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Hartnup disorder

Hartnup disorder is a genetic condition in which there is a defect in the transport of certain amino acids (such as tryptophan and histidine) by the small intestine and kidneys.

Causes

Hartnup disorder is a metabolic condition involving amino acids. It is an inherited condition. This condition occurs due to non-working copies of the *SLC6A19* gene. A child must inherit a copy of the variant gene from both parents to be seriously affected.

The condition most often appears between ages 3 to 9 years.

Symptoms

Most people show no symptoms. If symptoms do occur, they most often appear in childhood and may include:

- Diarrhea
- Mood changes
- Nervous system (neurologic) problems, such as abnormal muscle tone and uncoordinated movements
- Red, scaly skin rash, usually when skin is exposed to sunlight
- Sensitivity to light (photosensitivity)
- Short stature

Exams and Tests

The health care provider will order a urine test to check for high levels of neutral amino acids. The levels of other amino acids may be normal.

Your provider can test for the gene that causes this condition. Biochemical tests may also be ordered.

Treatment

Treatments include:

- Avoiding sun exposure by wearing protective clothing and using a sunscreen with a protection factor of 15 or higher

- Eating a high-protein diet
- Taking supplements containing nicotinamide
- Undergoing mental health treatment, such as taking antidepressants or mood stabilizers, if mood swings or other mental health problems occur

Outlook (Prognosis)

Most people with this disorder can expect to live a normal life with no disability. Rarely, there have been reports of severe nervous system disease and even deaths in families with this disorder.

Possible Complications

In most cases, there are no complications. Complications when they occur may include:

- Changes in skin color that are permanent
- Mental health problems
- Rash
- Uncoordinated movements

Nervous system symptoms can most often be reversed. However, in rare cases they can be severe or life threatening.

When to Contact a Medical Professional

Contact your provider if you have symptoms of this condition, especially if you have a family history of Hartnup disorder. Genetic counseling is recommended if you have a personal or family history of this condition and are planning a pregnancy.

Prevention

Genetic counseling prior to marriage and conception may help prevent some cases. Eating a high-protein diet may prevent amino acid deficiencies that cause symptoms.

References

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