

MBL
deficiency

Mannose-binding lectin (MBL) deficiency

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Supporting families affected
by primary and secondary
immunodeficiency

About this booklet

This booklet provides information on mannan-binding lectin (MBL) deficiency. It has been produced by the Immunodeficiency UK Medical Advisory Panel and Patient Representative Panel to help answer the questions patients and their families may have about this condition but should not replace advice from a clinical immunologist.

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Summary

Mannose-binding lectin (MBL) deficiency is a condition that affects the immune system. It is a fairly common condition, affecting approximately 5-30 people in every 100. People with this condition have low levels of an immune system protein called mannan-binding lectin in their blood.

People with MBL deficiency may be prone to recurrent infections, including infections of the upper respiratory tract and other body systems. Sometimes those affected may also contract more serious infections, such as pneumonia and meningitis. The exact symptoms caused by infections vary in frequency and severity, depending on the type of infection.

Infants and young children with MBL deficiency seem to be more susceptible to infections, but adults can also develop recurrent infections. In addition, affected individuals undergoing chemotherapy or taking drugs that suppress the immune system are especially prone to infections.

The treatment of health problems associated with MBL deficiency depends on the severity of the symptoms, but most people do not require any specific treatment. Breakthrough infections are treated with antibiotics as and when they occur. For more severe infections, regular, low dose prophylactic antibiotics may be prescribed. The quality of life for those affected is good if infections are mild and/or treated promptly.

How did I get MBL deficiency?

Genetic changes (known as mutations) in the *MBL2* gene can lead to MBL deficiency. This gene provides instructions for making the MBL protein that plays an important role in the body's immune response. MBL protein attaches to foreign invaders such as bacteria, viruses or yeast and turns on (activates) another important part of the immune system known as the complement system.

Variants in the *MBL2* gene reduce the production of the MBL protein, so sometimes MBL deficiency may be referred to as MBL protein deficiency. The reduction of MBL in the blood means that the body does not recognise and fight foreign invaders (e.g. bacteria, viruses or yeast) efficiently. That is why infections can be more common in people with this condition.

Family planning and MBL deficiency

The inheritance pattern of MBL deficiency is not fully understood. People may inherit an increased risk of developing MBL deficiency but not develop symptoms of the condition. Furthermore, not everyone with a change in the *MBL2* gene has decreased levels of MBL, and not everyone with decreased protein levels is prone to infection. For these reasons it is more usual to measure the protein level in the blood than undertake genetic studies. Researchers believe that a number of factors, including other genetic and environmental factors, are involved in the development of MBL deficiency.

What are the symptoms of MBL deficiency?

The symptoms you may recognise and which may have led your doctor to a diagnosis of MBL deficiency are those associated with having recurrent respiratory infections. These usually affect the upper airways and include infections of the throat and ear; the latter causing otitis media.

People with MBL deficiency may sometimes also get more serious infections, such as pneumonia and meningitis.

What are the common causes of infection in MBL deficiency?

Common infections in MBL deficiency are due to viruses, e.g. influenza and bacteria, such as *Pseudomonas aeruginosa* and *Staphylococcus aureus*.

How is MBL deficiency diagnosed?

The diagnosis is made using a sample of the patient's blood. This is tested to see if the MBL protein is present in normal levels or is reduced. If it is reduced, then the patient's condition is classified as being mild, moderate or severe deficiency, with severe being complete deficiency when there is no detectable MBL in the blood.

Treatment

Treatment of MBL deficiency depends on the severity of the symptoms. In most cases no regular treatment of any kind is needed. If infections are mild and infrequent, and the person's quality of life is not significantly affected, then treatment can be limited to the early use of antibiotics when an infection occurs. For more information on the use of antibiotics, please read the Immunodeficiency UK booklet *Use of antibiotics in the treatment of primary immunodeficiencies* available on our website at www.immunodeficiencyuk.org.

If infections are more severe and/or are occurring frequently and time is being lost from work or school, or in the case of a child if growth and/or development are being affected, then regular, low dose prophylactic antibiotics may be introduced to prevent the development of infections.

If there are recurrent bacterial infections, appropriate antibiotics should be chosen according to the sensitivities of the organism isolated on culture.

Practising good hygiene plays an important role in keeping people with MBL deficiency well. See our advice page on keeping well and healthy when you have a primary immunodeficiency at www.immunodeficiencyuk.org.

Children and MBL deficiency

Infants and young children with MBL deficiency seem to be more susceptible to infections. This is because the lectin pathway plays an important role in fighting bacterial infections during the period when the antibody protection from the mother decreases and the child's own antibody production is not fully working.

Are there any other associated health problems with MBL deficiency and how will my/my child's health be monitored?

At present there is no conclusive evidence to support the association of MBL deficiency with the development of particular medical conditions.

Doctors will monitor patients' health by clinical review (check-up) and infrequent blood tests. Most patients will be looked after by their GP in the long term unless a clinical immunologist has identified any complications or additional immune problems.

Immunisation

Good protection against common bacteria and viruses, in keeping with the recommended vaccine schedule, will help reduce infections in patients with MBL deficiency. Most vaccines are safe to be given to patients with MBL deficiency. The annual flu vaccine is usually advised.

Immunologists may test vaccine responses to make sure that no other subtle immune defects, such as specific antibody deficiency, are contributing to MBL deficiency to cause frequent infection.

Glossary of terms

antibody – a type of protein (immunoglobulin) that is produced by certain types of white blood cells (plasma cells – a type of B-cell). The role of antibodies is to fight bacteria, viruses, toxins and other substances foreign to the body.

antibody deficiency – covers a range of disorders resulting from the failure of the immune system to produce sufficient antibodies in the bloodstream to fight infections.

chemotherapy – a type of cancer treatment that uses drugs to destroy cancer cells.

complement system – a group of immune system proteins that work together to destroy foreign invaders (pathogens), trigger inflammation and remove debris from cells and tissues.

gene – the fundamental unit of inheritance that carries the instructions for how the body grows and develops.

immune system – the structures and processes that protect the body against infection and disease.

inheritance – the passing down of genetic information from parents to children.

meningitis – an infection of the meninges (protective membranes) that surround the brain and spinal cord. It can be bacterial, viral, tuberculous or inflammatory (non-infectious).

mutation – a permanent alteration to a gene where part of the DNA within the gene is different from what it should be. There may be an extra or missing part, for example. Mutations may affect the proper growth or development of a person. Mutations can have either positive or negative effects on an individual.

otitis media – inflammation or infection of the ear.

protein – one of the basic building blocks of life. Proteins make up the structure and determine the function of the cells that make up all the tissues of our bodies.

respiratory tract – the airway passage involved in breathing that leads from the mouth/nose to the lungs.

variants – any alteration in a gene from its natural start; it may be disease causing or a benign, normal variant.

Notes

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About Immunodeficiency UK

Immunodeficiency UK is a national organisation supporting individuals and families affected by primary and secondary immunodeficiency.

We are the UK national member of IPOPI, an association of national patient organisations dedicated to improving awareness, access to early diagnosis and optimal treatments for PID patients worldwide.

Our website has useful information on a range of conditions and topics, and explains the work we do to ensure the voice of patients with primary and secondary immunodeficiency is heard. If we can be of any help, please email us or call on the number above, where you can leave a message.

Support us by becoming a member of Immunodeficiency UK. It's free and easy to do via our website. Members get monthly bulletins.

Immunodeficiency UK is reliant on voluntary donations. To make a donation, please go to **www.immunodeficiencyuk.org/donate**



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