CONDITION

Bassen-kornzweig syndrome

Learn how Bassen-Kornzweig syndrome affects fat absorption, growth, nerves, and vision, and how nutrition and vitamin treatment help prevent complications.

WHAT IT IS
Rare inherited fat absorption disorder

BODY SYSTEM
Digestive system

MAIN SYMPTOMS
Chronic diarrhea, greasy stools, poor growth, balance problems

WHAT CAUSES IT
Inherited MTTP gene changes

HOW IT IS DIAGNOSED
MTTP genetic testing

MAIN TREATMENT
Nutrition support and vitamin replacement

CAN IT BE CURED?
No, lifelong treatment controls complications

IS IT AN EMERGENCY?
Sometimes, bleeding that will not stop

LONG-TERM OUTLOOK
Improved by early treatment, with advanced damage potentially irreversible

Overview

Bassen-Kornzweig syndrome is a rare inherited condition that prevents the body from properly absorbing and transporting certain fats. It is also called abetalipoproteinemia .

In this condition, the body cannot make normal amounts of several lipoproteins. Lipoproteins are particles that carry cholesterol, triglycerides, and fat-soluble vitamins through the bloodstream. The affected particles contain a protein called apolipoprotein B, or apoB.

Without these particles, fat and the vitamins A, D, E, and K are not absorbed or distributed normally. These are called fat-soluble vitamins because the body needs dietary fat to absorb them.

Symptoms often begin in infancy with diarrhea, greasy stools, vomiting, or poor growth. Over

time, especially without treatment, vitamin deficiencies can damage the nerves and the light-sensitive tissue at the back of the eye. Problems with balance, muscle control, sensation, and vision may develop.

Early recognition matters because nutrition support and vitamin replacement can improve growth and reduce the risk of long-term complications. Treatment usually continues throughout life and is coordinated by clinicians experienced in inherited metabolic or lipid disorders.

Symptoms

The timing and severity of symptoms vary. Digestive and growth problems often appear first, while nerve and eye problems may develop later.

Infants and young children may have:

Greasy stools are called steatorrhea . They occur when unabsorbed fat passes into the stool.

A blood test or blood smear may show acanthocytes . These are red blood cells with an irregular, spiky shape. Acanthocytes are a characteristic clue, although they can occur in other medical conditions.

Vitamin E deficiency is an important cause of later symptoms. Vitamin E protects nerve and cell membranes from damage. Long-term deficiency may cause:

Vitamin A and E deficiencies may affect the retina, the part of the eye that senses light. A person may first have difficulty seeing in dim light. Peripheral, or side, vision may gradually narrow. Further vision loss can occur if retinal damage progresses.

Other possible findings include anemia, low bone density, or easy bruising and bleeding. Bleeding may be related to vitamin K deficiency because vitamin K helps the blood clot normally.

Symptoms alone cannot confirm Bassen-Kornzweig syndrome. Several other conditions can cause poor fat absorption, very low cholesterol, growth concerns, or neurologic symptoms.

- Chronic diarrhea or frequent loose stools
- Pale, greasy, bulky, or unusually foul-smelling stools
- A swollen abdomen
- Vomiting
- Poor weight gain or slower-than-expected growth

- Difficulty tolerating foods with larger amounts of fat
- Reduced or absent reflexes
- Muscle weakness
- Numbness or reduced ability to sense vibration and body position
- An unsteady walk
- Poor coordination, known as ataxia
- Tremor or difficulty with fine movements
- Changes in speech

Causes and risk factors

Bassen-Kornzweig syndrome is caused by disease-causing changes in both copies of the MTTP gene. This gene provides instructions for making microsomal triglyceride transfer protein. That protein is needed to assemble apoB-containing lipoproteins in the intestine and liver.

When the protein does not work properly, the intestine cannot package absorbed fat into particles called chylomicrons. The liver also cannot make normal very-low-density lipoprotein, or VLDL. As a result, blood levels of triglycerides, low-density lipoprotein cholesterol, and apoB are extremely low or nearly absent.

The condition follows an autosomal recessive inheritance pattern. This means an affected person has inherited one altered MTTP gene from each parent. Parents usually carry one altered copy without having symptoms.

When both parents are carriers, each pregnancy has:

These chances apply separately to each pregnancy. They do not predict what will happen in a particular family.

The main risk factor is having two parents who carry an altered MTTP gene. The likelihood that both partners carry the same rare gene change may be higher when they are biologically related or come from a small population in which a particular variant is more common.

The condition is inherited. It is not caused by a person's diet, parenting, or lifestyle.

- A 25% chance that the child will have the condition
- A 50% chance that the child will be an unaffected carrier
- A 25% chance that the child will inherit neither altered copy

Diagnosis

Diagnosis usually begins with a review of symptoms, growth, diet, family history, and laboratory results. A clinician may ask about diarrhea, stool appearance, balance, vision, bleeding, and whether relatives have very low cholesterol or similar symptoms.

Testing may include:

A person with Bassen-Kornzweig syndrome typically has extremely low total cholesterol, triglycerides, LDL cholesterol, and apoB. However, laboratory patterns must be interpreted in context.

Genetic testing can identify disease-causing changes in both copies of MTTP and confirm the diagnosis. A genetic counselor can help explain the results, inheritance, and testing options for relatives.

Eye testing may include a retinal examination, visual-field testing, and an electroretinogram, which measures how the retina responds to light. A neurologic evaluation can assess reflexes, strength, sensation, walking, and coordination.

An intestinal biopsy may show fat collected inside intestinal cells. It is now needed less often when the clinical and genetic findings are clear.

Other causes of very low apoB levels must be considered. These include familial hypobetalipoproteinemia and chylomicron retention disease. Conditions affecting the pancreas, liver, or intestine can also cause fat malabsorption, but their lipid and genetic findings differ.

- A fasting lipid panel to measure cholesterol and triglycerides
- Measurement of apoB
- Blood levels of vitamins A, D, and E
- Clotting tests that can show the effects of vitamin K deficiency
- A complete blood count and blood smear to look for anemia and acanthocytes
- Liver enzyme tests
- Tests of stool fat when malabsorption needs to be assessed

Treatment and management

Treatment focuses on supporting growth, reducing digestive symptoms, replacing deficient vitamins, and protecting the eyes and nervous system. Care may involve a metabolic specialist,

lipid specialist, gastroenterologist, dietitian, neurologist, and eye specialist.

A reduced-fat eating plan can decrease diarrhea and greasy stools. However, children still need enough calories, protein, and nutrients to grow. A dietitian familiar with fat-malabsorption disorders can help balance these needs.

Medium-chain triglycerides may sometimes be used as an energy source because they are absorbed differently from most dietary fats. They do not provide all essential fatty acids, so they are not a complete replacement for other fats. The care team may plan specific sources of essential fatty acids while limiting fats that are harder to absorb.

Replacement of vitamins A, D, E, and K is central to treatment. Vitamin E is especially important for protecting nerve function. People with this condition may require forms or amounts different from routine over-the-counter supplements because absorption is impaired.

Vitamin treatment should be managed with laboratory monitoring. High amounts of fat-soluble vitamins can build up in the body and cause harm. Vitamin A needs particular care during pregnancy because excessive exposure can harm a developing fetus. Patients should not increase, stop, or combine supplements without discussing the plan with their clinical team.

Additional management may include:

Treatment started early may prevent or limit some complications. Nerve or retinal damage that is already advanced may not fully reverse, which makes ongoing treatment and monitoring important even when digestive symptoms improve.

- Nutrition support for poor growth
- Physical or occupational therapy for weakness, balance, or coordination problems
- Mobility or safety aids when needed
- Vision support and low-vision services
- Treatment of anemia, bone disease, or bleeding problems when present
- Support at school or work for vision or movement limitations

Complications

The main long-term complications result from poor fat absorption and chronic vitamin deficiency.

Neurologic complications can include peripheral neuropathy, loss of reflexes, muscle weakness, impaired position sense, and progressive ataxia. These changes can affect walking, hand control, speech, and independence.

Retinal degeneration can lead to night blindness, narrowing of side vision, and more severe visual impairment. Regular eye assessment can identify changes before a person notices major vision loss.

Vitamin K deficiency may interfere with normal blood clotting. Easy bruising, nosebleeds, bleeding gums, or prolonged bleeding may occur. Vitamin D deficiency can contribute to weak bones, while poor nutrition may affect growth and muscle health.

Fat can collect in the liver even though blood lipid levels are very low. Some people develop an enlarged liver, abnormal liver tests, or progressive liver disease. Liver health therefore remains part of follow-up.

Routine concerns such as increasing diarrhea, gradual changes in balance, or declining night vision should be reported promptly to the usual care team. They generally require timely clinical review rather than emergency services.

Seek urgent medical advice for new jaundice, persistent vomiting, rapidly worsening weakness, a sudden change in vision, or bleeding that is unusual for the person.

Call emergency services for bleeding that will not stop, vomiting blood, black or bloody stools, severe breathing difficulty, fainting, a seizure, or sudden severe neurologic symptoms. A serious head injury also needs prompt assessment when a clotting problem is possible.

Follow up

Bassen-Kornzweig syndrome requires lifelong follow-up. The exact schedule depends on age, symptoms, laboratory results, and response to treatment.

Regular visits may assess:

Laboratory results help the clinical team adjust vitamin and nutrition plans while avoiding both deficiency and excess. Taking supplements consistently is important, even when the person feels well.

New problems with balance, sensation, strength, night vision, bruising, or stool patterns should be reported between planned visits. These changes may mean treatment or testing needs review.

Relatives may benefit from genetic counseling or targeted testing after a diagnosis is confirmed. People planning a pregnancy can discuss carrier testing, reproductive options, and safe vitamin management with genetics and obstetric specialists.

- Height, weight, growth, and overall nutrition

- Digestive symptoms and dietary intake
- Blood counts, clotting, and fat-soluble vitamin levels
- Liver enzymes and other measures of liver health
- Muscle strength, reflexes, sensation, walking, and coordination
- Vision and retinal function
- Bone health when clinically appropriate

Sources

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