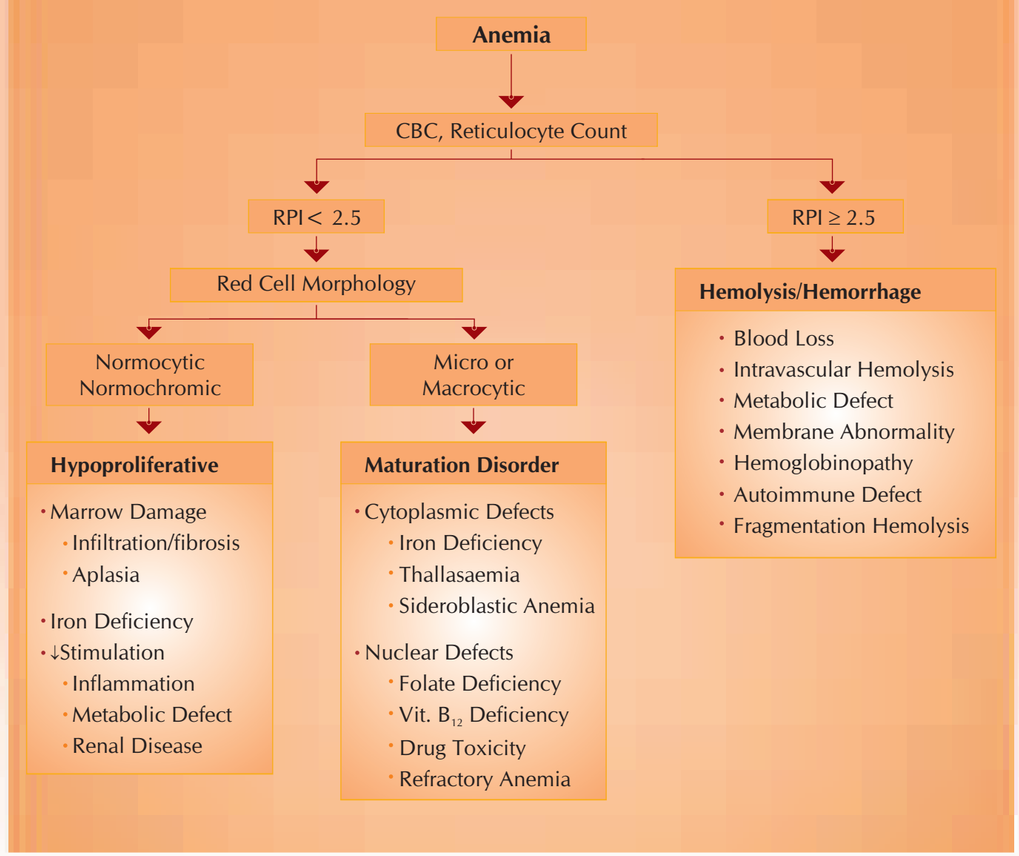
ANEMIA & HEMOGLOBINOPATHY

Anemia meaning "without blood", is a deficiency of red blood cells (RBCs) and/or hemoglobin. It is the most common disorder of the blood. There are several kinds of anemia, produced by a variety of underlying causes

Metropolis Test Menu & Test Profiles for Anemia & Hemoglobinopathy Diagnosis

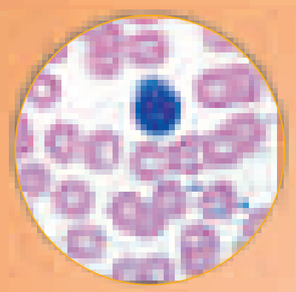
Test Menu
Abnormal Hemoglobin Studies (Capillary Electrophoresis), Acidified Ham's Test, Bilrubin (Indirect), Bone Marrow Examination, Cold Agglutin, Coombs-Direct, Coombs-Indirect, Cytogenics For Fanconi's Anemia, Erythropoetin, Ferritin, Free Hemoglobin, G6PD, Hemoglobinuria, Intrinsic Factor Antibody, Iron Studies, Lactate Dehydrogenase, Meth-Hemoglobin, Osmotic Fragility, Parietal Cell Antibody (Serum), Peripheral Blood Smear, Plasma Hemoglobin, Red Blood Cell Folate, Reticulocyte Count, Reticulocyte Index, Rh Antibody, Serum Folate, Total Iron Binding Capacity, Transferrin, Vitamin B_{12}, Active B_{12}, Thallasaemia studies, Sickling Test, C-Reactive Protein
Test Profiles
Anemia Profile (Maxi): Hemogram, Reticulocyte Count, Serum Haptoglobulin, Iron, Ferritin, Total Iron Binding Capacity, Transferrin Saturation, Vitamin B_{12}, Folate Levels (Serum & RBC), C-Reactive Protein, Osmotic Fragility Test, G6PD Deficiency, Coomb's Test (Direct), Abnormal Hemoglobin Studies
Anemia Profile (Hemolytic): Hemogram, Reticulocyte Count, Serum Haptoglobulin, G6PD Deficiency, Osmotic Fragility Test, Abnormal Hemoglobin Studies, Coomb's Test (Direct)
Anemia Profile (Mini): Hemogram, Iron, Ferritin, Total Iron Binding Capactiy, Transferrin Saturation, C-Reactive Protien
Anemia Profile (Nutritional): Iron Studies, Ferritin, Vitamin B_{12}, Folate Levels
Anemia Profile (Pernicious): Parietal Cell Antibody, Intrinsic Factor Antibody, Vitamin B_{12}, Hemogram
Thallasaemia Profile: Hemogram, Iron Studies, Abnormal Hemoglobin Studies (Capillary Electrophoresis)

Physiologic Classification of Anemia



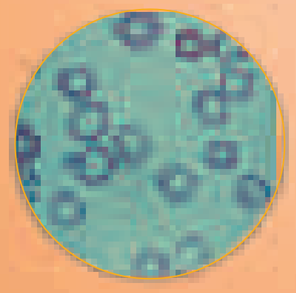
RPI: Reticulocyte Production Index; CBC: Complete Blood Count

Source: Harrison's Principles of Internal Medicine. Vol-I, 16th ed. USA: McGraw-Hill; 2005
J Assoc Physicians India. 2005 Dec; 53:1021-6.
Clinica Chimica Acta 2003; 330: 1 –30.
Ann Clin Biochem 1999; 36: 133-150.
Thalassemia, Beta; http://www.emedicine.com



Normocytic Normochromic Anemia

Macrocytic Anemia



Hypochromic Microcytic Iron Deficiency Anemia

Hemolysis

- Hemoglobinopathies are inherited disorders of globin [the protein component of hemoglobin (Hb)] affecting the structure, function or production of hemoglobin
- Thallasaemia and sickle cell anemia are the most common types of hemoglobinopathy
- General incidence of thallasaemia trait and sickle cell hemoglobinopathy in India varies between 3-17% and 1-44%, respectively

Sickle Cell Anemia

Thallasaemia Minor

Thallasaemia Major

A definitive diagnosis of hemoglobinopathies usually requires assays for abnormal hemoglobin and cytogenetic analyses

Abnormal Hemoglobin Studies by Capillary Electrophoresis
Specific Features and Advantages over HPLC
• Clear separation of HbE is possible
• Detects HbH and Barts even at low concentration
• Detection and presumptive identification of Hbs S, F, A, C, D, E, J, Q-India etc
• Simultaneous, quantitative results for percent HbA2, HbF, HbA and other abnormal hemoglobins
• Objective patient reports, unlike traditional methods

Test	**Specific Features**
Cytogenetics for Beta Thallasaemia	• Analysis of complete set of 23 loci including HbS and HbE detection • Can precisely determine whether a mutation is present any time after approximately 8 weeks of gestation • Diagnosis can be made using DNA obtained from amniocentesis or by chorionic villus sampling • DNA is amplified using the polymerase chain reaction technique and then is analyzed for the presence of the thallasaemia mutation using a panel of oligonucleotide probes corresponding to known thallasaemia mutations
HOLO Transcobalamin (Active B_{12})	• Serum Holo-Transcobalamin is a marker of Vitamin B_{12} status • This *in vitro* diagnostic assay offers quantitative measurement of the fraction of vitamin B_{12} bound to the carrier protein transcobalamin in the human serum or plasma • Identifies early subclinical stages of cobalamin deficiency • Hence, permits prompt initiation of treatment