, social workers, and dietitians for improved health and longevity.
## Disorders

<table>
<thead>
<tr>
<th>Disorders</th>
</tr>
</thead>
<tbody>
<tr>
<td>Phenylketonuria (PKU)</td>
</tr>
<tr>
<td>Maple syrup urine disease (MSUD)</td>
</tr>
<tr>
<td>Galactosemia</td>
</tr>
<tr>
<td>Endocrine disorders</td>
</tr>
<tr>
<td>• Hypothyroidism</td>
</tr>
<tr>
<td>• Hyperthyroidism</td>
</tr>
</tbody>
</table>
Phenylketonuria (PKU) – most common

- Absence of phenylalanine hydroxylase enzyme
- Inability to convert phenylalanine to tyrosine
- Tyrosine becomes conditionally essential
Symptoms occur at 3-6 months of age.
Phenylalanine

- Phenylalanine an essential amino acid
- It cannot be synthesized by body
- Needs to provide through food
PKU can be managed by???

• Dietary restriction of phenylalanine containing foods
• Controlled low-phenylalanine diet
• Levels of phenylalanine in the blood should be monitored
• Insufficient amounts leads to brain damage
• Essential requirement must be ingested each day with meal
• Care must also be taken to avoid the sweetener aspartame (L-aspartylphenylalanine) that is contained in many paediatric medicine

• Human milk has lower phenylalanine than cow’s milk include protein substitute for infant
• Assess kcal and protein needs

• Allow as much protein as possible for adequate growth from fruits, vegetables, limited amounts of grains

• Balance provided by metabolic formulas
• Growth retardation
• Bone status
• Amino acid deficiencies
• Over restriction
• Metabolic control during pregnancy
MAPLE SYRUP URINE DISORDER (MSUD)

- Inherited disorders

- Persons with this condition cannot break down the amino acids leucine, isoleucine, and valine

- Impairment of branched chain alpha keto acid dehydrogenase
• Newborns with MSUD appears normal & well
• After intake of protein containing feeds leads to
  - Seizures
  - Aponea
• If not treated death may occur, but it is manageable
• Restrict their diet to foods without leucine, isoleucine, and valine

• Must continue throughout life or symptoms will reoccur

• Supplements can be taken so that patients receive those essential amino acids

• Include orogastric feeding of branched chain amino acids free protein & energy sources within the first week of life

• Provide all other nutrients for optimal growth
Galactosemia

• Enzyme defect in galactose metabolism leading to failure to thrive, hepatomegaly, life-threatening sepsis in newborn period

  – Vomiting, jaundice upon initiation of milk feedings
  – Anorexia
  – Cirrhosis, ascites, edema, bleeding problems, enlarged spleen if milk feedings continue
Patients with galactosaemia are unable to metabolise galactose, most frequently due to a deficiency of the enzyme galactose-1-phosphate uridyl transferase.
Nutrition Interventions

• Exclusion of galactose/ lactose from diet
• Immediate reversal of symptoms results
• Exclusion of human milk, cow’s milk …
• Substitution of casein hydrolysate-containing formula
• Infant soy formulas
• Nutrition concerns
  • Provision of alternative sources of missing nutrients: vitamin D, calcium
  • Calcium supplements
  • Meet kcal, protein, vitamin and mineral needs
Hyperthyroidism

- Is a disturbance in which there is an excessive secretion of the thyroid gland with a consequent increase in the metabolic rate

Symptoms
- Weight loss
- Excessive nervousness
- Prominence of the eyes
- Enlarged thyroid gland
- Appetite is often increased
- Weakness
Modification of the diet

- High calorie (4000-5000)
- High protein diet (100-125g)
- Include snacks between meals
- Multivitamin mineral supplements are often given
- Avoid Caffeine containing foods
Hypothyroidism

- Decreased production of the thyroid hormone is known as myxedema
- Myxedema is characterized by a lowered rate of energy metabolism 30-40% below normal

Symptoms
- Muscular flabbiness
- Puffy face, eyelids & hands
- Sensitivity to cold
- Personality change to apathy & dullness
• Obesity is an occasional problem

• The energy metabolism has been reduced

• In other patients the appetite may be so poor that under nutrition results

• Increased Dietary fiber to prevent constipation

• A calorie restricted diet helps to obese patients