METABOLIC DISORDERS

Metabolic disorders

- Metabolism refers to all the chemical reactions taking place in the body to convert or use energy.
- Metabolic disorders are usually defined as inborn errors of metabolism, encompassing deficiencies in enzymes involved in the metabolism of carbohydrates, amino acids derived from proteins, and fatty acids liberated from lipids.
- There are hundreds of different genetic metabolic disorders, and their symptoms, treatments, and prognoses vary widely.

Causes of Inherited Metabolic Disorders

- In most inherited metabolic disorders, a single enzyme is either not produced by the body at all or is produced in a form that does not work.
- The code or blueprint to produce an enzyme is usually contained on a pair of genes.
- Most people with inherited metabolic disorders inherit two defective copies of the gene -- one from each parent.
- Both parents are "carriers" of the defective gene, meaning they carry one defective copy and one normal copy.

Causes of Inherited Metabolic Disorders

- In the parents, the normal gene copy compensates for the defective copy.
- Their enzyme levels are usually adequate, so they may have no symptoms of a genetic metabolic disorder.
- However, the child who inherits two defective gene copies cannot produce enough effective enzyme and develops the genetic metabolic disorder.

Lactose Intolerance

- Lactose intolerance is the inability to break down lactose.
- A person becomes lactose intolerant when his or her small intestine stops making enough of the enzyme lactase to digest and break down the lactose.
- When this happens, the undigested lactose moves into the large intestine.
- By a process called fermentation bacteria that are normally present in the large intestine interact with the undigested lactose to form lactic acid, hydrogen and other gases.

Lactose Intolerance

- **Types of Lactose Intolerance**
  - There are three main types of lactose intolerance, each with different causes:
  1. Primary Lactose Intolerance (Normal Result of Aging)
  2. Secondary Lactose Intolerance (Due to Illness or Injury)
  3. Congenital or Developmental Lactose Intolerance (Being Born with the Condition)
- The symptoms of lactose intolerance typically occur between 30 minutes and two hours after eating or drinking a milk or dairy product, and may include: Abdominal cramps, bloating, gas, diarrhea, nausea.
- Lactose intolerance can be diagnosed by these tests: blood test, stool acidity test and Hydrogen breath test.

- The mucopolysaccharidoses are a group of inherited metabolic diseases caused by the absence or malfunctioning of certain enzymes needed to break down molecules called **glycosaminoglycans** - long chains of sugar carbohydrates in each of our cells that help build bone, cartilage, tendons, corneas, skin, and connective tissue.
- Glycosaminoglycans (formerly called mucopolysaccharides) are also found in the fluid that lubricates our joints.

Hunter Syndrome

- Hunter syndrome is one type of a group of inherited metabolic disorders called mucopolysaccharidoses (MPSs), and Hunter syndrome is referred to as MPS II.
- Hunter syndrome symptoms vary and range from mild to severe. Symptoms aren't present at birth, but often begin around ages 2 to 4.

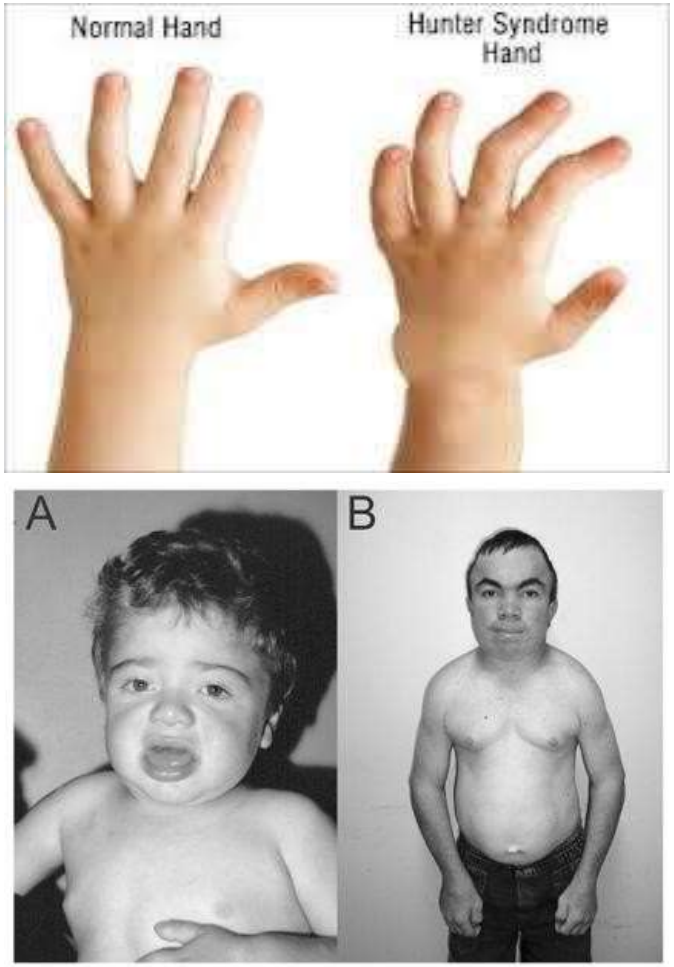

Figure 2 - Children with Hunter syndrome. A: a 2-year-old with a severe phenotype; B: an adult male with an attenuated phenotype.

Hunter Syndrome

Signs and symptoms may include:

- ✓ An enlarged head (macrocephaly)
- ✓ Thickening of the lips
- ✓ A broad nose and flared nostrils
- ✓ A protruding tongue
- ✓ A deep, hoarse voice
- ✓ Abnormal bone size or shape
- ✓ A distended abdomen
- ✓ Diarrhea
- ✓ White skin growths that resemble pebbles
- ✓ Joint stiffness
- ✓ Aggressive behavior
- ✓ Stunted growth
- ✓ Delayed development, such as late walking or talking

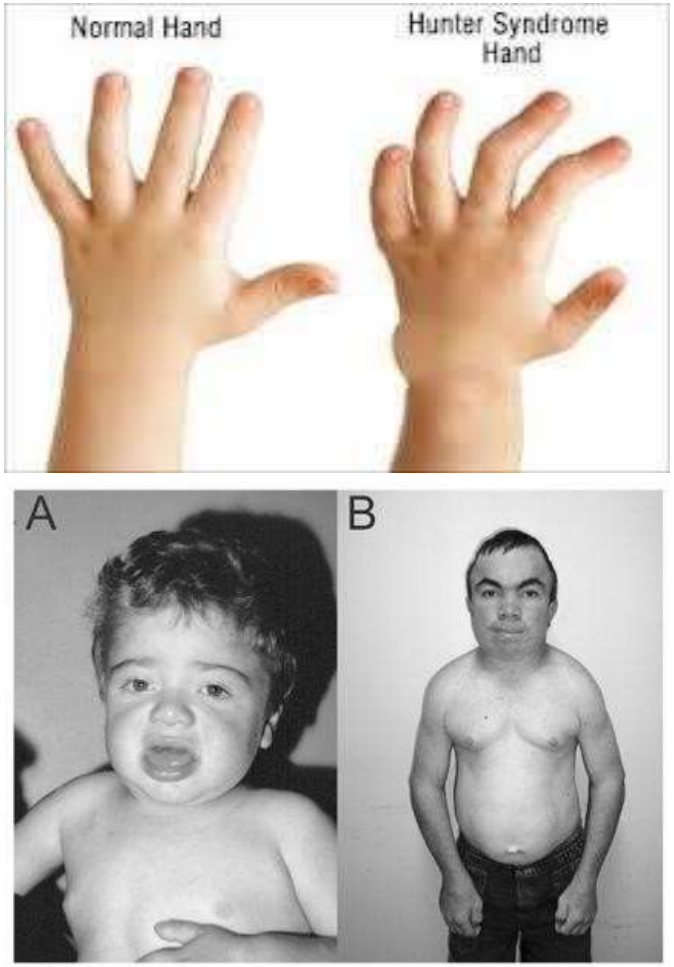

Figure 2 - Children with Hunter syndrome. A: a 2-year-old with a severe phenotype; B: an adult male with an attenuated phenotype.

Diabetes

- Diabetes mellitus (DM) is impaired insulin secretion and variable degrees of peripheral insulin resistance leading to hyperglycemia.
- Early symptoms are related to hyperglycemia and include polydipsia, polyphagia, polyuria, and blurred vision.
- Later complications include vascular disease, peripheral neuropathy, nephropathy, and predisposition to infection.

Diabetes

- There are 2 main categories of diabetes mellitus—type 1 and type 2 Diabetes.
- **Type 1 diabetes-**In type 1 diabetes mellitus (previously called juvenile-onset or insulin-dependent), insulin production is absent because of autoimmune pancreatic beta-cell destruction possibly triggered by an environmental exposure in genetically susceptible people.
- Type 1 DM generally develops in childhood or adolescence and until recently was the most common form diagnosed before age 30.

Diabetes

- **Type 2 diabetes-**In type 2 diabetes mellitus (previously called adult-onset or non–insulin-dependent), insulin secretion is inadequate because patients have developed resistance to insulin. The disease generally develops in adults and becomes more common with increasing age; up to one third of adults > age 65 have impaired glucose tolerance.
- Diagnosis is by measuring plasma glucose.
- Treatment is diet, exercise, and drugs that reduce glucose levels, including insulin and oral antihyperglycemic drugs.

Albinism

- Albinism refers to a range of disorders that result from a reduction or absence of the pigment melanin.
- The most common cause of albinism is an interruption in the functioning of the enzyme tyrosinase.
- Albinism is an autosomal recessive inheritance.

Albinism

- Primarily, albinism affects the hair, eyes, skin, and vision.
  - **Skin-**The skin will likely burn easily in the sun. It does not usually tan. Skin may have freckles, moles, usually pink in color due to the reduced quantities of pigment.
  - **Hair-**This can range in color from white to brown.
  - **Eye color-**This can also change with age and varies from very light blue to brown.
  - **Vision-**Albinism always affects vision. Changes to eye function can include: Nystagmus, Strabismus, Amblyopia, Myopia, Photophobia, Astigmatism
- An estimated 1 in 70 people carry the genes associated with albinism.
- There is no cure for albinism, but some symptoms can be treated.