

Terms in Blood & Genetic Tests

Term	Abbrev	Definition
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. Several causes of anemia.
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.
Band Neutrophils		Immature neutrophil (leukocyte)
Basophil		A granular leukocyte (white cell)
Bands, -blasts		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 Pressure	BP	The pressure blood exerts on the wall of blood vessels. BP is typically measured on the radial artery (on upper arm) as a fraction, such as 120/80. The numerator (120 in this example) is the maximum pressure that follows the heart's contraction (systole) to circulate blood through the body. The denominator (80 in this example) is the rhythmical expansion of the heart's cavities during which they fill with blood.
Bone Marrow		The spongy, fatty, vascular tissue inside bones that hosts the hematopoietic stem cells which produce blood cells.
Bone Marrow Aspiration		A technique for obtaining bone marrow tissue through a needle (usually in the pelvic bone) for examination.
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.
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.
Deoxyribonucleic acid	DNA	The cell nucleus material that contains genetic instructions.
Diagnosed, Diagnosis	Dx	description or determination of a disease, injury or abnormality.
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
Erythropoietin		A hormone controlling red cell production; it promotes RBC survival by protecting from apoptosis (programmed suicide cell death).
Fluorescence In Situ Hybridization	FISH	An analytical technique: a fluorescent probe that detects DNA sequences in situ (in normal position within a chromosome) and thereby detects abnormalities.
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.
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.
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).
International Normalized Ratio	INR	Test measuring time it takes blood to clot compare to an average; target range for warfarin dosing 2.0-3.0.
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
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.
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)
Mass Corpuscular Volume	MCV	Average size of the RBCs. average size of the red cells. MCV tends to be low if treated by phlebotomies, as iron deficiency, the goal of phlebotomies, causes red cells to be of smaller average size; normal range 80-100 fL (femtoliter, or 1x10 to minus 15th power of a liter).
Mean Corpuscular Hemoglobin	MCH	Amount of hemoglobin in the average RBC; reflects the average weight of hemoglobin in each cell; assumes homogeneous population of cells normal 27-33 pg (picograms)
Mean Corpuscular Hemoglobin Concentration	MCHC	The concentration of hemoglobin in an average RBC. It is a percentage relation between the hematocrit and hemoglobin values. % of average red cell which is filled with hemoglobin; normal 33.1-35.6.
Mean Platelet Volume	MPV	Measures the average amount (volume) of platelets. Used with platelet count to diagnose some diseases.
Monocyte	MONO	A large white blood cell that migrates into connective tissue; there it changes into a macrophage.
Myelo-		Of the bone marrow
Myelocyte		A bone marrow cell that occurs abnormally in the circulating blood.

Neutrophilia		Too many neutrophils, granulocytes
Oncologist, Oncology	Onc	Cancer doctor, study of cancers
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.
Platelet Count		The number of platelets in a given volume of blood. Either quoted as per liter (eg, reference range of $150-400 \times 10^9$ per liter) or per microliter (reference range of 150,000-400,000).
Platelets (thrombocytes)	PI	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
Post-Polycythemia Vera Myelofibrosis with Myeloid Metaplasia		Also called "secondary myelofibrosis" because it occurs after polycythemia vera.
Proliferative		Takes part in rapid and repeated production of offspring (e.g., new cells).
Protein		Complex molecule made up of a chain of amino acids. Proteins play several functions in the body's cells and organs. Proteins are antibodies, enzymes, messengers, transportation/storage, and support cell structure.
Prothrombin Time/International Ratio	PT/INR	Measures time for blood to clot after addition of tissue factor.
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.
Thrombocytopenia		A lower than normal number of platelets in the blood.
Thrombopoietin	TPO	Hormone that regulates megakaryocyte (platelet precursors) production, and thus platelets; it operates through its receptor (Mpl) to stimulate production.
Total Iron Binding Capacity	TIBC	Test of blood's capacity to bind iron to transferrin; normal 240-450 micrograms/deciliter.
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.
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.