

Glossary of Blood, MPN, and Mutation Terms

Acute Lymphocytic Leukemia	ALL	Excess of lymphoblasts. Most common type of cancer in children with cure rate up to 80%. Cure rate for adults is up to 60%.
Acute Myeloid Leukemia	AML	Most common type of acute leukemia. Phases are measured by levels of abnormal immature white cells (blasts): Chronic Phase: < 10% blasts; Accelerated Phase: 10-19% blasts; Blast Phase: 20+% blasts; Blast Crisis: > 20% blasts with fever, fatigue and enlarged spleen.
Agnogenic Myeloid Metaplasia	AMM	Also known as Primary Myelofibrosis. Myeloid tissue in organs other than bone (i.e., liver, spleen). see Primary Myelofibrosis
Allele burden		Allele is the variant form of a given gene. Varied combinations from each chromosome produce alleles such as blood type, eye color, skin pigmentation. With MPN, allele burden (amount of mutation) varies by person and over time.
Anemia		When the number of red blood cells is below normal, or hemoglobin is insufficient to carry enough oxygen throughout the body. Can result in fatigue, weakness, and shortness of breath.
Anisocytosis		Refers to changes in the size of red cells; reflected in MCV
Antibody		A molecule created to adhere to and interact with the antigen that triggered its synthesis. The antigen-antibody reaction is important to an immune response.
Antigen		A substance on the surface of a cell that triggers an immune response; it also reacts with the product of the response (the antibody). Part of the body's immune response.
Auto-immune		The body's immune system identifies an organ or system as foreign and attacks it.
Autogenic, Autologous		One's own cells, tissues
Band Neutrophils		Immature neutrophil (leukocyte)
Basophil		A granular leukocyte (white cell)
Blasts, Bands		Immature cells
Blood		Red fluid, consisting of red cells, white cells, platelets, and plasma. It circulates throughout the body, carrying oxygen to tissues in the arteries and carrying carbon dioxide from the tissues in the veins. Considered a body organ in liquid form.
Blood Cancer		An uncontrolled growth of abnormal, or malignant, blood cells that affect the function and the production of blood cells in the body.
Blood clot (Coagulation)		Blood forms a clot when it comes in contact with a "thrombogenic" substance causing the blood to convert from a liquid to a solid state (also known as coagulation). Thrombogenic substances include collagen, tissue factor and von Willebrand factor.
Bone Marrow		The spongy, fatty, vascular tissue inside bones that hosts the hematopoietic stem cells which produce blood cells.
Bone Marrow Biopsy	BMB	A procedure used to remove soft tissue, called marrow, from inside the bone (typically the posterior pelvic bone). Blood and tissue is tested for disease or disease progression. Can be done under local anesthetic, or with "conscious sedation" to ensure a minimum of discomfort from the procedure.

Bone Marrow Transplant	BMT	A procedure in which healthy bone marrow stem cells are used to replace diseased or damaged bone marrow. Also called a Stem Cell Transplant. Autologous transplants use the patient's own cells. Allogeneic transplants use cells from a donor for a patient.
Chronic Myeloid / Myelogenous Leukemia	CML	The bone marrow produces excessive white blood cells, caused by the Philadelphia chromosome or the BCR-ABL fusion gene. Since granulocytes are involved in this type of leukemia, it is also referred to as CGL or chronic granulocytic leukemia.
Clonal Proliferation		Describes diseases arising from a single cell that rapidly reproduces genetically identical cells
Cluster Differentiation	CD	System of classifying lymphocytes according to antigen collections on their cell surfaces. Also called CD markers, e.g., rituximab destroys B cells that have CD20 antigen on their surface.
Coagulation		The process by which the blood converts from a liquid to a semisolid mass (blood clot), caused by a thrombogenic substance.
Colony-Stimulating Factors	CSF	A hormone produced in the lining of blood vessels that stimulates the production of bone marrow cells, which includes stem cells and differentiated blood cells
Complete Blood Count	CBC	A comprehensive blood test used to measure the number of white blood cells, red blood cells, platelets, the amount of hemoglobin, and the level of hematocrit. The CBC also gauges the size of reds and platelets. See Hct, Hg or Hgb, MCH, MCHC, MCV, PT/PL, RBC, RDW, WBC, WCC
Cytogenetics		Technique to identify and analyze the number and integrity of a cell's chromosomes
Cytogenic analysis / Karyotype		Examining samples of blood or bone marrow to study the chromosomes in order to identify genetic disorders.
Cytokines		A class of proteins used for communication between cells to trigger the immune system. They regulate the intensity and duration of immune response to protect from infections. Cytokines include interferons, interleukins, lymphokines, and chemokines.
Cytopenia		low blood cell count in circulation. Generally due to either low production or cell destruction.
Deep Vein Thrombosis	DVT	A blood clot that forms in a deep vein of the body, usually the thigh or leg. If and when the clot breaks off and moves through the bloodstream, it becomes an embolism, which can get lodged in the brain, heart or lungs, causing severe damage.
Diagnosed, Diagnosis	Dx	description or determination of a disease, injury or abnormality.
Embolism		A blood clot that breaks off and moves through the bloodstream, lodges in a blood vessel and blocks it. An embolism can become lodged in the brain, heart, lungs or other area, causing severe damage.
Enzyme		A protein that catalyzes (launches) a specific chemical reaction, without being used up or converted in the reaction. Name usually ends in <i>-ase</i> .
Eosinophils	EOS	White blood cells (granulocytes) with rough, round granules of cytoplasm that stain with eosin.
Erythrocytes		Red blood cells; carry oxygen from lungs to the tissues

Erythrocytosis		Overproductions of red cells
Erythromelalgia		Sudden dilation of peripheral blood vessels, often triggered by heat or exertion; causes throbbing, burning, or severe itchiness of the skin. Usually affects hands and feet.
Erythropoietin		A hormone controlling red cell production; it promotes RBC survival by protecting from apoptosis (programmed suicide cell death).
Essential Thrombocythemia	ET	A blood cancer, characterized by the overproduction of platelets (thrombocytes) in the bone marrow. Also referred to as primary thrombocytosis. Essential thrombocythemia is one of the myeloproliferative neoplasms (MPNs).
Gene mutation		A change in the DNA sequence. Gene mutations that are often associated with MPNs include JAK2V617F mutation, MPL mutation and calreticulin (CALR) mutation.
Gout		Acute arthritis (swelling) of joints, typically the big toe. Due to excess uric acid that isn't processed through the kidneys. Uric acid crystalizes and accumulate in the joints.
Graft vs Host Disease	GvHD	Complication of allogeneic SCT in which the new immune cells in the transplanted marrow treat the recipient's tissues (the patient's) as foreign and cause an immunologic attack.
Granulocyte		Any group of white blood cells (e.g., basophil, eosinophil, or neutrophil) with a granule-containing cytoplasm.
Hem/Onc		Hematologist & Oncologist. A physician who specializes in blood diseases and cancers. Many hematologists treat tumor cancers as well as blood cancers.
Hematocrit	HCT	Percentage of red blood cells in a volume of whole blood. the percentage by volume of whole blood that consists of blood cells (the remainder is plasma). Measured in a centrifuged test tube; also called PCV packed cell volume or EVF-erythrocyte volume fraction; reference range for females is approximately 33-43; for males 40-53; quoted as a decimal number by some laboratories (ie 0.40-0.53).
Hematologist, Hematology	Hem	Blood specialist doctor, study of blood
Hematopoiesis		Formation and development of blood cells.
Hematopoietic stem cell	HSC	Self-renewing, basic cell that can develop into any type of specialized blood cell.
Hematopoietic Stem Cell Transplant	SCT, HSC	Major procedure where patient's immune system (blood system in the bone marrow) is reduced through chemotherapy, then replaced with healthy blood stem cells from donor (allogenic SCT) or from self (autologous SCT). Healthy stem cells delivered through an IV. MPN patients must have an allogenic transplant because their stem cells are defective.
Hemoglobin	Hg or Hgb	Molecule in the red blood cell that carries oxygen throughout the body. Labs report the concentration in grams of hemoglobin in one liter of blood although some report it in 100 milliliters (one deciliter). Normal female 12-16 g/deciliter, male 14-18 g/deciliter (ie 120-160, and 140-180 g/l) (see RBC).
Idiopathic		The cause for a disease process is unknown; also called Agnogenic
JAK2 positive	JAK2+	The somatic genetic mutation found in approximately 50 percent of myelofibrosis patients, 95% of polycythemia vera patients, and approximately 50% of essential thrombocythemia patients.

JAK2v617F	JAK2	Janus Kinase 2 (JAK2) is a gene that exists in all people and signals /drives the growth and division of cells. In 2005 a mutation was discovered (known as JAK2V617F+) in the JAK2 gene in people with myeloproliferative neoplasms (MPNs). This mutation affects the proper signaling of the JAK2 molecule.
Janus Kinase-Signal Transducer & Activator of Transcription	JAK-STAT	A signal transduction pathway, ie the molecular-level messaging that occurs within cells; STATs are phosphorylated by JAKs, go into cell nuclei, bind to specific elements and induce gene transcription.
Karyotype		The chromosomes and their characteristics.
Kinase		Enzymes also called phosphotransferases; they transfer phosphate groups to proteins which then act as on-off switches in many biochemical systems.
Leukocytes		White blood cells; kill micro-organisms (infection) that invades the body
Leukocytosis		Overproduction of white cells
Lymphocyte		A type of white blood cell (leukocyte) that is responsible for the immune response and aids in defending the body against disease. There are two primary types of lymphocytes: B cells and T cells.
Lymphocytosis		Too many lymphocytes, immature granulocytes
Macrocytosis, Macrocytes		Red cells larger than normal (above 100)
Macrophage		A tissue cell of the immune system; it engulfs and consumes foreign antigens (viruses and bacteria) and debris (e.g., dead tissue cells). Created from a monocyte (type of white blood cell)
Monocyte	MONO	A large white blood cell that migrates into connective tissue; there it changes into a macrophage.
MPL gene	MPL	A mutation of the MPL gene is known to cause MPN.
Multiple Myeloma	MM	A blood cancer where tumors of the bone marrow formed from marrow cells (such as myelocytes, plasma cells); usually active in several bones at the same time.
Mutation		The change or alteration of something. Gene mutation changes the way a gene functions.
Myelo-		Of the bone marrow
Myelocyte		A bone marrow cell that occurs abnormally in the circulating blood.
Myelodysplastic Syndrome	MDS	Previously "preleukemia", or "smouldering leukemia"; a diverse collection of conditions involving ineffective production (dysplasia) of the myeloid class of blood cells; closely related to, but different from, MPNs.
Myelofibrosis with Myeloid Metaplasia	MFMM	Also called Secondary Myelofibrosis. Blood is produced in the spleen and liver to compensate for reduced bone marrow effectiveness due to fibrosis. This secondary form may be due to other diseases including polycythemia vera, carcinomatosis, tuberculosis, and leukemia.
Myeloproliferative Disorder	MPD	Myeloproliferative diseases; now called <i>Myeloproliferative Neoplasms (MPN)</i>
Myeloproliferative Neoplasms/Neoplasia	MPN	A group of diseases of the blood and bone marrow which causes overproduction of red blood cells, platelets, or certain white blood cells. There are three classic types of MPNs: myelofibrosis (MF), essential thrombocythemia (ET) and polycythemia vera (PV).

Neutrophilia		Too many neutrophils, granulocytes
Oncologist, Oncology	Onc	Cancer doctor, study of cancers
Petechiae		Flat, red, pinpoint spots under the skin caused by micro bleeds
Phagocyte		A cell that consumes and destroys foreign material (like virus or bacteria) and dead tissue cells. Created from white blood cells. Neutrophil and Macrophage are phagocytes.
Pheresis, Apheresis		A laboratory procedure, in which the blood is filtered, separated, and a specific component is retained, while the remainder is returned to the individual. (Plateletpheresis is the collection of platelets only).
Philadelphia Chromosome		An abnormality of chromosome 22, which is associated with chronic myeloid leukemia (CML), a myeloproliferative neoplasm.
Phlebotomy, Venesection	Phleb	Withdrawing blood from the body, usually in large amounts, for treatment purposes. Phlebotomy is a mainstay of treatment for the polycythemia vera (PV) to lower hemoglobin and hematocrit levels.
Plasmapheresis		A laboratory procedure in which the plasma is removed from the blood.
Platelet Count		The number of platelets in a given volume of blood. Either quoted as per liter (eg, reference range of 150-400 x 10 ⁹ per liter) or per microliter (reference range of 150,000-400,000).
Plateletpheresis		A laboratory procedure in which the platelets are removed from the blood.
Platelets (thrombocytes)	Pl	Small blood cell particles produced in bone marrow; repair damaged blood vessels by sticking together (clot) to prevent blood loss
Poikilocytosis		Change in the shape of the red cells
Polycythemia Vera	PV	Disease of the bone marrow where the stem cells produce excessive clonal red cells that rapidly multiply and are released into the bloodstream. Causes high red cell count (hematocrit) and Hemoglobin and increased blood volume and viscosity; can cause thrombosis, migraines, strokes. White cells and platelets may also increase. A Myeloproliferative Neoplasm
Polycythemia Rubra Vera	PRV	see Polycythemia Vera
Post-Polycythemia Vera Myelofibrosis with Myeloid Metaplasia		Also called "secondary myelofibrosis" because it occurs after polycythemia vera.
Primary		Refers to a disease that is initiated independently of any other diseases; compare to "secondary" disease which results from another disease or action.
Primary Myelofibrosis	PMF	A disorder of the bone marrow that disrupts normal production of blood cells. It causes excessive scarring in the bone marrow. Symptoms include enlarged spleen and anemia. Myelofibrosis is one of the myeloproliferative neoplasms (MPNs).
Proliferative		Takes part in rapid and repeated production of offspring (e.g., new cells).
Protein		A chain of amino acids
Red cell distribution width	RDW	Shows if the cells are all the same or different sizes or shapes. degree to which red cells vary in size from one another; normal 10.9-13.7

Red Cell Mass	RCM	Test for volume/mass of circulating red blood cells used to confirm diagnosis of PV; normal males 24-32 mL/kg, females 21-27. Rarely used now as JAK2 and EPO tests generally preferred.
Reynaud's Syndrome		A disorder in which the fingers or toes experience decreased blood circulation and the skin color intermittently changes from pale white to blue/purple, then to red.
Secondary Myelofibrosis	sMF	Myelofibrosis occurring as a progression from another of the MPNs, generally following PV, ET, or CML.
Secondary Polycythemia	SP	High hemoglobin and hematocrit. Can be caused by smoking, high altitude, chronic heart, lung or kidney disease; resolves when cause removed, or underlying disease is treated. Driven by an increase in the cytokine EPO, produced by the kidneys when oxygen levels are chronically low.
Thrombocytopenia		A lower than normal number of platelets in the blood.
Thrombocytosis or Thrombocythemia		Overproduction of platelets
Thrombopoietin	TPO	Hormone that regulates megakaryocyte (platelet precursors) production, and thus platelets; it operates through its receptor (Mpl) to stimulate production.
Tyrosine Kinase Inhibitors	TKI	A targeted cancer therapy that interferes with the signals that tell a cell to grow and divide. This therapy can reduce or stop cancer cells from growing, and, in some cases, it causes the cell to die.
von Willebrand Disease	vWD	Bleeding disorder caused by deficiency of VW Factor which helps platelets clump and stick to blood vessel walls; usually hereditary but Acquired VWD may occur in ET due to sequestration of Factor by adhesion to the high number of platelets.
White Blood Cells	WBC	Blood cells that fight infection and anything it perceives as foreign
White Blood Cells, Leukocytes	WBC	Blood cells that are part of the immune system and also help fight infection. Also called leukocytes.
White Cell Count	WCC	normal range $4-11 \times 10^9$ per liter (ie 4-11 billion cells per liter, sometimes quoted as 4000-11,000 per microliter). The precise range varies slightly between laboratories and regions
Wild Type	WT	refers to a normal cell or gene as it occurs in nature. Wild Type is in contrast to a mutation.
	BCR-ABL	Gene mutation: Philadelphia Chromosome translocation (see Ph) to form the oncogene which causes CML