The frequency of homozygous deletion of a developmentally regulated vh gene (humhv3005) is increased in patients with chronic idiopathic thrombocytopenic purpura.

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摘要

Abstract

Little is known of the genetic factors that may contribute to the development of chronic idiopathic thrombocytopenic purpura (cITP). We have previously shown that a developmentally regulated Vh gene (Humhv3005) is absent in 10/41 (24%) of patients with systemic lupus erythematosus while it is absent in only 7/88 (8%) of normal controls. This finding suggests that a homozygous deletion of an Ig variable (V) gene may alter the immune system and thus predispose the host to an autoimmune disorder. We have analyzed the same gene in 44 patients with cITP and found that Humhv3005 and like genes were absent in a higher percentage of patients (14 of 44, 31.8%) than they were absent in either normals (7/88, 8%, p = 0.002) or thrombocytopenic patients without cITP (6/53, 11.3%, p = 0.042); the hv3005 deletion frequency in the latter group did not differ from that in normals (P = 0.74). These data suggest that deletions of Humhv3005 and/or highly homologous Vh genes may predispose individuals to the development of cITP, and may contribute toward production of pathogenic antiplatelet antibodies