

Disease associated with
carbohydrate metabolism

- The metabolism of the carbohydrates galactose, fructose, and glucose is linked through interactions between different enzymatic pathways, and disorders that affect these pathways may have symptoms ranging from mild to severe or even life-threatening.
- Clinical features include various combinations of hypoglycemia (low blood sugar), liver enlargement, diabetes (high blood sugar) and muscle pain.
- Most of these disorders can be treated, or at least controlled, with specific dietary interventions.

Disorders of carbohydrate metabolism include

1. Lactose intolerance

2. Congenital sucrase-isomaltase deficiency (CSID)

3. Galactosemia

4. Glycogen storage diseases

5. Hereditary fructose intolerance

6. Diabetes mellitus

1. Lactose intolerance

- Lactose of milk cannot be hydrolysed due to deficiency of lactase
- Accumulation of lactose in intestinal tract, which is “osmotically active” & holds water, producing diarrhoea.
- Accumulated lactose is also fermented by intestinal bacteria which produce gas & other products, producing flatulence & abdominal pain

2. Congenital sucrase-isomaltase deficiency (CSID)

- Congenital sucrase-isomaltase deficiency (CSID) is a rare inherited metabolic disorder characterized by the deficiency or absence of the enzymes sucrase and isomaltase.
- Symptoms may include watery diarrhea resulting in abnormally low levels of body fluids (dehydration), abdominal swelling (distension), and/or abdominal discomfort. In addition, some affected infants may experience malnutrition, resulting from malabsorption of essential nutrients, and/or a delay in growth and weight gain.

3. Galactosemia

- Galactosemia is a rare, hereditary disorder of carbohydrate metabolism that affects the body's ability to convert galactose (a sugar contained in milk, including human mother's milk) to glucose (a different type of sugar).
- The disorder is caused by a deficiency of an enzyme galactose-1-phosphate uridylyl transferase (GALT) which is vital to this process.
- A metabolite that is toxic to the liver and kidneys builds up. The metabolite also damages the lens of the eye, causing cataracts.

4. Glycogen storage diseases

- Glycogen storage diseases are carbohydrate metabolism disorders that occur when there is a defect in the enzymes that are involved in the metabolism of glycogen, often resulting in growth abnormalities, weakness, a large liver, low blood sugar, and confusion.
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- Glycogen storage diseases are caused by the lack of an enzyme needed to change glucose into glycogen and break down glycogen into glucose.
- Typical symptoms include weakness, sweating, confusion, kidney stones, a large liver, low blood sugar, and stunted growth.

5. Hereditary fructose intolerance

- Hereditary fructose intolerance is a carbohydrate metabolism disorder that is caused by a lack of the enzyme needed to metabolize fructose.
- Very small amounts of fructose cause low blood sugar levels and can lead to kidney and liver damage.
- Fructose intolerance disorders occur when parents pass the defective genes that cause these disorders on to their children.
- Typical symptoms include low blood sugar, sweating, confusion, and kidney damage.

6. Diabetes mellitus

- Diabetes mellitus refers to a group of diseases that affect how your body uses blood sugar (glucose). Glucose is vital to your health because it's an important source of energy for the cells that make up your muscles and tissues. It's also your brain's main source of fuel.
- Chronic diabetes conditions include type 1 diabetes and type 2 diabetes. Potentially reversible diabetes conditions include prediabetes and gestational diabetes. Prediabetes occurs when your blood sugar levels are higher than normal, but not high enough to be classified as diabetes

If you have diabetes:

- Your pancreas doesn't make any insulin or enough insulin.

Or

- Your pancreas makes insulin but your body's cells don't respond to it and can't use it as it normally should.
- If glucose can't get into your body's cells, it stays in your bloodstream and your blood glucose level rises.

Some of the signs and symptoms of type 1 diabetes and type 2 diabetes are:

- Increased thirst
- Frequent urination
- Extreme hunger
- Unexplained weight loss
- Presence of ketones in the urine (ketones are a byproduct of the breakdown of muscle and fat that happens when there's not enough available insulin)
- Fatigue
- Irritability
- Blurred vision
- Slow-healing sores