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A new alpha-globin variant with increased oxygen affinity in a Swiss family : Hb Frauenfeld [alpha 138(H21)Ser–

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A new alpha-globin mutation [alpha 138(H21)Ser–] was found in a 55-year-old male proband with an erythrocytosis known since his youth. Cation exchange high performance liquid chromatography (HPLC) revealed an additional peak eluting slightly before Hb A indicating the presence of a variant. The peak area of the variant was approximately one-third that of Hb A suggesting an alpha-globin variant. Matrix-assisted laser desorption ionization-time-of-flight mass spectrometry analysis confirmed the mutation at the protein level. The variant is also detectable with isoelectric focusing and reversed phase HPLC. DNA analysis revealed a heterozygous sequence mutation at codon 138 of the alpha2 gene. A Ctransition at the second nucleotide of the codon indicated a Ser–exchange. The variant showed increased oxygen affinity and was named Hb Frauenfeld.
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