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Characteristics of Japanese Thalassemia

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ABSTRACT Thalassemia is relatively rare in Japan. Of 387 b-thalassemia cases of Japanese, homozygotes for b^+- and b^0-thalassemias were twenty-two (5.7%) and one (0.26%), respectively. The rest (94%) were of b-thalassemia trait. Ten different kinds of b-thalassemia mutations comprised about 80% of all cases. Half of these "common" mutations were unique to Japanese and another half were possibly from abroad. Sixty-nine a-thalassemia cases were analyzed, in which HbH and a-thalassemia trait comprised nineteen and fifty, respectively. Most of the a^o- and a^+-thalassemia chromosomes were of Southeast Asian type (--SEA) and $-a^{3.7}$ type, respectively. The frequency of a^+-thalassemia as well as triplication of a-globin gene was high in northern Japan. Recent increase in the number of immigrants from Southeast Asia seems to raise the number of a-thalassemia found in Japan. They comprise at least 20% of the a-thalassemia. Six mutants classified into dominant-type b-thalassemia were found. All of them exhibited moderate anemia and marked anisopoikiocytosis. Heinz body varied in degree from copious to rare or even absent. Any mutation at initiation codon demonstrated marked microcytosis and erythremia. The breakpoint determination for large deletion-type thalassemia became feasible by estimation of gene dosage and PCR. Thus, the precise breakpoints for Filipino-type a^o-thalassemia (--FIL) and Japanese type-2 db-thalassemia were disclosed. The genetic diagnosis for these thalassemias are now readily conducted by gap PCR. The characterization of several new large deletions is being carried out.

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