CONDITION

Abercrombie syndrome

Learn how the historical term Abercrombie syndrome relates to amyloidosis, including its symptoms, diagnosis, treatment, complications, and follow-up care.

WHAT IT IS
Disease caused by abnormal amyloid deposits

BODY SYSTEM
Heart and circulation, kidneys, nervous and digestive systems

MAIN SYMPTOMS
Tiredness, shortness of breath, swelling, numbness or tingling

WHAT CAUSES IT
Abnormally folded proteins from varied causes

HOW IT IS DIAGNOSED
Biopsy with Congo red staining and protein typing

MAIN TREATMENT
Type-specific protein-targeting treatment and supportive care

CAN IT BE CURED?
No, damage may not fully reverse

IS IT AN EMERGENCY?
Sometimes, with severe difficulty breathing

LONG-TERM OUTLOOK
Variable, from slow progression to irreversible organ damage

Overview

Abercrombie syndrome is a historical name for amyloidosis . It is not usually used as a diagnosis in modern medical practice. If this term appears in an older medical record, it generally refers to disease caused by abnormal amyloid deposits in body tissues.

Amyloid forms when certain proteins fold into an abnormal shape and collect outside cells. These deposits can interfere with how an organ works. Amyloidosis may affect one area, called localized amyloidosis, or several organs, called systemic amyloidosis.

Systemic amyloidosis is not a single disease. Doctors classify it by the protein forming the deposits. The exact type matters because the causes, treatments, and outlook differ greatly. Important forms include:

The heart, kidneys, nerves, liver, and digestive system are among the organs that may be affected. Some forms progress slowly, while others can damage organs more quickly. Early and accurate protein typing helps doctors choose the right treatment and avoid therapies that will not work for that form.

- AL amyloidosis , caused by abnormal antibody fragments called light chains
- ATTR amyloidosis , caused by the transthyretin protein
- AA amyloidosis , associated with long-term inflammation
- Less common forms linked to other proteins, long-term dialysis, or inherited conditions

Symptoms

Symptoms depend on the type of amyloidosis, the organs involved, and the amount of organ damage. Early symptoms may be mild or resemble those of more common conditions. Having one or more of these symptoms does not mean a person has amyloidosis.

Possible symptoms include:

Heart involvement can make the heart muscle thick and stiff. The heart may then have trouble filling with blood, even if its pumping strength appears normal. Kidney involvement may cause protein to leak into the urine and can eventually reduce kidney function. Nerve involvement may affect sensation, muscle control, digestion, blood pressure, or bladder function.

Some manifestations are subtle. For example, carpal tunnel syndrome affecting both hands may occur years before certain forms of ATTR cardiac amyloidosis become apparent. This finding is common in the general population, however, and does not establish a diagnosis by itself.

- Tiredness, weakness, or reduced ability to exercise
- Shortness of breath, especially with activity or when lying flat
- Swelling in the feet, ankles, legs, or abdomen
- Unexplained weight loss or poor appetite
- Foamy urine, which can be a sign of protein loss through the kidneys
- Numbness, tingling, burning pain, or weakness in the hands and feet
- Dizziness when standing because of a drop in blood pressure
- Diarrhea, constipation, nausea, or feeling full after eating a small amount
- Easy bruising, including bruising around the eyes

- Enlargement of the tongue, although this is uncommon and mainly associated with AL amyloidosis
- Irregular or unusually slow or fast heartbeats
- Carpal tunnel syndrome, which can cause hand numbness and weakness

Causes and risk factors

In AL amyloidosis , abnormal plasma cells in the bone marrow produce light-chain proteins that form amyloid. Plasma cells are immune cells that normally make antibodies. AL amyloidosis may occur on its own or with a plasma-cell disorder such as multiple myeloma.

ATTR amyloidosis involves transthyretin, a protein made mainly in the liver. In hereditary, or variant, ATTR amyloidosis, a change in the `TTR` gene makes the protein more likely to form deposits. The pattern of heart and nerve disease varies among gene variants and families.

Wild-type ATTR amyloidosis does not result from an inherited `TTR` variant. It usually develops later in life and mainly affects the heart, although tendons and ligaments may also be involved.

AA amyloidosis occurs when long-lasting inflammation causes persistently high levels of serum amyloid A protein. It may be associated with chronic inflammatory diseases or long-standing infections. Better control of many inflammatory conditions has reduced this risk, but AA amyloidosis can still occur.

Other uncommon forms include beta-2 microglobulin amyloidosis in people who have received dialysis for many years. Certain inherited variants in other proteins may also cause systemic amyloidosis. Localized amyloid can form in places such as the skin, airway, bladder, or eye without causing systemic disease.

Risk depends on the specific form. Relevant factors may include older age, a family history of hereditary amyloidosis, a plasma-cell disorder, persistent inflammatory disease, or long-term dialysis. Most symptoms associated with amyloidosis have more common explanations, so risk factors and test results must be considered together.

Diagnosis

Because "Abercrombie syndrome" does not identify a modern amyloidosis type, a clinician reviewing that term will usually begin by clarifying the original diagnosis and looking for evidence of current organ involvement.

Evaluation may include a medical and family history, a physical examination, and tests chosen

for the person's symptoms. Blood and urine tests can assess kidney and liver function, protein loss, blood cell counts, and signs of heart strain or injury.

When AL amyloidosis is possible, doctors usually check for abnormal light chains with serum free light-chain testing and immunofixation of blood and urine. A routine blood protein test alone cannot reliably exclude AL amyloidosis.

Imaging may help assess affected organs. An electrocardiogram records the heart's electrical activity. An echocardiogram uses ultrasound to examine heart structure and function. Cardiac magnetic resonance imaging can show patterns of heart muscle injury.

A nuclear medicine bone-tracer scan can support a diagnosis of ATTR cardiac amyloidosis in carefully selected situations. Before interpreting such a scan as ATTR, clinicians must test for a monoclonal protein, which can be a sign of an abnormal plasma-cell process. A scan should not be used alone to distinguish all forms of cardiac amyloidosis.

A biopsy removes a small tissue sample for laboratory examination. The sample may come from abdominal fat, bone marrow, or an affected organ. Congo red staining can identify amyloid. Specialized analysis, often mass spectrometry, is then used to determine the protein type. Correct typing is essential because the appearance of amyloid under a microscope does not reveal its cause.

If ATTR amyloidosis is diagnosed, genetic testing can distinguish hereditary ATTR from the wild-type form. Genetic counseling can help a person understand the test and what a hereditary result may mean for biological relatives.

Treatment and management

Treatment is based on the amyloid protein type, the organs affected, and the person's overall health. Care often involves specialists in hematology, cardiology, kidney disease, neurology, or genetics.

For AL amyloidosis , treatment targets the abnormal plasma cells so they stop making harmful light chains. This usually involves combinations of plasma-cell-directed medicines. Some medically suitable patients may be considered for an autologous stem cell transplant, which uses the person's own blood-forming stem cells after intensive treatment. Treatment selection requires specialist assessment because heart, kidney, and nerve involvement can affect safety.

For ATTR amyloidosis , medicines may stabilize the transthyretin protein or reduce its production. The appropriate option depends on whether disease mainly affects the heart, nerves, or both, as well as regulatory approval and access in the patient's location. Selected

people with hereditary disease may be considered for an organ transplant, although medicines have changed the role of transplantation.

For AA amyloidosis , controlling the underlying inflammatory disease or infection is central. Lowering ongoing inflammation can reduce production of serum amyloid A and may help preserve organ function. Treatment varies according to the underlying condition.

Supportive care treats the effects of organ involvement. It may include carefully managed diuretics like furosemide or hydrochlorothiazide to reduce fluid buildup, treatment for nerve pain, nutrition support, or dialysis for severe kidney failure. Some people may be evaluated for heart or kidney transplantation.

Cardiac amyloidosis can change how the body responds to medicines commonly used for heart failure or high blood pressure. A person should not start, stop, or change these medicines without speaking with the clinician managing their amyloidosis.

Management may also include adjusting dietary sodium or fluid intake, monitoring weight, and preventing falls when low blood pressure or nerve damage is present. These plans should be individualized rather than followed as universal restrictions.

Complications

Possible complications depend on where amyloid accumulates. Heart deposits may lead to heart failure, abnormal rhythms, fainting, or blood clots. Kidney disease may cause major protein loss, swelling, or kidney failure.

Nerve damage can cause chronic pain, weakness, balance problems, or loss of normal blood-pressure control. Digestive involvement may contribute to poor nutrition, bleeding, diarrhea, constipation, or difficulty absorbing nutrients. Liver involvement can cause enlargement and reduced function.

Organ damage may continue if the amyloid-producing process is not controlled. Even after treatment lowers production of the abnormal protein, existing deposits may take time to clear, and some damage may not fully reverse. This is why follow-up includes both laboratory measures of treatment response and continued assessment of organ function.

Follow up

Follow-up is tailored to the amyloidosis type and treatment. Routine visits may include blood and urine tests, heart biomarkers, kidney testing, electrocardiograms, imaging, and checks for nerve or digestive symptoms. People receiving treatment also need monitoring for side

effects.

Daily weight tracking may be recommended when heart or kidney involvement causes fluid retention. A rapid change in weight, increasing swelling, worsening breathlessness, new palpitations, reduced urine output, or repeated dizziness should be reported promptly to the care team. These symptoms may require an earlier assessment rather than waiting for a routine appointment.

Seek emergency care for severe difficulty breathing, chest pressure, fainting that does not quickly resolve, vomiting blood, black or bloody stools, or signs of a stroke such as sudden facial drooping, arm weakness, trouble speaking, or loss of balance.

People with hereditary ATTR amyloidosis may benefit from genetic counseling. Adult biological relatives can discuss whether testing is appropriate and what a result could mean. Testing relatives should be voluntary and accompanied by clear information about possible medical, emotional, and insurance implications.

Sources

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