HBB loss of heterozygosity in the hemopoietic lineage gives rise to an unusual sickle-cell trait phenotype

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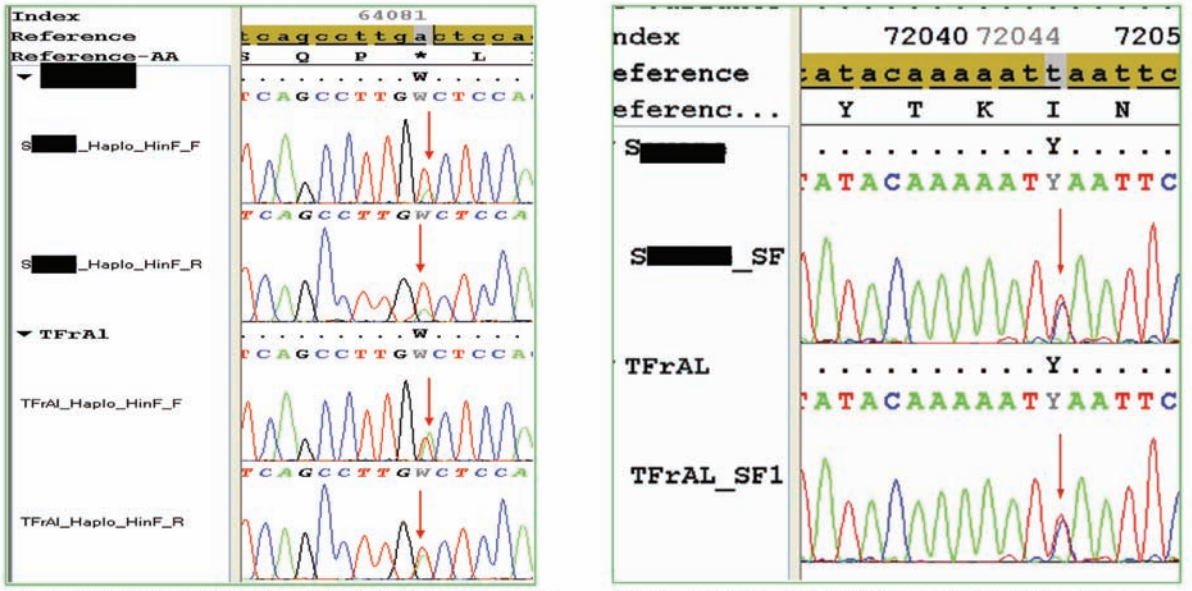
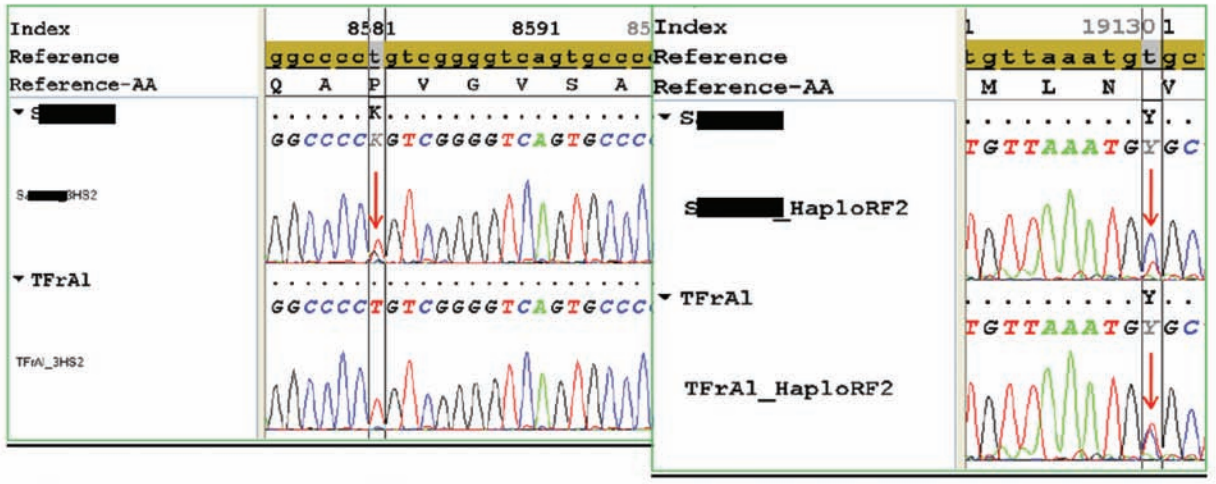


(A): Last identified position with peaks disequilibrium at Sanger Sequencing for the propositus: U01317.1:g.64081. Up: Propositus. Down: Internal control (mother of the propositus who bears the same SNP at the heterozygous state but not the somatic deletion).

(B): First identified position without peaks disequilibrium at Sanger direct sequencing for the propositus (U01317.1:g.72044). Up: Propositus. Down: Internal control (mother of the propositus who bears the same SNP at the heterozygous state but not the somatic deletion).

Online Supplementary Figure S1. 3'-breakpoint determination.



(A): Part of Sanger direct sequencing of the *HS2* gene (5' extremity of the β-globin gene cluster). For the propositus (up), peaks disequilibrium can be observed at position U01317.1:g.8581 for a T>G SNP. No comparison could be made with the mother of the propositus (down) as she did not bear this particular SNP.

(B): Part of Sanger direct sequencing near the *HBE* gene (about 10 kb after the *HS2* gene). For the propositus (up), peaks disequilibrium can be observed at position U01317.1:g.19130 for a T>C SNP, contrary to the mother of the propositus (down) who also bears the same SNP at the heterozygous state but not the somatic deletion (internal control).

Online Supplementary Figure S2. 5'-breakpoint determination.