Sickle Cell Disease (HBB)

Sickle cell disease is caused by mutations in the HBB gene, which encodes for the protein beta-globin, a component of hemoglobin. The main form of the disease is sickle cell anemia, where the red blood cells are bent into a sickle shape. Sickle cells break down more quickly than normal cells, often resulting in anemia. The irregular cell shape tends to block blood vessels, which can lead to pain and ischemia of organs, including strokes. Jaundice and damage to the spleen often occur. In some cases, pulmonary hypertension can occur and lead to heart failure. Other than sickle cell formation, other abnormal forms of hemoglobin can form. The various symptoms of sickle cell anemia shorten live expectancy to about 40 to 60 years.

Sickle cell disease tends to be concentrated in particular ethnic groups. About 1 in 500 African Americans have sickle cell disease, while the figure is about 1 in 1,000 to 1 in 1,400 for Hispanic Americans. In all about 100,000 Americans suffer from sickle cell disease. As the population of the USA is around 321 million, this means that about 1 in 3210 Americans has the disease, making it the most common inherited blood disease. The faulty gene is autosomal recessive, typically requiring both parents to be asymptomatic carriers of the faulty gene copy.

Sources

Bender M.A. & Seibel, G.D. (2003), “Sickle Cell Disease,” in Pagon, R.A. et al., editors, GeneReviews [Internet]. See http://www.ncbi.nlm.nih.gov/books/NBK1377/

NIH, Genetics Home Reference: HBB gene. See http://ghr.nlm.nih.gov/gene/HBB

NIH, Genetics Home Reference: Sickle Cell Disease.
See http://ghr.nlm.nih.gov/condition/sickle-cell-disease

Recombine Website. Sickle Cell Anemia.
See https://recombine.com/diseases/sickle-cell-anemia

US Census Bureau http://www.census.gov/popclock/