



PRIMARY IMMUNODEFICIENCIES

PRIMARY IMMUNODEFICIENCIES AND INFECTIONS



KEY ABBREVIATIONS

| | |
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| CGD | Chronic Granulomatous Disease |
| CVID | Common Variable Immunodeficiency |
| WAS | Wiskott-Aldrich Syndrome |
| IgE | Immunoglobulin E |
| HSCT | Haematopoietic stem cell transplantation |
| IG | Immunoglobulin |
| IPOPI | International Patient Organisation for Primary Immunodeficiencies |
| PID | Primary immunodeficiency |

Primary immunodeficiencies and infections (1st edition)

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INTRODUCTION: WHAT IS INFECTION?

This booklet explains the range of infections that can affect people with primary immunodeficiencies and how they can best be prevented and treated.

Infection occurs when the body is invaded by microorganisms (such as bacteria, fungi, viruses and parasites) that are not normally present. Infections can affect a particular part of the body (such as the throat or lungs) or they can spread to affect the whole body.

Infections are communicable diseases, meaning that they can be spread by air, contaminated food, water, blood or direct contact.

Common infections include:

- Respiratory tract infections, including pneumonia and infections of the sinuses, ear passages and throat
- Skin infections, such as dermatitis or abscesses (infected areas containing pus)
- Bowel infections, such as gastroenteritis and *Clostridium difficile* infection
- Infections of the central nervous system, including the brain – such as meningitis and encephalitis
- Bloodstream infection and sepsis.

Primary immunodeficiencies (PIDs) are rare diseases that occur when components of the immune system of the body are either not present or are not working normally. The immune system is a complex system that protects the body from infections. By compromising the immune system, PIDs leave people more vulnerable to infections.

The following sections explain in more detail about why infections are particularly problematic for people with PIDs, how infections are investigated and treated, and the important steps that patients, parents and carers can take to prevent them.

SPECIFIC ISSUES FOR PATIENTS WITH PIDs

There are various reasons why infections are particularly problematic for people with PIDs and why patients should try to avoid them:

COMMON AND SEVERE INFECTIONS

Patients with PIDs can catch the same infections that affect other people. However, these infections tend to be more frequent and more severe in people with PIDs than in other people. Recurrent infections (such as pneumonia, diarrhoea and infections of the inner ear and the sinuses) are often the first signs that someone has a PID. Infections can also be more difficult to treat in people with PIDs, and therefore they may last longer and become 'chronic' infections.

Different PIDs affect different parts of the immune system (as explained in the IPOPI leaflet '*Classification of PIDs*'). A patient's susceptibility to infections therefore depends on which type of PID they have. For example:

- Respiratory tract infections are commonly associated with antibody deficiencies (e.g. common variable immunodeficiency; CVID) or complement deficiencies. Lower respiratory tract infections (e.g. pneumonia) are also common with chronic granulomatous disease (CGD), T cell deficiencies and CD40L deficiencies.
- Skin infections are often associated with many PIDs, especially phagocyte cell PIDs (e.g. CGD) combined immunodeficiencies (e.g. severe combined immunodeficiency, Wiskott-Aldrich syndrome (WAS), CD40L deficiency), CVID and hyper IgE syndrome.

'OPPORTUNISTIC' INFECTIONS

In addition, people with PIDs are more susceptible to certain infections that usually only affect people with weakened immunity – these are known as 'opportunistic' infections. Opportunistic infections can be caused by bacteria or viruses that would usually not cause infection in people with a normal immune system, or by more unusual organisms such as fungi or parasites.

DIFFERENT SYMPTOMS

Infections can cause many different symptoms, depending on which part of the body is affected, the type of infecting organism, and the severity of the infection. Common, general symptoms include fever (high temperature), pain and redness at the site (e.g. the skin or throat).

Importantly, patients with PIDs may not have the same symptoms that other people typically have. For example, PID patients may not have a fever, or they may show only mild symptoms even though their infection is severe or even life-threatening. This means that patients, parents and carers need to be very wary about infections, and must seek medical care if they think the patient might have one.

INVESTIGATING AND TREATING INFECTIONS

GETTING MEDICAL CARE

Patients, parents and carers must promptly contact their doctor if an infection is suspected, to allow prompt diagnosis and treatment. Some infections can be treated at home, but serious infections may need hospital care. In some cases, a consultation with an infectious diseases doctor may be needed.

Patients should always keep the details of their PID, medication, PID doctor and next of kin at hand, in case urgent medical care is needed. It may help to keep this information all together in a medical card or using the PID Genius app.

School staff should be aware of the needs of a student with a PID and the action to take if an infection is suspected, including alerting the patient's family.



PID GENIUS APP

IPOPI has developed the PID Genius app to help people living with PIDs organise their information and to make it available at any time, including in case of emergencies. PID Genius is available for download from [Google Play](#) and the [App Store](#).

INVESTIGATIONS

Various examinations and tests may be necessary to investigate a suspected infection. As well as a physical examination, patients may also need blood tests and scans (e.g. X-rays).

Often a sample (such as sputum, pus or blood) will be taken to help doctors identify the infecting organism and choose the best antimicrobial treatment. Identifying the organism is important because treatment can be lengthy or complex in PID and so doctors need to be sure they are giving the correct medications.

ANTIMICROBIALS

Antimicrobials (or anti-infectives) kill or suppress infecting micro-organisms and are used to treat and prevent infections. Antimicrobials include antibiotics (which work against bacteria), antivirals (which work against viruses) and antifungals (which work against fungi).

Each antimicrobial works against a certain range, or 'spectrum', of micro-organisms. This is why it helps, when choosing an antimicrobial, to know which organism is causing the infection. If doctors cannot identify the organism (or while they await test results) they may prescribe 'broad spectrum' antimicrobials covering the range of germs likely to be causing the infection.

People with PIDs may need higher doses or longer courses of antimicrobials than other people, because their infections can be harder to treat. Sometimes more than one antimicrobial may be necessary.

Antimicrobials may be given orally (as tablets, capsules or liquids), topically (e.g. as creams or drops for the eyes or ears) or by injection for more serious infections. Less commonly, antibiotics may also be inhaled (or 'nebulised') for certain respiratory tract infections.

IMPORTANT POINTS ABOUT TAKING ANTIMICROBIALS

Take antimicrobials exactly according to the label or patient information sheet, or the instructions from your doctor or pharmacist.

Only take antimicrobials prescribed for you – never "borrow" them from a friend or family member.

Tell your healthcare team if: your symptoms continue to get worse, you are unable to take the prescribed antimicrobials, or you experience unacceptable side effects.



SUPPORTIVE CARE

Several other types of treatment might be used to help relieve the symptoms of an infection, including:

- Fluids to avoid dehydration, e.g. especially in diarrhea.
- Medicines to reduce fever and pain.
- ‘Bronchodilator’ medicines to help breathing in patients with bronchitis (inflammation of the bronchial tubes of the lungs).
- Cough suppressants and expectorants to water down mucus in respiratory tract infections.
- Deep breathing exercises and respiratory interventions such as chest or sinus drainage, chest physiotherapy or positive airway pressure.
- Surgical drainage may be needed for some types of infected abscesses.

PREVENTING INFECTIONS

There are many things that patients with PIDs, parents and carers can do to help prevent infections, while maintaining as normal a life as possible.

HYGIENE AND PERSONAL CARE

Good hygiene is very important. This means simple things, such as:

- Washing the hands regularly and carefully, especially before meals and after using the toilet, outdoor activities and playing with pets
- Cleaning and dressing cuts and scrapes
- Catching coughs and sneezes in tissues – this is important both for patients and family members
- Careful brushing of the teeth and regular visits to the dentist
- Good food hygiene – a good nutritious diet is important, but patients should avoid raw or undercooked dishes (e.g. meats, eggs, cheeses and raw fish), contaminated water, or water of unknown origin or stored for long period in same container
- Some patients may be advised to avoid swimming in lakes or ponds and to minimise contact with dirt, dust or soil and contact with pets and other animals.

Where possible, patients should avoid contact with people with infections. For example, patients should avoid crowded places and consider wearing a face-mask during the winter influenza season. Parents of children with severe PIDs should ask the school to inform them of any infection outbreaks and then seek advice from the PID healthcare team. Parents should also ensure that the school enforces infection control measures, such as making sure that children with diarrhoea and vomiting remain off school for 48 hours after the episode.

Other important steps that patients and families can take to prevent infections include:

- Stopping smoking, to reduce the risk of respiratory infections
- Seeking a doctor's advice before getting any piercings, tattoos or similar procedure
- Seeking treatment for eczema (see PIDs and Allergies booklet), as scratching itchy skin can cause skin infections
- Practising safe sex (including the use of condoms where appropriate) to avoid sexually transmitted diseases
- Making sure, if surgery is needed, that the surgeon knows that a person has a PID.



VACCINATION

Vaccination (or immunisation) is the administration of a vaccine containing components of bacteria or viruses to protect against infections by these organisms. Children are routinely vaccinated against various preventable viral and bacterial infections, and other vaccines may be advised for travellers.

The appropriate use of vaccines for PID patients and their families depends on the specific PID and so it is very important to get individualised advice from the PID doctor. In general, vaccines that may benefit people with PIDs should be given and not avoided. However, most patients with PIDs should not receive certain vaccines called 'live' or 'live-attenuated' vaccines (including measles, mumps and rubella and oral polio vaccine) as these may cause patients to have infections.

Vaccination of family members and close contacts is highly recommended (e.g. influenza) to reduce the risk of infection to PID patients, but this should always be based on the PID doctor's advice.



TRAVEL

Having a PID should not prevent patients from travelling, but certain precautions are advisable. Patients should discuss travel plans with their PID doctor, who can advise on infection prevention issues, including the need for vaccines and malaria prevention. Some patients with severe PID may be advised to avoid countries where there is a high risk of infections.

While travelling, patients should take extra care with food hygiene and should avoid local water (including ice cubes) where this may be unsafe. Patients should also take precautions to avoid sunburn and insect bites, and should report any possible symptoms of infection (e.g. diarrhoea). Patients may be advised to take some treatment for traveller's diarrhoea with them.

Caution is required if a yellow fever vaccination certificate is required for travel, as this is a live vaccine. In this case patients/parents should ask for a letter from the immunologist to explain and may need to seek an exemption certificate from a specialist travel clinic.

IMMUNOGLOBULIN REPLACEMENT THERAPY

Immunoglobulin replacement therapy provides protection against infections. It is the most important therapy for many PIDs, although it does not remove the need for the other infection precautions above.

Immunoglobulin replacement treatment is prescribed according to each patient's individual circumstances. It can be given by intravenous or subcutaneous injection, and some patients can self-treat at home. More information can be found in the IPOPI leaflets, '*When to give IG replacement therapy*' and '*Immunoglobulin replacement therapy – One size does not fit all*'.

ANTIMICROBIALS

Patients with PIDs are sometimes prescribed antimicrobials to protect against infections, sometimes for long periods. This is known as 'prophylaxis'. Doctors consider the benefit/risk balance on an individual patient basis because of possible side effects and the risk of antimicrobial resistance (when microorganisms become resistant to the effects of antimicrobial drugs).

TREATING OTHER CAUSES

In some cases, some non-infectious medical conditions may contribute to a recurrent infection. For example, respiratory tract infections can result from allergic rhinitis and asthma, and eczema can lead to skin infections – hence it is important that these conditions are identified and treated (see '*PIDs and allergies*' booklet).

FURTHER INFORMATION AND SUPPORT

This booklet has been produced by the International Patient Organisation for Primary Immunodeficiencies (IPOPI). Other booklets are available in this series. For further information and details of PID patient organisations in 63 countries worldwide, please visit www.ipopi.org.



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