# Hereditary Hemolytic Anemias Outline

- Membrane Defects
  - Hereditary Spherocytosis (extravascular hemolysis)
  - Hereditary Elliptocytosis (extravascular hemolysis)
- Enzyme Defects
  - G6PD Deficiency (extravascular hemolysis)
  - Pyruvate Kinase Deficiency (extravascular hemolysis)
- Globin defects
  - Hemoglobinopathies
  - Thalassemias

# **The Hereditary Hemolytic Anemias**

**Mary Louise Turgeon** 

#### The Hereditary Hemolytic Anemias:

The Hereditary Hemolytic Anemias William C. Mentzer, Gail M. Wagner, 1989 Hereditary Hemolytic Anemias Source Wikipedia, 2013-09 Please note that the content of this book primarily consists of articles available from Wikipedia or other free sources online Pages 28 Chapters Alpha thalassemia Beta thalassemia Congenital hemolytic anemia Delta thalassemia Glucose 6 phosphate dehydrogenase deficiency Hemoglobinopathy Hereditary elliptocytosis Hereditary persistence of fetal hemoglobin Hereditary spherocytosis Hereditary stomatocytosis Hexokinase deficiency Minkowski Chauffard syndrome Pyruvate kinase deficiency Sickle cell disease Sickle cell trait Southeast Asian ovalocytosis Triosephosphate isomerase deficiency Excerpt Sickle cell disease SCD or sickle cell anaemia SCA or drepanocytosis is an autosomal recessive genetic blood disorder with overdominance characterized by red blood cells that assume an abnormal rigid sickle shape Sickling decreases the cells flexibility and results in a risk of various complications. The sickling occurs because of a mutation in the hemoglobin gene Life expectancy is shortened In 1994 in the US the average life expectancy of persons with this condition was estimated to be 42 years in males and 48 years in females but today thanks to better management of the disease patients can live into their 80s or beyond Sickle cell disease occurs more commonly among people whose ancestors lived in tropical and sub tropical sub saharan regions where malaria is or was common Where malaria is common carrying a single sickle cell gene sickle cell trait confers a fitness Specifically humans with one of the two alleles of sickle cell disease show less severe symptoms when infected with malaria Sickle cell anaemia is a form of sickle cell disease in which there is homozygosity for the mutation that causes HbS Sickle cell anaemia is also referred to as HbSS SS disease haemoglobin S or permutations of those names In heterozygous people that is those who have only one sickle gene and one normal adult haemoglobin gene the

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