



PRIMARY IMMUNODEFICIENCIES

# MANAGEMENT AFTER DIAGNOSIS



## ABBREVIATIONS

ALT	Alanine transaminase
AST	Aspartate aminotransferase
BUN	Blood urea nitrogen
CBC	Complete blood count
CT	Computed tomography
GGT or gamma GT	Gamma-glutamyltransferase
IG	Immunoglobulin
IVIG	Intravenous IG
MRI	Magnetic resonance imaging
PID	Primary immunodeficiency
SCIG	Subcutaneous IG
UTI	Urinary tract infection

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## INTRODUCTION

**This booklet explains how routine medical tests enable your physician to manage your health if you have a primary immunodeficiency.**

Primary immunodeficiencies (PIDs) are rare diseases that occur when components of the immune system are either missing or not working properly. There are over 250 types of PID, which vary widely in their clinical signs and symptoms.

If you have been diagnosed with a PID, regular consultations with your physician and other healthcare specialists are likely to be an important part of managing your health. You will probably meet with your physician on a regular basis if your health is stable and more frequently if you have complications. Routine blood tests, urine tests and scans will usually be carried out and other tests may be recommended, depending on your specific PID and health status. It may be useful to see other specialists regularly too, e.g. respiratory specialists, to manage any related conditions and have more detailed tests.

### WHAT ARE THE TESTS FOR?

The results of routine medical tests are used to:

- track the progress of your PID
- decide which treatments are necessary, e.g. immunoglobulin (IG) replacement therapy, bone marrow transplantation, gene therapy or additional medication
- check the health of your major systems and organ functions, e.g. immune system, blood, kidneys, liver, lungs
- screen for any side-effects associated with treatments.

Your physician will be checking to see if your levels are within normal ranges and will use the results to help with any treatment decisions or to decide if additional tests are needed to give more detailed information.

## BLOOD TESTS

Blood tests are common medical tests and are included in routine check-ups for many conditions. The procedure is quick and easy and usually doesn't require any special preparation.

A small blood sample is normally taken from a vein in your arm using a needle and a test tube. In children, samples are usually taken from the back of the hand after it has been numbed with anaesthetic cream.

Blood samples are usually examined and tested in a laboratory rather than the physician's clinic so results may not be available immediately. The sample is used to check the levels of blood cells, as well as substances found in the liquid part of the blood (plasma).

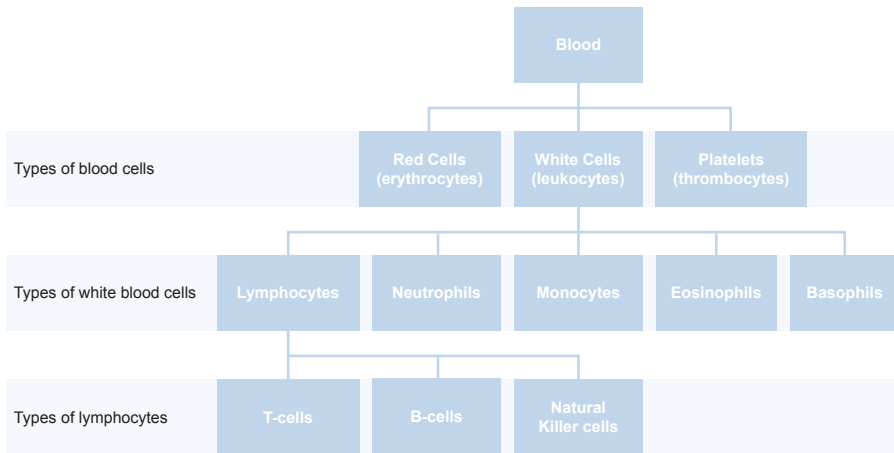
If you are nervous about blood tests, let your physician and nurse know. It can help to look away and to talk to someone to distract yourself.

## BLOOD COMPONENTS

The main components of blood are:

Red blood cells (erythrocytes)	Carry oxygen from the lungs around the body.
White blood cells (leukocytes)	Form part of the immune system and help defend the body against infection.
Platelets (thrombocytes)	Help blood to clot.
Plasma	The liquid part of blood is a mixture of water and chemicals, such as proteins, glucose and salt.

FIGURE: KEY COMPONENTS OF BLOOD



## BLOOD CELL TESTS

The test that your physician is most likely to request is called a haemogram or complete blood count (CBC), which provides information about the cells in your blood. Results will show the levels of the three main cell types: red blood cells, white blood cells and platelets. For many PID patients, the lymphocyte count is important as problems with this type of white blood cell can be the cause of the PID.

## IMMUNOGLOBULIN (ANTIBODY) TESTS

Your physician may also request tests to measure the levels of IGs (antibodies) in your blood serum. There are five types of IG: IgA, IgD, IgE, IgG and IgM. The results will enable him/her to decide if IG replacement therapy is necessary, if not already prescribed.

If you are already receiving IG replacement therapy, your physician may look at your trough level to check if you are receiving the correct dose. This is the lowest level of IG in your blood and it occurs just before your next infusion is due with intravenous IG (IVIG) replacement therapy. With subcutaneous IG (SCIG) therapy, it can be measured at any time between infusions.

## LIVER TESTS

Your blood sample can also be used to check if your liver is functioning correctly. When it is healthy, it releases various substances into the blood and these levels may be raised if you have a liver disorder or if you are taking antibiotics. The tests are known as liver function tests and usually include measuring:

- Alanine transaminase (ALT). When the liver is injured or inflamed, levels of this enzyme usually rise
- Aspartate aminotransferase (AST). Raised levels of this enzyme are seen if the liver, heart or other muscles are injured
- Gamma-glutamyltransferase (GGT or gamma GT). High levels are associated with high alcohol intake or liver problems
- Bilirubin. Raised levels indicate problems in liver or bile function.

## KIDNEY TESTS

To check on the health of your kidneys, your physician may order tests that check your levels of blood urea nitrogen (BUN) and creatinine:

- BUN is a waste product formed from the breakdown of proteins. High levels of BUN indicate that your kidneys may not be working properly (or that you are simply dehydrated)
- Creatinine. Also a waste product, this is produced by muscles. High levels may also indicate kidney function issues.

## OTHER BLOOD TESTS

The tests described above are the ones that are most routinely requested by physicians for their PID patients. A number of other tests can provide more detailed information and your physician may request these, if required.



TESTS	NORMAL RANGES (ADULTS)	WHAT DO THEY MEAN?
Red blood cells	Men: 5-6 million cells/ $\mu$ L Women: 4-5 million cells/ $\mu$ L	Red blood cells carry oxygen from your lungs to the rest of your body. Low red blood cell levels cause anaemia and may be due to bleeding, vitamin B12 deficiency, bone marrow problems or iron deficiency.
White blood cells	4,500-10,000 cells/ $\mu$ L	White blood cells form part of your immune system, which fights infections and diseases. Abnormal white blood cell levels might be a sign of infection or an immune system disorder.  A CBC measures your overall number of white blood cells. A complete white cell count or differential blood test can measure the amounts of different types of white blood cells.
Platelets	140,000-450,000 cells/ $\mu$ L	Platelets are blood cell fragments that help your blood clot by sticking together to repair cuts or breaks in blood vessel walls.  Low platelet levels can cause internal bleeding and may suggest a bleeding disorder (not enough clotting). High levels might indicate a thrombotic disorder (too much clotting).
IGs (antibodies)	IgG: 767-1590 mg/dL IgA: 61-356 mg/dL IgM: 37-286 mg/dL	In normal serum, there are five types of IGs, approximately 80% is IgG, with the rest comprising IgA (15%), IgM (5%), IgD (0.2%) and trace amounts of IgE



Liver function tests	ALT: 7-55 U/L AST: 8-48 U/L GGT or gamma GT: 9-48 U/L Bilirubin: 0.1-1.0 mg/dL	Liver tests measure levels of certain enzymes and proteins in your blood. Some measure how well your liver is performing its normal functions. Other tests measure enzymes that the liver cells release when damaged or diseased.
Kidney function tests	BUN: 7-20 mg/dL (2.5-7.1 mmol/L) Creatinine: 0.6-1.3mg/dL (53-115 $\mu$ mol/L)	Blood tests for kidney function measure levels of BUN and creatinine. Both of these are waste products that the kidneys filter out of the body. Abnormal BUN and creatinine levels might suggest a kidney disease or disorder.

**Note:** Normal ranges may vary depending on the reference range used by each laboratory and by age of the patient (normal ranges for children will be lower). Occasionally laboratories will present their results with different units. Ask your physician to explain your results.



## URINE TESTS

A urine test is another type of medical test often included in routine check-ups. Urine is created in the kidneys and is made up of water and waste products that need to be removed from the body. There are many different components and what you eat and drink, how much you exercise and how well your kidneys are working affect their levels.

A routine urine test can be carried out in your physician's office or clinic. You may be asked to collect a sample at home and bring it with you for testing or to collect a sample during your visit. The physician or nurse will usually use a test strip or dipstick to test your sample and results are available immediately. The strip changes colour in response to the sample and these colour changes tell your physician/nurse if there are any abnormal levels.

For patients with PIDs, physicians usually check for white blood cells or large amounts of protein (proteinuria). If there are abnormal results, a sample may be sent to a laboratory for tests that can provide more detailed information.

TESTS	NORMAL RANGES (ADULTS)	WHAT DO THEY MEAN?
White blood cells	Zero	Normally there are no blood cells in urine. Their presence can indicate a kidney or urinary tract infection (UTI)
Proteinuria	Albumin:creatinine ratio: >30 mg/mmol  Albumin concentration: >200 mg/L	Healthy adults usually excrete 80-150 mg/day of protein in urine. Proteinuria may be the first sign of renal disease

**Note:** Normal ranges may vary depending on the reference range used by each laboratory and by age of the patient (normal ranges for children will be lower). Occasionally laboratories will present their results with different units. Ask your physician to explain your results.

## SCANS

Your physician may suggest that you have a scan as part of your routine check-up. There are four main types of scans: ultrasound, X-rays, computed tomography (CT) scans and magnetic resonance imaging (MRI) scans. All are used to produce images of your internal organs, for example, your lungs, intestines and liver, which are difficult to explore from the outside. Patients with PIDs should ask for low-dose radiation for all scans that use X-rays.

Ultrasound scans	They use sound waves to produce images of internal tissues. The procedures are typically external (to examine organs in the abdomen), internal (vaginal or rectal examinations) or endoscopic (via the mouth for lung or stomach examinations).
X-rays	X-rays are commonly used to produce internal images, particularly of bones. They are also useful for highlighting lung infections, such as pneumonia, especially in children.
CT scans	Sometimes called CAT scans, they use X-rays and computers to rapidly produce more detailed images than normal X-rays. They are best suited for studying bones and internal organs and are particularly useful in children.
MRI scans	These use magnetic fields and radio waves to produce images. Although they take longer than CT scans, the radiation dose is much lower and they provide more detailed images of soft tissues.

Not all PID patients require scans on a routine basis. In some patients, they might be useful in picking up conditions at an early stage before they cause symptoms so that treatment can be started.

When you visit your physician, it is important to take your treatment diary with you, which should contain information you have collected about any infections, treatment side-effects, absences from school or work, infusion records, problems with your lungs, intestines, skin, etc. This will be helpful in guiding decisions about tests that may be required.

## 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 52 countries worldwide, please visit [www.ipopi.org](http://www.ipopi.org).

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