Unusual low sickle cell hemoglobin level

In the January 2013 issue of this Journal, Joly and Colleagues have described an unusual case of sickle cell trait.[1] The patient, originating from Ivory Coast, exhibits a low HbS level (12%) with no phenotypic manifestations. The mutation was found on a β-globin cluster haplotype of the Benin type. The Authors argued that the low percentage of HbS in circulating blood results from a somatic deletion of the HBB gene bearing the β^S mutation. Surprisingly, the patient's relatives (mother and son) display a higher, yet unusually low HbS level of approximately 34-35%.

A very similar case had been described some years ago when a Moroccan female exhibited a low HbS level of approximately 11%.[2] The β^S chromosome haplotype was also of the Benin type. Molecular analyses have demonstrated the occurrence of a point mutation at the promoter element CACCC (CACCC → CATCC) in cis of the sickle cell mutation.[3] One would wonder whether the authors of the recent work on the Ivorian case have extended the sequencing analysis down to the promoter region of the β^S-globin allele.

Faouzi Baklouti,[1] and Jean Delaunay[2]

[1]mRNA Metabolism in Normal and Pathological Cells; CNRS, Université Lyon 1, Villeurbanne; [2]INSERM U 779, Faculté de Médecine Paris-Sud, Univ. Paris-Sud, Le Kremlin-Bicêtre, France

Correspondence: faouzi.baklouti@univ-lyon1.fr

doi:10.3324/haematol.2013.087973

Key-words: HBB cell, sickle cell, loss of heterozygosity, hemopoietic lineage.

Information on authorship, contributions, and financial & other disclosures was provided by the authors and is available with the online version of this article at www.haematologica.org.

References

1. Joly P, Schluth-Bolard C, Lacan P, Barro C, Pissard S, Labalme A, et al. HBB loss of heterozygosity in the hemopoietic lineage gives rise to an unusual sickle-cell trait phenotype. Haematologica. 2013;98(1):e7-8.
2. Baklouti F, Francina A, Dorleac E, Richard G, Rosenberg D, Godet J, et al. Association in cis of beta +-thalassemia and hemoglobin S. Am J Hematol. 1987;26(3):237-45.
3. Baklouti F, Ouazana R, Gonnet C, Lapillonne A, Delaunay J, Godet J. Beta+-thalassemia in cis of a sickle cell gene: occurrence of a promoter mutation on a beta s chromosome. Blood. 1989;74(5):1817-22.