Polycythemia: Causes, symptoms, and management

In the clinical setting, most blood disorders involve a loss or reduction in the number of red blood cells (erythrocytes), excessive production of red blood cells presents its own problems also. These disorders that result from an overproduction of red blood cells are collectively known as polycythemia.

Polycythemias can be classified on the basis of either a total increase in the number of erythrocytes (absolute increase) or an increase in the concentration of blood cells secondary to dehydration.

An increased concentration of red blood cells due to dehydration is easily corrected by ?uid replacement and, therefore, we shall focus on absolute polycythemia.

Causes and pathophysiology of polycythemia

The most common underlying cause for absolute polycythemia is a physiological response to hypoxia, causing secretion of a hormone known as erythropoietin. An increase in erythropoietin hormone secretion leads to increased production of erythrocytes. This condition is referred to as secondary polycythemia.

In people with chronic obstructive pulmonary disorder (COPD) or congestive heart failure, or those who live in high altitude areas, the body is likely to respond in a similar manner and to develop this condition.

Individuals with abnormal hemoglobin can also develop secondary polycythemia. Some tumors can also predispose an individual to the development of an absolute increase in the number of red blood cells such as renal cell carcinoma, hepatoma, and cerebellar haemangioblastomas. This is because each of these tumors is associated with inappropriate EPO secretion.

Primary polycythemia, also known as polycythemia vera or polycythemia rubra vera. This is a rare condition marked by increased erythrocyte, white cell and platelet production, as well as splenomegaly.

This condition has an age of onset of 55–60 years of age, though it can occur at younger ages. Primary polycythemia is associated with changes in the bone marrow, with hyperplasia of the myeloid, erythroid and megakaryocyte precursor cells.

The majority of cases are associated with the JAK2 V617F mutation of the Janus kinase 2 gene. This gene encodes a tyrosine kinase that is involved in erythropoiesis.

Signs and symptoms

Increased viscosity in patients who have polycythaemias predisposes them to an increase in incidental thrombus formation, leading to occlusion of blood vessels of virtually all sizes, and

marked tissue and organ ischemia and, ultimately, infarction.

In association with the change in blood viscosity, blood ?ow becomes sluggish and individuals will manifest signs such as plethora and engorgement of retinal and cerebral vessels.

Symptoms of this condition include headache, drowsiness, delirium, changes to vision, chorea and behavior alterations, including delirium, mania, and psychotic depression.

Death due to cerebral thrombosis is more common in polycythemia, remarkably there are few cardiovascular disturbances and myocardial infarctions are relatively rare.

Patients may have extreme, painful itching skin that is exacerbated by heat or water.

Diagnosis

A full hemogram is performed to determine hematocrit and red blood cell count, as well as total blood volume. This is the mainstay of the diagnosis of polycythaemias.

Since polycythemia is an increase in circulating red blood cells, there is an increase in the amount of hemoglobin. This will have a negative e?ect on the veracity of pulse oximetry measurements.

If hemoglobin levels are increased and oxygen levels are normal, there is a low percentage of hemoglobin that is bound with oxygen. Therefore, the pulse oximeter will indicate lower oxygen saturation(hypoxia) although clinically, the person may be adequately oxygenated.

Something to note is that; before having confidence in the pulse oximeter measurement, determine that the person is not polycythaemic.

Management

The goal of treatment for low-risk individuals is to reduce erythrocyte production and the increase in blood volume using regular phlebotomy. This is initially performed two to three times per week and then every three to four months to maintain near-normal hematocrit levels.

In addition to this, a low-dose aspirin treatment can be used to reduce the incidence of incidental thrombus formation that can be triggered by routine phlebotomy.

Individuals who are at higher risk require intervention with cytotoxic agents or drugs.

Use of radioactive phosphorous (phosphorous-32) suppresses increased erythrocyte production and has lasting e?ects subsequent to a single exposure, with an e?ective period of 12–18 months.

Treatment is well-tolerated with few side-e?ects, although acute leukemia is a possible side-e?ect of treatment.

More commonly, hydroxyurea is a highly e?ective non-radioactive myelosuppressive agent that is used and is associated with lower risks of thrombus formation and leukemia.

Some people can be either tolerant or resistant to hydroxyurea and the use of interferon-alpha or

anagrelide, which suppresses platelet production, may be indicated, although interferon-alpha has a high degree of toxicity.

Despite a link to the presence of mast cells in the skin, antihistamines provide little relief.

The prompt and appropriate treatment provides a therapeutic remission and extends the life of the individual by about 10–15 years compared to those who do not receive appropriate treatment in the early stages of their illness who generally die within two years of symptom onset