

Understanding primary immunodeficiency

A patient's advice sheet

What is immunodeficiency?

The immune system helps to protect us from infection - when this system fails we call the condition an immunodeficiency.

Primary immunodeficiencies are very rare diseases and some are so unusual that they only affect one or two people in the world. Any immunodeficiency means that you are likely to catch rather more infections than is normal. This is probably why you have been referred to a Consultant Immunologist.

Some immunodeficiencies are so severe they are diagnosed soon after birth. But others leave you at risk to just one or two types of bacteria, and it could be many years before you are exposed to these bacteria. In addition, because these diseases are so rare, many doctors do not take them into consideration when diagnosing possible causes of recurrent, severe, or unusual infections. As a result, it is possible to suffer long term ill health and even permanent damage, before you are diagnosed.

These diseases cannot be cured, but with proper care, most people can enjoy a reasonable quality of life.

What causes primary immunodeficiencies?

The cause is frequently found to be a faulty gene. Some immunodeficiencies are caused by a single faulty gene and others by a combination of genes. The fault may be inherited, be a new mutation in one of your genes or it may have been inherited from one or both of your parents. Most commonly, a faulty gene does not matter, unless the same fault is inherited from both parents. This is

most common when cousins have children together. Information about gene defects that cause primary immunodeficiencies is increasing rapidly due to new research. Your hospital doctor will be happy to discuss how this new information could affect your diagnosis and treatment.

Gene therapy

Where a single faulty gene is found to cause the immunodeficiency, gene therapy, which means a normal gene is transferred to a patient to replace a faulty gene, is the best treatment of all. The new gene can correct the immune deficiency and cure the disease. This therapy is available for a few defined immunodeficiencies. Bone marrow transplantation is also an option in certain cases where the immunodeficiency is very severe.

There is no link between Primary Antibody Deficiency and the Acquired Immunodeficiency Syndrome (AIDS). Patients with primary antibody deficiency diseases are not infectious and once treatment is started, they should start to feel better and suffer fewer infections.

There are a whole range of different primary immunodeficiencies. **Antibody deficiencies** are the largest group of primary immunodeficiencies. Antibodies are a group of proteins also known as immunoglobulins. This is the main group of proteins which help kill bacteria. There are three types of antibodies called IgG, IgA and IgM which work in different ways to protect us.

Antibody deficiency can range from a complete inability to make antibodies of any type, to

more subtle illnesses, where one type of immunoglobulin is missing. Sometimes it may be that normal levels of antibodies are present, but they are not able to attack certain groups of bacteria.

What treatment is available?

Patients who lack a number of their own antibodies to fight infections can be given replacement therapy via an infusion of antibodies. This is called immunoglobulin replacement therapy. The immunoglobulins are prepared from donated blood.

Immunoglobulin therapy

Immunoglobulin replacement therapy can be given in the Day Case Unit. If it is given in hospital, it is most frequently given straight into your blood stream via an intravenous drip of immunoglobulin (IVIg). It takes a few hours and should not be painful. Your specialist centre can provide training so that you are able to carry out treatments in your own home, with someone to help you. This regime should keep most infections at bay and dramatically reducing the time spent off work due to sickness. Although the treatment cannot reverse damage already caused by severe recurrent infections, it will prevent more problems developing. Papworth Hospital has a recognised home therapy training centre.

Immunoglobulin can also be given subcutaneously (SCIg) (just under the skin). This makes it simpler to administer yourself at home. However, this process needs to be repeated more frequently than IVIg, as smaller volumes are administered each time. It is useful for young children, patients with 'difficult' veins and for patients who want the freedom of deciding when and where to have their infusions.

Your immunology team will discuss with you the best way for you to receive your treatment, so that you can continue to live your life with as little disruption as possible.

The objective of immunoglobulin replacement therapy is to keep your immunoglobulin levels in the normal range for your age and protect you from infections.

Side effects from either IVIg or SCIg are rare and are most likely to happen during the first few infusions.

Other medicines which may be prescribed to you

Even on adequate treatment there may be 'breakthrough' infections. When these occur you must begin antibiotic therapy immediately. As soon as you begin to feel ill you must contact your GP to obtain a prescription for antibiotics. The surgery staff should be made aware of your condition and ensure that your request is given priority.

Safety of immunoglobulin

Immunoglobulin is prepared from human blood. There is potentially a risk of infection from blood borne viruses such as Hepatitis and HIV (the AIDS virus). However, there have been **no** cases of HIV or Hepatitis B being transmitted in this way. All blood donations are screened for Hepatitis B & C, HIV, and new variant CJD and the purification stages reduce the possibility of infection to a minute level.

There are several preparations available and your Consultant Immunologist will decide which one is best for you. Changing preparations is not advised under normal circumstances.

Specialist care

Ideally patients with primary immunodeficiencies should be looked after at a centre specialising in this condition. The Immunology Department at Papworth Hospital is a specialist centre for diagnosis and management of primary and secondary immunodeficiency and it is very important that you are seen regularly by its team of experts.

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Physiotherapy

Many people with primary immunodeficiencies are not diagnosed until they have suffered many chest infections, which may have damaged their lungs. If this is the case, you may be referred to a physiotherapist who will teach you exercises to help your breathing.

Alternative therapies

No 'alternative' therapy can affect your ability to make effective antibodies. However, in theory, something which helps you to relax and feel good in yourself is unlikely to harm you. But please, consult the Immunology team before embarking on any new treatment. And remember, it is vital that you always receive regular immunoglobulin replacement therapy.

Who do I need to tell?

Your GP will be kept informed of your progress and treatment. However, because it is rare, your doctor may not be fully familiar with your condition. Information and advice is always readily available from the team at Papworth Hospital when you come to your clinic appointments and through the Lung Defence Telephone Support Service.

If you are referred for any surgery, you must inform the surgeon of your condition, and tell the Immunology team what is happening. This is because you are more susceptible to infections than an average patient and your antibiotic cover and/or your immunoglobulin therapy may need to be increased to cope with surgery.

It is also a good idea to tell your dentist of your condition and, if you have an accident, the Accident and Emergency Department.

Immunisations

Regular replacement immunoglobulin therapy will keep you supplied with antibodies against most diseases. This replaces the need for vaccinations and you should **not** receive any 'live' vaccines. Although such vaccines contain

an organism that has been treated to make it harmless to people with a normal immune system, they could adversely affect you.

Your lifestyle

- It is important that you look after yourself with a sensible diet and exercise. Swimming, cycling and walking will all improve your general state of health. If you were diagnosed before major problems occurred, then you can look forward to a normal life style and to a normal life span.
- As chest infections are a particular problem, it is **vital** that you do not smoke, and that you ask **others** not to smoke around you and to allow you to live in a **smoke free environment**.
- If you intend to have children, please discuss this with your Immunologist, to obtain the best possible advice and treatment.
- You are more at risk of getting food poisoning and so **good** kitchen hygiene is **vital**.
- Pets should not present a health problem, as long as normal care is taken.
- Once you are on regular immunoglobulin replacement therapy you should have fewer interruptions to your work due to illness.
- Young patients may go away to college with pre-arranged treatment support at a local Immunology Centre.

Travel

Holidays and business trips abroad can be undertaken and even far flung exotic destinations can be enjoyed. A travel information leaflet is available to provide information relevant to keeping you well during your visit. Please ask your Specialist Immunology Nurse for a copy of this.

On-going support and further information

For more information please ask your Specialist Immunology Nurse.

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