URL of this page: //medlineplus.gov/ency/article/000359.htm

Hereditary fructose intolerance

Hereditary fructose intolerance is a disorder in which a person lacks the protein needed to break down fructose. Fructose is a fruit sugar that naturally occurs in the body. Man-made fructose is used as a sweetener in many foods, including baby food and drinks.

Causes

This condition occurs when the body is missing an enzyme called aldolase B. This substance is needed to break down fructose.

If a person without this substance eats fructose or sucrose (cane or beet sugar, table sugar), complicated chemical changes occur in the body. The body cannot change its stored form of sugar (glycogen) into glucose. As a result, blood sugar falls and dangerous substances build up in the liver.

Hereditary fructose intolerance is inherited, which means it can be passed down through families. If both parents carry a nonworking copy of the aldolase B gene, each of their children has a 25% (1 in 4) chance of being affected.

Symptoms

Symptoms can be seen after a baby starts eating food or formula.

The early symptoms of fructose intolerance are similar to those of galactosemia (inability to use the sugar galactose). Later symptoms relate more to liver disease.

Symptoms may include:

- Convulsions
- Excessive sleepiness
- Irritability
- Yellow skin or whites of the eyes (jaundice)
- Poor feeding and growth as a baby, failure to thrive
- Problems after eating fruits and other foods that contain fructose or sucrose
- Vomiting

Exams and Tests

Physical examination may show:

- Enlarged liver and spleen
- Jaundice

Tests that confirm the diagnosis include:

- Blood clotting tests
- Blood sugar test
- Enzyme studies
- Genetic testing
- Kidney function tests
- Liver function tests
- Liver biopsy
- Uric acid blood test
- Urinalysis

Blood sugar will be low, especially after receiving fructose or sucrose. Uric acid levels will be high.

Treatment

Removing fructose and sucrose from the diet is an effective treatment for most people. Complications may be treated. For example, some people can take a medicine to lower the level of uric acid in their blood and decrease their risk for gout.

Outlook (Prognosis)

Hereditary fructose intolerance may be mild or severe.

Avoiding fructose and sucrose helps most children with this condition. The prognosis is good in most cases.

A few children with a severe form of the disease will develop severe liver disease. Even removing fructose and sucrose from the diet may not prevent severe liver disease in these children.

How well a person does depends on:

- How soon the diagnosis is made
- How soon fructose and sucrose can be removed from the diet
- How well the enzyme works in the body

Possible Complications

These complications may occur:

- Avoidance of fructose-containing foods due to their effects
- Bleeding
- Gout
- Illness from eating foods containing fructose or sucrose
- Liver failure
- Low blood sugar (hypoglycemia)
- Seizures
- Death

When to Contact a Medical Professional

Contact your health care provider if your child develops symptoms of this condition after feeding starts. If your child has this condition, experts recommend seeing a doctor who specializes in biochemical genetics or metabolism.

Prevention

Couples with a family history of fructose intolerance who wish to have a baby may consider genetic counseling.

Most of the damaging effects of the disease can be prevented by decreasing fructose and sucrose intake.

Alternative Names

Fructosemia; Fructose intolerance; Fructose aldolase B-deficiency; Fructose-1, 6-bisphosphate aldolase deficiency

References

Bonnardeaux A, Bichet DG. Inherited disorders of the renal tubule. In: Yu ASL, Chertow GM, Luyckx VA, Marsden PA, Skorecki K, Taal MW, eds. *Brenner and Rector's The Kidney*. 11th ed. Philadelphia, PA: Elsevier; 2020:chap 44.

Kishnani PS, Chen Y-T. Defects in metabolism of carbohydrates. In: Kliegman RM, St. Geme JW, Blum NJ, Shah SS, Tasker RC, Wilson KM, eds. *Nelson Textbook of Pediatrics*. 21st ed. Philadelphia, PA: Elsevier; 2020:chap 105.

Mojica A, Weinstock RS. Carbohydrates. In: McPherson RA, Pincus MR, eds. *Henry's Clinical Diagnosis and Management by Laboratory Methods*. 24th ed. Elsevier; 2022:chap 17.

Scheinman SJ. Genetically based kidney transport disorders. In: Gilbert SJ, ed. *National Kidney Foundation's Primer on Kidney Disease*. 8th ed. Philadelphia, PA: Elsevier; 2023:chap 37.

Review Date 4/24/2023

Updated by: Anna C. Edens Hurst, MD, MS, Associate Professor in Medical Genetics, The University of Alabama at Birmingham, Birmingham, AL. Review provided by VeriMed Healthcare Network. Also reviewed by David C. Dugdale, MD, Medical Director, Brenda Conaway, Editorial Director, and the A.D.A.M. Editorial team.