

Publication**A new alpha-globin variant with increased oxygen affinity in a Swiss family
: Hb Frauenfeld [alpha 138(H21)Ser–****Journal Article (Originalarbeit in einer wissenschaftlichen Zeitschrift)****ID** 1192763**Author(s)** Hochuli, Michel; Zurbruggen, Karin; Schmid, Marlis; Speer, Oliver; Rochat, Philippe; Frauchiger, Beat; Kleinert, Peter; Schmutz, Markus; Troxler, Heinz**Author(s) at UniBasel** [Frauchiger, Beat](#) ;**Year** 2009**Title** A new alpha-globin variant with increased oxygen affinity in a Swiss family : Hb Frauenfeld [alpha 138(H21)Ser–<Phe, TCC<TTC (alpha 2)]**Journal** Hemoglobin**Volume** 33**Number** 1**Pages / Article-Number** 54-58**Keywords** -Globin variant, PolyCAT A, High performance liquid chromatography (HPLC), Mass spectrometry (MS), Erythrocytosis

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 C transition 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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