

For Presentation to American Society of Human Genetics
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HELL, J. V., KODANI, H., BRENER, R., and ANDERSON, R. C., A comparison of the frequency of five genes in Japanese and Caucasian populations.

Assuming panmixia, the incidence of the gene responsible for any rare autosomal recessive trait present in a population may be calculated from the relationship $q = \frac{c(1 - k)}{(16k - 15c - ck)}$, where q = the frequency of the recessive gene responsible for the trait in question, k = the proportion of first cousin marriages among the parents of affected persons, and c = the proportion of first cousin marriages among the population as a whole. Departures from panmixia, such as of course occur in human populations, tend to invalidate the use of the above formula, but the extent of the error cannot be estimated until more is known of the breeding structure of human populations. It is felt that the formula, if used judiciously, is capable of yielding a first approximation to gene frequencies. -- An attempt has been made through the use of this formula to determine the frequencies in Japanese and European Caucasian populations of the autosomal recessive genes responsible for complete albinism, infantile amaurotic idiocy, ichthyosis congenita, congenital total color blindness, and xeroderma pigmentosum, assuming for each disease that all cases are due to the same recessive gene. The factor c has been given a value of 0.01 for European populations, while for Japanese populations, following our own investigations, c has been given a value of 0.06. k has been estimated for the various diseases from a summary of the literature; there is a considerable potential error in this estimate. There appears to be no significant difference in the frequencies in European Caucasian and Japanese populations of the recessive genes responsible for the first four diseases mentioned, but the last gene, responsible for xeroderma pigmentosum, may be more frequent in Japanese.

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