Sickle hemoglobin (HbS) allele and sickle cell disease: a HuGE review.

Abstract: Sickle cell disease is caused by a variant of the beta-globin gene called sickle hemoglobin (Hb S). ... either two copies of Hb S or one copy of Hb S plus another beta-globin variant (such as Hb C) are required for disease expression. ... individuals with sickle cell disease exhibit significant morbidity and mortality. Symptoms include chronic anemia, acute chest syndrome, stroke, ... Disease expression is variable and is modified by several factors, the most influential being genotype. Other factors include beta-globin cluster haplotypes, alpha-globin gene number, and fetal hemoglobin expression. ... newborn screening, better medical care, parent education, and penicillin prophylaxis have successfully reduced morbidity and mortality due to Hb S.