



Polycythaemia (raised red cell count)

Written by Dr Claire Harrison, Consultant Haematologist, St Thomas' Hospital, London and Professor Samuel J Machin, Department of Haematology, University College London Hospital

What is polycythaemia?

Doctors use the terms polycythaemia and erythrocytosis if you have too many red blood cells.

Red cells make up the majority of blood cells. They transport oxygen through the body, so their main content is the oxygen-carrying protein, haemoglobin.

Red cells have a special doughnut-like shape, which allows them to bend and squeeze through the smallest of blood vessels (see Figure 1).

The number of red cells you have varies according to age and sex. Men have higher levels than women and newborn babies often have more than adults.

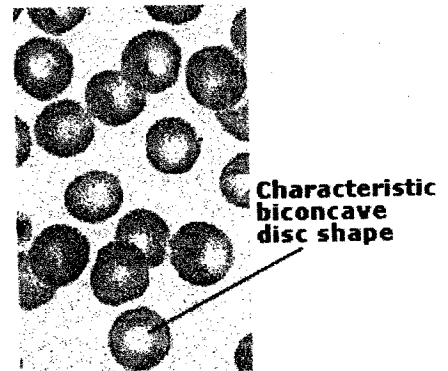


Figure 1: Normal red blood cells

Did you know?

If you have too few red cells, or a low haemoglobin, this is known as anaemia.

It is possible to be anaemic and have a relatively high number of red cells.

The main problem caused by polycythaemia is that the high number of red cells increases the blood's 'thickness' (viscosity). Blood flow to organs is reduced and, rarely, blood clots can form.

- If there is an actual increase in red blood cells, you have **absolute erythrocytosis**.
- If there is no increase in red cells, but they are more concentrated, this is known as **apparent erythrocytosis**.

What are the symptoms of a raised red cell count?

There are many symptoms that can be a feature of increased blood thickness but these are often rather vague. They include:

- a ruddy complexion
- headache
- blurred vision or patchy loss of vision
- confusion
- in extreme cases, stroke or coma.

The risk of clotting (thrombosis) increases if you have other risk factors for blood vessel disease such as previous clot, high blood pressure or diabetes.

Causes of a raised red cell count

Absolute erythrocytosis may be caused by an abnormality within the red blood cells (primary erythrocytosis) or by another condition (secondary erythrocytosis).

A raised red cell count most often develops from another disease. Usually, this is because a problem outside of the bone marrow stimulates the production of red blood cells. These diseases can be split into two main groups:

- those that cause low oxygen levels in the blood, for example from blockage to a kidney artery or lung disease.
- those that cause enhanced erythropoietin production, for example because of a tumour. Erythropoietin is the hormone that regulates the production of red blood cells.

Less often it occurs due to an abnormality of the red cells in the bone marrow itself, otherwise known as polycythaemia rubra vera (PV).

Genes and raised red cell count

It is rare for a person to inherit an erythrocytosis - either as a problem within the red cells themselves or one of the conditions that causes it.

Congenital red cell disorders are caused by a mutation in:

- the erythropoietin cell receptor that stimulates the synthesis of red cells
- the von Hippel Lindau protein that affects the production of erythropoietin
- proline dehydroxylase enzymes that also affect the quantity of erythropoietin.

Other inherited disorders include:

- a type of haemoglobin that binds more tightly to oxygen; a lower concentration of a compound called 2,3 diphosphoglycerate (2,3,DPG). In both, less oxygen is delivered to the tissues of the body. The body then compensates by increasing the total quantity of haemoglobin and thus red cells.
- a higher-than-normal level of erythropoietin.

Causes of absolute erythrocytosis

Primary (caused by an abnormality within the red blood cells)	Congenital (present at birth)	Abnormal erythropoietin cell receptor, von Hippel Lindau protein or proline dehydrogenase enzyme
	Acquired (develops some time after birth)	<u>Polycythaemia vera</u>
		Other myeloproliferative disease (essential thrombocythaemia)
Secondary (caused by an abnormality outside of the red blood cells)	Congenital	Inherited high erythropoietin levels Abnormal haemoglobin with the increased oxygen affinity Decreased level of metabolite (2,3 Diphosphoglycerate)
	Acquired (increased erythropoietin)	Conditions causing low oxygen levels: chronic lung disease, some types of congenital heart disease, sleep apnoea Kidney disease: tumours (cancers such as hypernephroma), cysts (usually benign), block to urine flow (hydronephrosis), after a kidney transplant
		Liver disease: hepatoma (liver cancer), cirrhosis, hepatitis Tumours: lung cancer, fibroids in the womb, abnormal blood vessels in the brain (cerebellar haemangioma) Endocrine (hormonal) abnormalities: Cushing's syndrome, tumour of the adrenal glands (phaeochromocytoma)
Idiopathic (unknown cause)		Idiopathic erythrocytosis

Apparent erythrocytosis

Apparent erythrocytosis, where red cells are more concentrated, can be caused by many things, such as:

- obesity
- fluid loss
- diuretics (water pills)
- smoking
- high blood pressure
- alcohol
- kidney disease
- stress.

The increase may be temporary and resolve once the cause is addressed, for example if a person loses weight, stops smoking or taking the diuretic.

Any increase in the volume of red blood cells should be monitored, because it may be the start of a true erythrocytosis.

How is a raised red cell count diagnosed?

A battery of different tools is used to decide if and why a patient has an erythrocytosis. An increase in red cells can first show in a blood test result as:

- an increase in red cell numbers
- a rise in haemoglobin
- a rise in the packed cell volume (haematocrit). This is a laboratory measure of the volume of red cells in the blood.

The initial blood test should be repeated to confirm the abnormal result. You will normally then be asked about your medical history, symptoms, medications and smoking habits.

A physical examination will also be performed to look for signs of possible underlying disorders.

Unless there is an obvious cause or the haematocrit is clearly very abnormal, a red cell mass study will be performed. This is often carried out in a hospital's nuclear medicine department.

The principle is to attach a very weak radioactive dye to the red blood cells and to a protein in the plasma (the liquid part of the blood). This involves mixing the dyes with a sample of your blood and then returning it to your body. The dyed cells distribute themselves among your red cells, making it possible to calculate what the total mass of red cells must be. This calculation can also be done using special radiosensitive cameras.

This red cell mass study determines if there is:

- absolute erythrocytosis (raised red cell mass, normal plasma volume)
- apparent erythrocytosis (normal red cell mass, reduced plasma volume).

Other diagnostic tests that may be performed include:

- blood tests of kidney and liver function
- measurement of iron, folic acid and vitamin B12, which are all important in red cell production
- measurement of oxygen levels in the blood - usually with a probe clipped onto your finger
- urine test for the presence of blood, sugar or other abnormality
- chest X-ray to check that the lungs and heart appear normal
- ultrasound of the abdomen to check the kidneys, liver and for any increase in the size of the spleen or fibroids in the womb.

Depending on the results of these tests, you may need further tests that are more specific to the likely underlying cause of your raised red cell count. These include:

- a bone marrow sample (performed under local anaesthetic)
- tests of lung function
- an echocardiogram to examine the structure of the heart

- genetic testing for mutations in JAK2 (common in polycythaemia vera), the erythropoietin receptor, von Hippel Lindau protein and proline dehydroxylase enzymes
- measurement of how tightly haemoglobin 'holds on to oxygen' (oxygen dissociation curve)
- a sleep study will be requested if a condition called sleep apnoea is suspected; this will involve an overnight stay in a special laboratory to see if prolonged breath-holding attacks occur when you sleep.

If an inherited condition is suspected, family members may be asked to have blood tests.

How is a raised red cell count treated?

In general, treatment is directed towards the underlying cause of the polycythaemia. If it's not possible to cure the underlying disease, your doctor will decide whether you need specific treatment to reduce your red cell count.

Term watch

Venesection: a hypodermic needle is inserted into a large vein to collect a set volume of blood.

Polycythaemia is treated by removing blood (venesection) at varying intervals until a satisfactory level of haematocrit (blood thickness) is obtained.

In patients with heart disease or a history of fainting, fluid is returned at the same time so that the circulating blood volume remains relatively constant.

If the cause of the raised red cell count is unknown (idiopathic erythrocytosis), you will require continual monitoring: It may turn out to have a medically significant cause which declares itself later, and requires treatment.

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