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Brief Communication

Carbonic Anhydrase-I Polymorphism in a Philippine Aboriginal Population

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SUMMARY

Polymorphism of carbonic anhydrase-I (CA_1) was found by electrophoresis in an aboriginal group of Mindanao, Philippines, with a remarkably high frequency of variant types. The frequency of the variant allele was estimated at .256. The variant isozyme designated CA_1 3Negrito (CA_1 3N) is electrophoretically indistinguishable from the "Guam" variant and may be regarded as a potential anthropological marker in the Western Pacific.

INTRODUCTION

Most genetic variants of human CA_1 detected by electrophoresis have been known to be infrequent [1], although Blake and Kirk [2] recently discovered the polymorphic occurrence of several CA_1 variants among the Australian aboriginals.

During the course of population genetic studies of the aboriginal groups of the Philippines [3, 4], an electrophoretic variant indistinguishable from CA Ic Guam, or CA_1 3Guam, according to the naming system recently proposed by Blake [5], was discovered with a remarkably high frequency in a so-called Negrito group of northern Mindanao. The finding seems to be of particular genetic and anthropological significance and will be described below.

MATERIALS AND METHODS

Blood specimens from 82 subjects belonging to the Mamanwa tribe were obtained by venipuncture in the Province of Agusan del Norte, northern Mindanao. They were kept cool with wet ice and transported by air to Tokyo, where red cells were separated and preserved in liquid nitrogen.

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CA_1 types were examined by horizontal starch gel electrophoresis using a Tris-EDTA-Borate buffer (pH 8.6), and 4-methylumbelliferyl or β-naphthyl acetate as substrate [6].

RESULTS AND DISCUSSION

The zymogram of the variant CA_1 phenotypes is shown in figure 1. Both the heterozygote and the variant homozygote presumably were found. The variant was tentatively named CA_1 3N, the heterozygote being CA_1 1-3N, and the homozygote CA_1 3N. However, the comparison run with samples of CA_1 1-3Guam, as well as the Australian variants, showed that the isozyme CA_1 3N is electrophoretically indistinguishable from CA_1 3Guam (N. M. Blake, personal communication, 1978).

As shown in table 1, the variant phenotypes were found to be remarkably common in the present study. The frequency of the postulated variant allele (.256) is probably the highest for the CA_1 variants ever detected.

The Mamanwas are the aboriginal group inhabiting hilly areas of the northernmost part of Mindanao. They are regarded as one of the Negrito groups, the true aboriginals of the Philippines. They are not as small as the Aetas of Luzon, but are very dark-skinned and frizzly-haired, forming a sharp contrast to the other inhabitants of the Philippines who are predominantly Mongoloids.

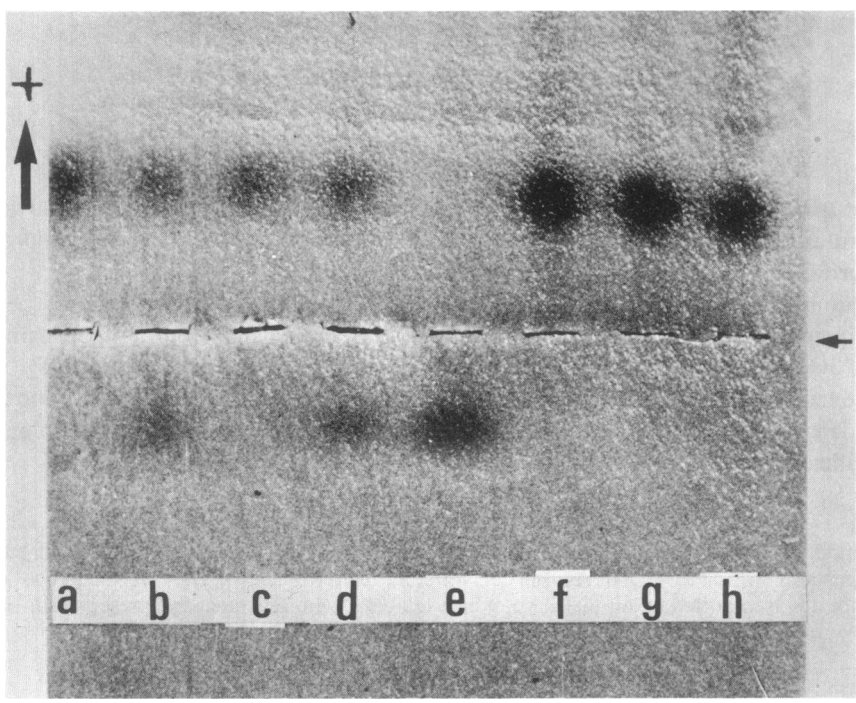

FIG. 1. —Starch gel showing variant CA_1 phenotypes. β-Naphthyl acetate was used to stain CA_1 isozymes. *a, c, f, g, h*: CA_1 1; *b, d*: CA_1 1-3N (heterozygote); *e*: CA_1 3N (homozygote). *Small arrow* indicates position of start.

TABLE 1

DISTRIBUTION OF THE VARIANT CA_1 PHENOTYPES AMONG 82 SAMPLES FROM MAMANWA, NORTHERN MINDANAO

NUMBER	PHENOTYPES				GENE FREQUENCY
	CA_1 1	CA_1 1 – 3N	CA_1 3N	Total	χ^2 (1 df) = .0159, $P > .70$
Observed	46	30	6	82	CA_1^1: .7439
Expected	45.38	31.24	5.38	82	CA_1^{3N}: .2561
					SE: .0341

Unfortunately, no detailed information was available as to the degree of inbreeding among the Mamanwa subjects from whom the blood samples were obtained, or the rate of admixture from the surrounding groups. However, the fact that the subjects who possess the variant CA_1 type came from various parts of the Agusan and Surigao provinces indicates that this variant may have wide distribution among the Mamanwa population. Also, it is unlikely that the variant was introduced by admixture from the other groups, since the screening for CA_1 variants carried out in other Philippine populations, including those living adjacent to the Mamanwas, thus far failed to detect a variant.

CA_1 variants, probably identical to CA_1 3Guam, have been reported to occur sporadically among populations of the Western Pacific, that is, among Micronesians (Guam and Saipan), Indonesians (Java), Filipinos (U. S. residents), and Malays [1, 5]. Investigation of the primary structure of the CA_1 3N enzyme is necessary to identify this variant as CA_1 3Guam. At present, however, it may be postulated that the variant was once present at high frequency in a certain aboriginal group of Southeast Asia or the Western Pacific, of which the Mamanwas may be one of the few survivors.

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A SYMPOSIUM ON GENETICALLY-DETERMINED DISEASE will take place in Dunedin, New Zealand, on February 18 and 19, 1980. Invited speakers will discuss the genetic basis and pathogenesis of disease in the light of newly-revealed mechanisms of molecular biology. There will be sessions on recessively-inherited, autoimmune, and dominantly-inherited disorders. Papers describing studies on disease with a genetic predisposition, relevant to the above approach, are invited. The program coordinator is Dr. D. D. Adams. Enquiries should be addressed to: Dr. R. J. M. Gardner, Hon. Secretary Genetics Symposium, Department of Paediatrics & Child Health, University of Otago Medical School, PO Box 913, Dunedin, New Zealand.