

# ANEMIA & HEMOGLOBINOPATHY

## Metropolis Test Menu & Test Profiles for Anemia & Hemoglobinopathy Diagnosis

### Test Menu

Abnormal Hemoglobin Studies (Capillary Electrophoresis), Acidified Ham's Test, Bilirubin (Indirect), Bone Marrow Examination, Cold Agglutin, Coombs-Direct, Coombs-Indirect, Cytogenetics 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<sub>12</sub>, Active B<sub>12</sub>, Thallasaemia studies, Sickling Test, C-Reactive Protein

### Test Profiles

**Anemia Profile (Maxi):** Hemogram, Reticulocyte Count, Serum Haptoglobin, Iron, Ferritin, Total Iron Binding Capacity, Transferrin Saturation, Vitamin B<sub>12</sub>, 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 Haptoglobin, G6PD Deficiency, Osmotic Fragility Test, Abnormal Hemoglobin Studies, Coomb's Test (Direct)

**Anemia Profile (Mini):** Hemogram, Iron, Ferritin, Total Iron Binding Capacity, Transferrin Saturation, C-Reactive Protein

**Anemia Profile (Nutritional):** Iron Studies, Ferritin, Vitamin B<sub>12</sub>, Folate Levels

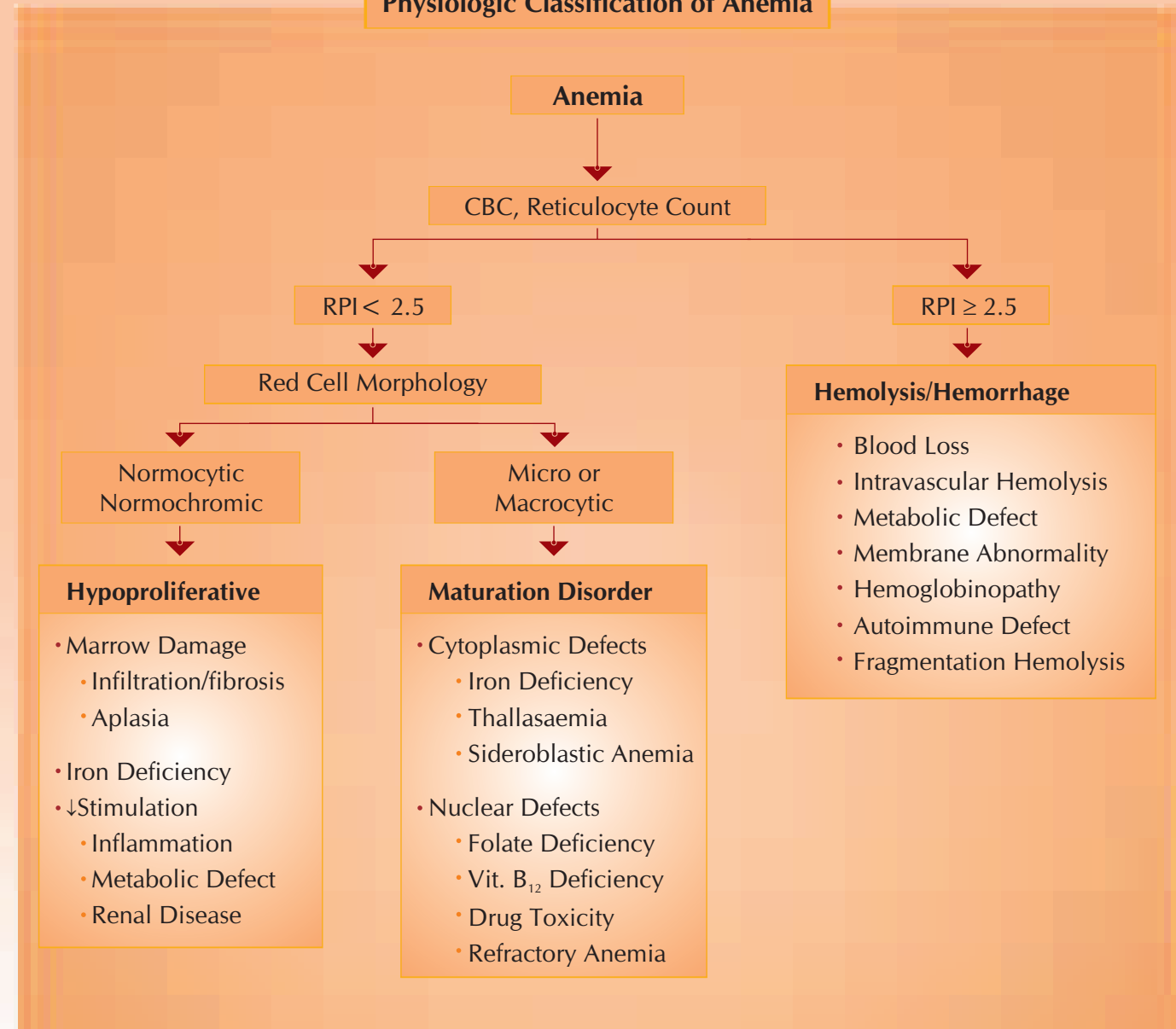
**Anemia Profile (Pernicious):** Parietal Cell Antibody, Intrinsic Factor Antibody, Vitamin B<sub>12</sub>, Hemogram

**Thalassaemia Profile:** Hemogram, Iron Studies, Abnormal Hemoglobin Studies (Capillary Electrophoresis)

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.  
 Thalassaemia, Beta; http://www.emedicine.com

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

### Physiologic Classification of Anemia



RPI: Reticulocyte Production Index; CBC: Complete Blood Count

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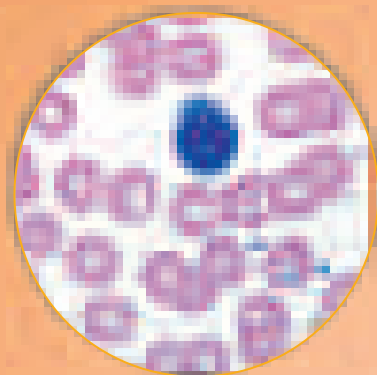
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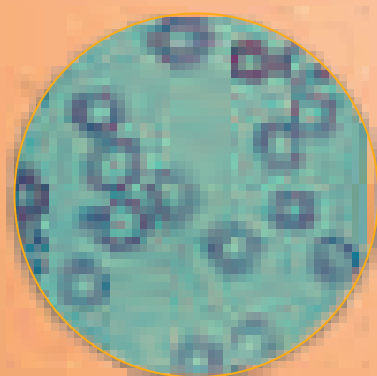
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Normocytic Normochromic Anemia



Macrocytic Anemia



Hypochromic Microcytic Iron Deficiency Anemia

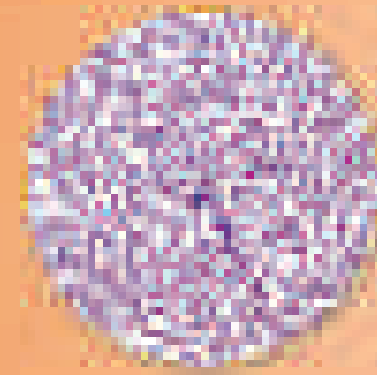


Hemolysis

- 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 HbA<sub>2</sub>, HbF, HbA and other abnormal hemoglobins
- Objective patient reports, unlike traditional methods

Test	Specific Features
Cytogenetics for Beta Thallasaemia	<ul style="list-style-type: none"> <li>• Analysis of complete set of 23 loci including HbS and HbE detection</li> <li>• Can precisely determine whether a mutation is present any time after approximately 8 weeks of gestation</li> <li>• Diagnosis can be made using DNA obtained from amniocentesis or by chorionic villus sampling</li> <li>• 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</li> </ul>
Holo Transcobalamin (Active B <sub>12</sub> )	<ul style="list-style-type: none"> <li>• Serum Holo-Transcobalamin is a marker of Vitamin B<sub>12</sub> status</li> <li>• This <i>in vitro</i> diagnostic assay offers quantitative measurement of the fraction of vitamin B<sub>12</sub> bound to the carrier protein transcobalamin in the human serum or plasma</li> <li>• Identifies early subclinical stages of cobalamin deficiency</li> <li>• Hence, permits prompt initiation of treatment</li> </ul>