Hemoglobin Hakkari: an autosomal dominant form of beta thalassemia with inclusion bodies arising from de novo mutation in exon 2 of beta globin gene.

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Public Summary:

Scientific Abstract:
Certain beta globin gene mutations produce a thalassemia major phenotype in the heterozygous state. While most such patients have thalassemia intermedia, we describe a young Guatemalan child with a de novo mutation in the beta globin gene, codon 31 T --> G (Hemoglobin Hakkari), who developed severe anemia at the age of 10 months and remains transfusion-dependent. The substitution of B13 leucine with arginine in the beta globin results in alteration of a critical heme contact point resulting in an extremely unstable variant hemoglobin and a clinical picture that is characterized by ineffective erythropoiesis and numerous intracytoplasmic inclusions within the erythrocyte precursors of the bone marrow. .