

GLOSSARY OF MEDICAL TERMS

Ageusia – Loss of taste.

Agranulocytosis - An acute condition marked by severe decrease in white blood cells and by fever, exhaustion, chills, swollen neck, and sore throat sometimes with local ulceration; believed to be basically a response to the side effects of certain drugs.

Alopecia - Sudden loss of hair in defined patches with little or no inflammation.

Anaphylaxis – Extreme sensitivity to a foreign protein or drug; can be severe and sometimes fatal; causes a drop in blood pressure, difficulty breathing, fainting, itching, and hives.

Anemia – A condition caused by too few red blood cells in the bloodstream, resulting in insufficient oxygen to tissues and organs.

Anorexia – Prolonged loss of appetite and distaste for food.

Aplastic anemia - Anemia that is characterized by defective function of the blood-forming organs (as the bone marrow) and is caused by toxic agents or is idiopathic in origin. Also called *hypoplastic anemia*.

Ascites – The accumulation of fluid in the abdominal cavity, most commonly caused by cirrhosis of the liver.

Basal ganglia – Group of cells deep in the brain that initiate and control movement.

Bile – A yellowish green digestive fluid produced in the liver necessary for the digestion and absorption of fat.

Biochemical testing - Measuring the amount of a substance in the body through blood or urine analysis.

Ceruloplasmin - A blood glycoprotein to which copper is bound during transport and storage.

Chelator - binds the excess copper in the body and increases the excretion of copper in the urine.

Cholestasis – A reduction or stoppage of **bile** flow between the liver and the upper part of the small intestine.

Cirrhosis - A chronic progressive disease of the liver characterized by the replacement of healthy cells with scar tissue (**fibrosis**).

Cupriuria - The presence of excess copper in the urine.

Cutaneous macular atrophy – Spotted skin rash.

Diplopia - A disorder of vision in which two images of a single object are seen because of unequal action of the eye muscles (*double vision*).

Dysarthria - Difficulty in speaking words due to poor coordination of the speech muscles. Speech is slurred and there are uncontrolled fluctuations in volume.

Dysphagia: Slow movement of the tongue, lips, throat and jaws that causes drooling and difficulties in swallowing, caused by dystonia of the vocal chords. The voice may be hoarse, tone and volume may be diminished causing the speech to have a soft whisper-like quality

Dystonia - The condition of a sustained increase in muscle tone, sometimes with contractions or spasms of muscles of the shoulders, neck, and trunk. It frequently causes twisting and repetitive movements or abnormal postures, due to disease involving the basal ganglia of the brain.

Edema - The swelling of soft tissues as a result of excess water accumulation.

Elastosis perforans serpiginosum (EPS lesions) - Ring-shaped small, localized, superficial, solid elevations of skin, possibly occurring in groups. They may be discolored in varying hues of red, brown, or black. The outer layer of skin is thickened around a central plug of the skin's elastic tissue which is extruded through the outer layer of skin. EPS lesions have been identified as a possible side effect of long-term use of penicillamine.

Encephalopathy - A condition used to describe the harmful effects of liver failure on the central nervous system. Features include confusion ranging from confusion to unresponsiveness (coma). Symptoms generally related to the liver's inability to properly detoxify the blood and is associated with elevated blood ammonia levels. Treatment includes different methods to eliminate the production of ammonia from the gastrointestinal tract and to trap substrate for ammonia production so that it will not be absorbed by the gastrointestinal (GI) tract.

Endoscopy - A procedure using an endoscope , a small, flexible instrument that is a tube with a light and a lens on the end used to look into the esophagus, stomach, small and large intestine etc.

Esophageal varices - Stretched veins in the walls of the lower part of the esophagus and sometimes the upper part of the stomach and rarely in the small intestine and rectum. A complication of portal hypertension (increased blood pressure in the portal vein caused by [liver disease](#)). May cause massive bleeding.

Fatty liver (steatosis) – The build-up of fat in the liver cells.

Fibrosis – An abnormal thickening and scarring of the liver tissue.

Goodpasture's syndrome – A condition characterized by rapid destruction of the kidneys and hemorrhaging of the lungs. It is an autoimmune disease produced when the patient's immune

system attacks cells presenting the Goodpasture antigen, which are found in the kidney and lung, causing damage to these organs; caused by a possible toxicity of penicillamine treatment.

Hematemesis – Vomiting of blood; may be red, appear as coffee grounds, brown or black.

Hematuria – The presence of blood or blood cells in the urine.

Hemolysis - Breakdown of red blood cells, causing fewer than normal red cells to be available in the circulation to transport oxygen.

Hepatitis – Inflammation of the liver.

Hepatomegaly – Enlargement of the liver.

Hepatosplenomegaly – Enlargement of the liver and spleen.

Hyperkeratosis – Thickening of the outer layer of the skin.

Hypogeusia - Decreased sensitivity to taste.

Jaundice – Yellow or greenish hue to the skin and/or whites of the eyes caused by elevated bilirubin (formed when red blood cells are broken down; bilirubin taken up and transported by the liver into bile that is excreted into the intestine).

Kayser-Fleisher ring - A brown or greenish brown ring of copper deposits around the cornea; can only be seen with a slit-lamp by an ophthalmologist or optometrist early on, but may be visible to the naked eye when very large.

Leukopenia - An abnormal reduction in the number of white blood cells (leukocytes) circulating in the blood, most commonly caused by a reaction to various drugs.

Lichen planus - Shiny, flat-topped bumps that often have an angular shape. These bumps have a reddish-purplish color with a shiny cast due to a very fine scale. The disease can occur anywhere on the skin, but often favors the inside of the wrists and ankles, the lower legs, back, and neck.

Lupus erythematosus - A disorder characterized by skin inflammation, especially over the nose and cheeks – “butterfly rash”; or red scaly patches.

Lupus nephritis – Inflammation of the kidney associated with systemic **lupus erythematosus** that is typically characterized by **proteinuria** and **hematuria**, and that often leads to renal failure.

Lymphadenopathy - Abnormal enlargement of the lymph nodes.

Melena – Passing of dark blackish stools, indicating a bleeding disorder in the upper gastrointestinal track.

Myasthenic syndrome - Progressive weakness and exhaustibility of voluntary muscles without atrophy or sensory disturbance.

Nephrolithiasis - A condition marked by the presence of renal calculi (kidney stones).

Metallothionein Inducer - Removes copper from the body by increasing the amount of metallothionein in the cells of the intestines. Copper is bound within these cells and excreted through the stool.

Nephrotic syndrome - A collection of symptoms that affect the kidneys, resulting in a severe, prolonged loss of protein into the urine, decreased blood levels of protein (especially albumin), retention of excess salt and water in the body, and increased levels of fats (lipids) in the blood.

Obliterative bronchitis – Acute or chronic inflammation in the lung, causing closure of the bronchial tubes.

Optical axial neuritis – Inflammation of the nerve of the eye.

Portal hypertension – an increase in the pressure within the portal vein (the vein that carries blood from the digestive organs to the liver). The increase in pressure is caused by an increase in resistance to the blood flow through the liver due to swelling or scarring of the liver.

Proteinuria - The presence of excess protein in the urine.

Pseudobulbar palsy - A set of clinical signs including slowed slurred speech; difficulty with swallowing; weakness of face, tongue, and swallowing muscles; a tendency for uncontrollable laughter or crying; and brisk jaw and gag reflexes.

Psychosis – A serious mental disorder (as schizophrenia) characterized by defective or lost contact with reality, often with hallucinations or delusions.

Ptosis - A sagging or prolapse of an organ or part (renal *ptosis*); the drooping of the upper eyelid from paralysis of the third nerve.

Serous retinitis – Inflammation of the retina of the eye.

Serum free copper (Non-ceruloplasmin bound copper) – The amount of serum free copper is the amount of copper circulating in the blood which is not bound by ceruloplasmin. This is the copper which is "free" to accumulate in the liver and other organs. To calculate serum free copper, use the following formula:

(Total Serum Copper in $\mu\text{g/dL}$) - (Ceruloplasmin in $\text{mg/dL} \times 3$) = Free Copper
(Normal range is 5 to 15 $\mu\text{g/dL}$)

Sideroblastic anemia - Large numbers of iron-containing red blood cells in the bone marrow.

Splenomegaly – Enlargement of the spleen.

Thrombocytopenia - Persistent decrease in the number of blood platelets

Toxic hepatitis – Drug-induced inflammation of the liver.

Tremor - Involuntary, somewhat rhythmic movements of the muscles that cause various parts of the body to move uncontrollably.

µg – Microgram.

µMoles – Micromoles.

Varices – (*See Esophageal varices*)

