Haplotypes and alpha globin gene analyses in sickle cell anaemia patients from Kenya.

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Abstract:

Over 60 patients from the Luo and Luhya tribes of Western Kenya, aged 1-23 years, with severe sickle cell anaemia were evaluated through haematological and gene mapping analyses. Nearly all (56 of 58 tested) were homozygous for haplotype 20 (Antonarakis et al, 1984) which is also frequently present in SS patients of the Central African Republic. All patients had a severe haemolytic anaemia with low Hb F levels and low levels of G gamma chains. An alpha-thalassaemia-2 heterozygosity (-alpha-alpha alpha; -3.7 kb deletion) was present in 26 of 53 patients tested; one patient was a homozygote [f(-alpha) = 0.255]. The alpha-thal-2 was type I in all but one subject with this deficiency; the one exception had an alpha-thal-2 heterozygosity, type II. Heterozygosity for the alpha-thal-2 did not affect the clinical condition nor the haematology; Hb F levels were somewhat lower in SS patients with -alpha/alpha alpha than in those with alpha alpha/alpha alpha. A high frequency was observed for the absence of an Xba I restriction site 5' to the zeta globin gene; the frequency of this anomaly [f(Xba I-)] was estimated at 0.39 for the chromosome with two alpha globin genes and at 0.74 for that with the alpha-thal-2 deletion. An Apa I restriction site polymorphism was observed in the IVS-II of the alpha 2 globin gene; 13 alpha 2 genes of 53 normal (alpha alpha/) chromosomes had this restriction site which was absent in the hybrid alpha globin gene of the -alpha/chromosome.