Bassen-Kornzweig syndrome

Bassen-Kornzweig syndrome is a rare disease passed down through families. The person is unable to fully absorb dietary fats through their intestines.

Causes

Bassen-Kornzweig syndrome is caused by changes in the *MTTP* gene that tells the body to create lipoproteins (molecules of fat combined with protein). These variants make it hard for the body to properly digest fat and essential vitamins.

Symptoms

Symptoms include:

- Balance and coordination difficulties
- Curvature of spine
- Decreased vision that gets worse over time
- Developmental delay
- Failure to thrive (grow) in infancy
- Muscle weakness
- Poor muscle coordination that usually develops after age 10
- Protruding abdomen
- Slurred speech
- Stool abnormalities, including fatty stools that appear pale in color, frothy stools, and abnormally foul-smelling stools

Exams and Tests

There may be damage to the retina of the eye (retinitis pigmentosa).

Tests that may be done to help diagnose this condition include:

- Apolipoprotein B blood test
- Blood tests to look for vitamin deficiencies (fat-soluble vitamins A, D, E, and K)

- "Burr-cell" malformation of the red cells (acanthocytosis)
- Complete blood count (CBC)
- Cholesterol studies
- Electromyography
- Eye exam
- Nerve conduction velocity
- Stool sample analysis

Genetic testing may be available for genetic variants in the *MTTP* gene.

Treatment

Treatment involves large doses of vitamin supplements containing fat-soluble vitamins (vitamin A, vitamin D, vitamin E, and vitamin K).

Linoleic acid supplements are also recommended.

People with this condition should talk to a dietitian. Diet changes are needed to prevent stomach problems. This may involve limiting intake of some types of fat.

Supplements of medium-chain triglycerides are taken under the supervision of a health care provider. They should be used with caution, because they may cause liver damage.

Outlook (Prognosis)

How well a person does depends on the amount of brain and nervous system problems.

Possible Complications

Complications may include:

- Blindness
- Mental deterioration
- Loss of function of peripheral nerves, uncoordinated movement (ataxia)

When to Contact a Medical Professional

Contact your provider if your infant or child has symptoms of this disease. Genetic counseling can help families understand the condition and the risks of inheriting it, and learn how to care for the person.

Prevention

High doses of fat-soluble vitamins may slow the progression of some problems, such as retina damage and decreased vision.

Alternative Names

Abetalipoproteinemia; Acanthocytosis; Apolipoprotein B deficiency

References

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