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# Screening of *HBB*S* in populations from Alentejo and implementation of a SNaPshot® based system for *HBB*S* haplotyping

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It is well demonstrated that the frequency of sickle cell allele, *HBB*S,* reaches the highest values in regions of endemic malaria, due to resistance to the malaria infection conferred by *HBB*S*. Its presence in Portugal has been explained as the consequence of past population migrations, which in part also explain the uneven distribution of *HBB*S* across the country, where it tends to increase in frequency from the north towards the south-central regions.
In order to better understand how *HBB*S* was introduced in Portugal, the screening of *HBB* was performed in 266 individuals from Alentejo (Coruche, Serpa (Pias) and Alcácer do Sal). Then, to further infer the origin of the detected *HBB*S* chromosomes, their haplotypic background, encompassing the entire cluster of β-globin genes, was examined through a SNaPshot® Multiplex system that was purposely implemented as an alternative to the conventional RFLP-based method, and that allowed to interrogate the most informative positions defining the *HBB*S* haplotypes.
19 individuals harboured the c.20A>T mutation that underlies *HBB*S*, whose frequency was 2.2, 2.9 and 7.7% in Coruche, Alcácer do Sal and Serpa, respectively. The last value is the highest up to now reported in the country, where foci of high prevalence of HBB*S carriers have been previously identified in south regions.
Among the chromosomes bearing HBB*S, 29% harboured the Benin haplotype, 13.1% the Senegal and 10.5% the Bantu. Remarkably, whereas in Alcáçer do Sal and Coruche only Senegal and Bantu haplotypes were found, in Serpa all haplotypes were Benin.
Taking into account the global distribution of *HBB*S* haplotypes, the findings obtained reinforce a scenario before proposed positing that the introduction of *HBB*S* in south Portugal was mediated by gene influx events with distinct sources: one from the region encompassing the Mediterranean basin (captured by the Benin haplotype), and other from sub-Saharan Africa, likely afforded by the transatlantic slave trade (captured by the Senegal and Bantu haplotypes).