

# Living with Primary Immunodeficiency

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# What is Primary Immunodeficiency (PID)?

Your doctor will have told you that you or your child has a condition called Primary Immunodeficiency (PID), also known as Primary Antibody Deficiency (PAD).

PID is a genetic condition where your immune system is missing some of the key parts that make it work properly. This means that it is difficult for your body to fight infection and you are probably ill very often. People with PID tend to catch one infection after another and some of them, such as ear, sinus and chest infections, don't seem to clear up with treatment as well as expected.



Besides being painful, frustrating and sometimes frightening, having constant infections can damage your body's tissues and, in more severe cases, can even be life threatening.

Some clinicians estimate that there may be as many as 100 different forms of PID and not all of them are severe. The few which are severe become apparent almost immediately after birth, and others may be diagnosed within the first year of life.

Other, milder forms may not show up until people reach their teenage years, or during early adulthood.

At the moment there is no cure for PID, but it can be treated so that infections can be avoided or minimised. Many people living with PID are able to lead very normal lives and get on with ordinary activities, just like everybody else.



# X-Linked Agammaglobulinaemia (XLA)

Children with XLA make virtually no antibodies – the blood proteins called immunoglobulins that fight infection – because they have either too few or none of the special white blood cells called B cells which make them.

It is called X-Linked because the mutated gene responsible for the condition is located on the X chromosome. This means it affects only males because they have just one X chromosome, whereas females have two and only one is likely to be faulty.

Baby boys who have inherited XLA appear healthy for the first few months because they are protected by the Immunoglobulin G (IgG) they received from their mothers via the placenta before birth and in the breast milk after they were born. However, as the protective effect of this IgG begins to fade, the baby can develop a steady stream of bacterial infections that may cause ear, sinus, eye and skin problems, as well as pneumonia. Boys with XLA are also susceptible to common viruses that can cause diarrhoea and weight loss.

Although XLA cannot be cured, it can be controlled with immunoglobulin therapy, which should prevent most infections and allow many children to lead normal, active lives.



# Common Variable Immunodeficiency (CVID)

CVID is the name given to a group of disorders characterised by low levels of different types of immunoglobulins and sometimes, by too few Immunoglobulin A (IgA) antibodies.

It can occur in children of either sex, but is more common in the second or third decades of life, and most patients have no family history of CVID.

People with CVID may have normal numbers of B cells, but they don't work properly. Their T cells may also have a variety of defects. These cells are another type of specialised white blood cell responsible for making powerful chemicals which help control our immune response.

Like most immunodeficiencies, CVID can cause frequent bacterial infections, typically involving the airways. Many patients experience repeated bouts of pneumonia and some may develop infections in their joints, bones and skin, as well as immune system illnesses including anaemia and rheumatoid arthritis.

Treatment with infection-fighting drugs called antibiotics can help to control infections, but the key to managing the condition is immunoglobulin therapy. This raises the antibody levels to help ward off infections so that many patients can enjoy an ordinary life.



# IgG Subclass Deficiency

This is caused by a lack of specific antibodies – usually one or two of the four subclasses of Immunoglobulin G, which are IgG1, IgG2, IgG3 or IgG4. Because overall IgG levels may be near normal, it is necessary to measure each of the subclasses to make a positive diagnosis.

As each subclass plays a slightly different role in fighting infection, each type of deficiency leaves the patient vulnerable to specific types of infection, particularly sinus and lung infections. This may be as severe as those seen with broader immunoglobulin deficiencies such as XLA or CVID.

Treatment normally comprises a course of antibiotics to control and prevent infections. Immunoglobulin therapy is usually reserved for children who are not seriously ill, and those who are not responding to antibiotics alone.



# Severe Combined Immunodeficiency (SCID)

SCID is one of the rarest forms of PID and is often the most severe. It is the result of a combined deficiency involving B and T cells. In some children the deficiency can be total, and almost any infection can be life-threatening for them.

SCID is three times more common in boys than girls because the X-linked deficiency is the most common form of SCID.

The symptoms which appear in the first few weeks of life are persistent thrush or extensive nappy rash. Weakened by chronic diarrhoea, the baby may fail to gain weight. Ensuing infections tend to be overwhelming and even viruses that are not usually harmful to humans can pose a serious threat to children with SCID.

Major and early intervention is necessary in order for children to survive infancy, and involves bringing any current infection under control using strong antibiotics. Immunoglobulin therapy may help to bolster the immune response, but the long-term solution is to arrange for a bone marrow transplant (BMT).



# Signs and symptoms

The most usual problem in PID is an increased susceptibility to infections which may be common, severe, lasting or hard to resolve.

Although healthy youngsters may get frequent coughs, colds and earaches (around 2-3 ear infections a year), children with PID can get one infection after another, or two to three different infections at one time. Weakened by infection they may fall behind in growth and development. Despite taking antibiotics, the infections of PID often drag on or keep coming back, becoming chronic.

Common problems include:

- Chronic sinusitis (infection and inflammation of the air passages in the bones of the cheeks and forehead)
- Chronic bronchitis (infection and inflammation of the airways leading to the lungs)
- Pneumonia (infection in the deep parts of the lungs)



Besides all the infections, some immunodeficiency diseases produce other immune system problems, including autoimmune disorders where the immune system gets out of control and mistakenly attacks the body's own tissues. This causes, for example, anaemia (a lack of red blood cells), or diabetes (a disorder caused by insufficient amounts of insulin).

Organs can be targeted too, producing diseases such as rheumatoid arthritis or systemic lupus erythematosus. Rheumatoid arthritis affects the joints, nerves, lungs and skin. Systemic lupus erythematosus affects skin, muscles, joints, kidneys and other organs, causing rashes, joint pain, fatigue and fever.



# Diagnosis

Occasionally the signs and symptoms of PID are so characteristic that the diagnosis is obvious. However, in most cases it is not clear if a long succession of illnesses is simply 'ordinary' infections or the result of an immunodeficiency.

Experts estimate that about half of the children who see a doctor for frequent infections are normal, 30% may have allergies and 10% have other serious disorders. Just 10% turn out to have an immunodeficiency.<sup>1</sup>

If PID is suspected, the doctor will want a full patient history with a list of past infections, details about how long they lasted and how frequent they were, as well as how well they responded to antibiotic treatment. The doctor will also explore the family history to see if any relatives have been diagnosed with PID or shown any unusual susceptibility to infections.

A medical examination will also be required and the doctor will assess growth rate and whether the patient is the correct weight for their height. Changes in the lungs will also be looked for, as well as the presence of rashes or sores and enlarged liver and swollen joints. All this will be supported by simple blood screening.

In fact, it takes just two, routine tests – total blood counts and quantitative immunoglobulin count – to detect most, but not all, immunodeficiencies.

#### Reference:

1. [www.nichd.nih.gov/publications/pubs/primary\\_immuno.cfm#SignsandSymptoms](http://www.nichd.nih.gov/publications/pubs/primary_immuno.cfm#SignsandSymptoms)



## THE 10 WARNING SIGNS FOR DIAGNOSIS OF PID<sup>1</sup>

1. Eight or more new ear infections within a year
2. Two or more serious sinus infections within a year
3. Two or more months on antibiotics with little or no affect
4. Two or more pneumonias within a year
5. Failure of an infant to gain weight or grow normally
6. Recurrent deep skin or organ abscesses
7. Persistent thrush in mouth or on skin, after age one
8. Need for intravenous antibiotics to clear infections
9. Two or more deep-seated infections such as meningitis, osteomyelitis, cellulitis or sepsis
10. A family history of primary immune deficiency

## Treatment

Treating PID involves not only curing infections but also correcting the underlying immunodeficiency. Any associated conditions, such as autoimmune disorders, will also need special attention.

The treatment of PID is discussed in detail in one of the other booklets in this pack.

A Patient Information Service from:



For full prescribing information please contact:  
Bio Products Laboratory, Dagger Lane, Elstree,  
Hertfordshire, WD6 3BX, United Kingdom

Tel: +44 (0)20 8258 2622  
Fax: +44 (0)20 8258 2611  
Email: [info@bpl.co.uk](mailto:info@bpl.co.uk)

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