CONDITION

Immunodeficiency diseases

Immunodeficiency is reduced immune protection that can make infections more frequent, severe, or difficult to clear.

WHAT IT IS
Reduced immune protection against infections and some other conditions

BODY SYSTEM
Immune system

MAIN SYMPTOMS
Repeated infections, severe infections, poor growth, persistent diarrhoea

WHAT CAUSES IT
Inherited immune changes or acquired immune suppression

HOW IT IS DIAGNOSED
Medical history, blood tests, and sometimes genetic testing

MAIN TREATMENT
Infection control and therapies that replace or restore immunity

CAN IT BE CURED?
Sometimes - transplant or gene therapy corrects selected types

IS IT AN EMERGENCY?
Sometimes - breathing trouble, confusion, or signs of sepsis

LONG-TERM OUTLOOK
Many people do well with ongoing specialist care

Overview

Immunodeficiency means that part of the immune system is missing or does not work as expected. The immune system includes white blood cells, antibodies, lymph nodes, the spleen, and other tissues that protect the body from infection.

Immunodeficiency is an umbrella term covering many different conditions. Some mainly affect antibodies, which recognise germs. Others affect immune cells, proteins called complement, or several parts of the immune system at once.

A primary immunodeficiency , now often called an inborn error of immunity , results from a genetic change affecting immune function. It may be found in infancy, but milder forms can remain unnoticed until adolescence or adulthood.

A secondary immunodeficiency develops because of another condition or treatment. Possible causes include HIV, some blood cancers, poor nutrition, loss of immune proteins, and medicines that suppress immune activity.

Having frequent minor colds does not necessarily mean you have immunodeficiency. Doctors look for an unusual pattern, such as infections that are severe, repeatedly return, involve uncommon germs, or do not improve as expected.

Early recognition matters. Treatment can reduce infections, protect organs from repeated damage, and address other immune problems that may occur alongside infection.

Symptoms

Symptoms differ according to which part of the immune system is affected. They may begin in early childhood or appear much later.

Possible signs include:

Some immunodeficiencies also cause immune activity against the body's own tissues. This is called autoimmunity . It can lead to low blood cell counts, joint inflammation, bowel inflammation, or other symptoms.

Not everyone has constant infections. Some people first seek care because of swollen lymph nodes, an enlarged spleen, unexplained inflammation, or an autoimmune condition.

The number of infections alone does not establish the diagnosis. Exposure to young children, smoking, asthma, allergies, and structural problems in the lungs or sinuses can also cause repeated respiratory infections.

- Repeated ear, sinus, chest, skin, or gut infections
- Infections that are unusually severe or slow to clear
- Infections that return soon after treatment
- Infection with germs that rarely cause illness in healthy people
- Persistent diarrhoea or difficulty absorbing nutrients
- Poor growth or weight gain in a child
- Persistent mouth thrush or fungal infections
- Deep skin or organ abscesses
- A family history of serious or unusual infections

Causes and risk factors

Primary immunodeficiency is caused by a genetic change that alters immune development or function. Some changes are inherited from one or both parents. Others occur for the first time in the affected person.

A family history can increase the chance of an inherited condition, especially when relatives have had early deaths from infection, repeated severe infections, or a known immune condition. However, many people diagnosed with primary immunodeficiency have no obvious family history.

Secondary immunodeficiency is more common and is acquired later. Causes can include:

Age also affects immunity. Babies have an immune system that is still developing, while immune responses may become less effective in later life. These normal age-related changes are not automatically an immunodeficiency.

- HIV infection
- Blood cancers such as leukaemia, lymphoma, or myeloma
- Chemotherapy and some targeted cancer treatments
- Steroids and other immune-suppressing medicines
- Loss of immune proteins through certain kidney or bowel conditions
- Removal of the spleen or reduced spleen function
- Severe undernutrition
- Major burns, serious illness, or some long-term medical conditions

Diagnosis

There is no single test for every immunodeficiency. Diagnosis starts with the pattern of infections, other symptoms, medicines, medical history, and family history.

Initial blood tests may include a complete blood count, which measures different blood cells. The numbers of lymphocytes and neutrophils are particularly useful because these white blood cells have important roles in fighting infection.

Tests may measure immunoglobulins, also called antibodies. Doctors may also check whether your immune system produces protective antibodies after vaccination. This is different from simply checking whether you have received a vaccine.

Further testing can assess:

Genetic testing may confirm a specific type, but it does not identify every case. Sometimes a genetic result is uncertain and must be interpreted alongside symptoms and immune test results.

Testing often involves an immunology specialist. Repeat tests may be needed because infections, medicines, age, and laboratory differences can temporarily affect results.

- The number and types of immune cells
- How well certain white blood cells function
- Complement proteins that help antibodies destroy germs
- HIV or other possible acquired causes
- Kidney, liver, and nutritional health
- Genetic changes linked to inborn immune conditions

Treatment and management

Treatment is matched to the affected part of the immune system and the cause. The goals are to control infections, prevent avoidable complications, and replace or restore immune function when possible.

Infections may need prompt treatment with antibiotics like amoxicillin or doxycycline , antiviral medicines, or antifungal medicines. Some people with a well-defined ongoing risk are given preventive antimicrobial treatment. This is planned individually because unnecessary use can cause side effects and contribute to antimicrobial resistance.

Immunoglobulin replacement therapy supplies antibodies collected from donated human plasma. It can be given into a vein or under the skin. It helps people whose bodies cannot make enough effective antibodies, but it does not replace every part of the immune system.

Treatment for secondary immunodeficiency may include managing the underlying condition or adjusting an immune-suppressing medicine when medically appropriate. Medicines should not be stopped without advice, because sudden changes may worsen the condition they are controlling.

Some severe inherited forms can be treated with a stem cell transplant. This replaces the blood-forming cells that produce immune cells. Transplant can be curative for selected conditions, but it involves significant risks and is not suitable for every person.

Gene therapy is available for a small number of specific inherited conditions. It aims to correct a person's own blood-forming cells. Access and suitability depend on the exact genetic

diagnosis and available specialist programmes.

Vaccination plans require individual review. Inactivated vaccines do not contain live germs, although they may produce a weaker response in some people. Certain live vaccines may be unsafe in severe immunodeficiency. Close household contacts may also need vaccine advice.

Complications

Repeated infection can damage the lungs, sinuses, ears, skin, or other organs. Long-term airway damage called bronchiectasis may develop after repeated chest infections. It can cause a regular cough, mucus production, and further infections.

Poor absorption from the gut can affect weight, growth, and nutrition. Chronic inflammation may also cause tiredness or pain.

Some immunodeficiencies increase the chance of autoimmune conditions. Others are associated with a higher risk of particular blood cancers, especially lymphoma. The level of risk is not the same for every type.

Complications can often be reduced through early infection treatment, regular monitoring, and care tailored to the specific immune problem.

Prevention

Inherited immunodeficiency cannot usually be prevented. Genetic counselling can explain inheritance and reproductive options when a specific genetic change is known, but it cannot change the condition in someone who already has it.

Some secondary causes are preventable. Measures that reduce HIV transmission can prevent HIV-related immune damage. Good nutrition can help prevent immunodeficiency caused by severe undernutrition.

Immune-suppressing treatment is sometimes essential. Its risks are managed by using an appropriate treatment plan, monitoring for infection, and reviewing whether ongoing suppression is needed. Do not reduce or stop prescribed medicine on your own.

Everyday steps such as handwashing, dental care, food safety, and avoiding close contact with people who are actively unwell may lower exposure to infection. Extreme isolation is rarely practical and can harm wellbeing.

Living with it and what to expect

The long-term course is shaped by the exact immune defect, how early it is recognised, previous organ damage, and the response to treatment.

Some people have mild antibody problems and lead full lives with occasional treatment and monitoring. Others need regular immunoglobulin replacement, preventive medicines, or frequent specialist visits. Severe combined immune conditions may require transplant or gene therapy early in life.

Treatment may reduce infections without removing every symptom. For example, immunoglobulin replacement can protect against many bacterial infections, but it cannot correct structural lung damage that has already developed. Ongoing lung care may therefore remain important.

A personal infection plan can make day-to-day decisions clearer. It may cover which symptoms to report promptly, where to seek help outside normal hours, and whether samples should be collected before treatment.

Regular follow-up may include blood tests, lung assessment, growth and nutrition checks, or monitoring for autoimmune problems. Appointments may be more frequent after a new diagnosis or treatment change.

School, work, exercise, travel, relationships, and family life are possible for many people. Practical planning may include carrying a medical summary, arranging enough medicine for travel, and checking vaccine or infection advice before visiting certain destinations.

The emotional effect can also change over time. Repeated illness and uncertainty may cause worry or social isolation. Support from family, mental health services, or an immunodeficiency support organisation can be useful.

When to get medical help

Arrange routine medical advice if you or your child has repeated infections, infections that keep returning, persistent thrush, unexplained weight loss, poor growth, long-lasting diarrhoea, or a strong family history of severe infection.

If you already have an immunodeficiency, follow your agreed plan for fever and new infection symptoms. Seek prompt medical advice for a new fever, worsening cough, painful skin infection, persistent vomiting or diarrhoea, or symptoms that are not improving as expected.

Get urgent or emergency help for:

These symptoms can occur with sepsis or another serious problem. Use your local emergency

number rather than waiting for a routine appointment.

- Severe difficulty breathing, blue or grey lips, or struggling to speak
- New confusion, unusual drowsiness, collapse, or difficulty waking
- A rapidly spreading rash with fever
- Severe weakness with cold, mottled, or unusually pale skin
- Signs of severe dehydration, such as very little urine
- Symptoms of a serious allergic reaction during treatment

Common questions

Is immunodeficiency contagious?

No. An immunodeficiency itself cannot spread between people. However, the infections a person develops may be contagious. HIV can be transmitted through specific body fluids, but the immune damage caused by HIV is not spread through ordinary contact.

Does getting lots of colds mean I have immunodeficiency?

Usually not. Children and adults can have several routine viral infections, especially with frequent school, childcare, or workplace exposure. Medical assessment is more important when infections are unusually severe, persistent, difficult to treat, or caused by uncommon germs.

Can immunodeficiency be cured?

Sometimes. Stem cell transplant or gene therapy can correct selected inherited types. Treating an acquired cause may allow immune function to recover. Many other forms are managed long term rather than cured.

Can someone with immunodeficiency have vaccines?

Often, yes, but the safest plan depends on the type and severity of immune impairment. Some vaccines may work less well, and certain live vaccines can be unsafe in severe immunodeficiency. A specialist vaccination plan is important.

Does immunodeficiency always start in childhood?

No. Some severe inherited forms appear during infancy, but milder genetic conditions may not be recognised until adulthood. Secondary immunodeficiency can develop at any age because of illness, infection, or treatment.

Does immunodeficiency affect life expectancy?

It can, but the outlook differs greatly between specific types. Early diagnosis and treatment have improved outcomes for many people. Severe infections, organ damage, autoimmune complications, and some cancers can affect long-term health.

Sources

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