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Genetic markers in a Malaysian population: variants of uridine monophosphate kinase (UMPK), phosphoglycolate phosphatase (PGP) and pancreatic amylase (AMY_2)

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Summary. Three genetic markers, red-cell UMPK, PGP and serum AMY_2 were investigated in Malaysians of Malay, Chinese and Indian ancestries using starch-gel and agarose-gel electrophoresis. UMPK was found to be polymorphic in all three races. Variants were observed for PGP in Malays; in Indians it is a polymorphic marker whereas it is monomorphic in Chinese. AMY_2 was polymorphic only in Indians. The $UMPK^1$ frequencies in Malays, Chinese and Indians, respectively, are 0·851, 0·880 and 0·942. The PGP^1 frequencies are 0·991, 1·000, 0·962, and the AMY_2^1 frequencies are 1·000, 1·000 and 0·983.

1. Introduction

The enzyme UMP kinase, also known as pyrimidine nucleoside monophosphate kinase, is important in the catalysis of the first step in the phosphorylation of UMP (uridine monophosphate) to UDP (uridine diphosphate) in the complicated metabolic pathway leading to products required for RNA and DNA synthesis.

Genetic polymorphism of UMPK, demonstrable by starch-gel electrophoresis of human red-cell lysates, was first reported by Giblett, Anderson, Chen, Teng and Cohen (1974). They reported the inheritance of different phenotypes of this enzyme to be of an autosomal co-dominant type, controlled by three alleles — $UMPK^1$, $UMPK^2$ and $UMPK^3$.

Gene frequency data of this enzyme on various populations especially in the south-east Asian region, is rather limited. UMPK seems a useful genetic marker and we decided to survey the three major ethnic groups in Malaysia for its gene frequency distribution.

The functional role of human phosphoglycolate phosphatase (PGP) is obscure. It may have an important regulatory influence on oxygen transport in man. In their investigation Barker and Hopkinson (1978) found the enzyme PGP to be very polymorphic among Europeans. The six electrophoretic phenotypes they observed were determined by three alleles, PGP^1, PGP^2 and PGP^3, at an autosomal locus. Also in this report we present results of a study of this locus on the Malays, Chinese and Indians of Malaysia.

Human amylase is the enzyme involved in the endohydrolysis of 1,4-α-glycosidic linkages in polysaccharides containing at least three 1,4-α-linked glucose units. Kamaryt and Laxova (1965, 1966) reported the existence of genetic variants of human amylase enzymes and that the amylase gene products found in the serum are determined by two closely linked autosomal loci, AMY_1 (the salivary locus) and AMY_2 (the pancreatic locus). Family data by Merritt, Rivas, Bixter and Newell (1973) and Kompf, Sibert and Ritter (1979) confirmed the formal hypothesis of an autosomal dominant mode of inheritance for each variant of AMY_2.

Data on the distribution of AMY_2 variants for several populations of south-east

Asia are limited, so we have surveyed the three major races of Malaysia for AMY_2 distribution.

2. Materials and methods

Blood samples were collected into heparinized tubes from healthy unrelated donors of Malay, Chinese and Indian ancestries through the National Blood Transfusion Service Centre, Kuala Lumpur. The red cells were separated from the sera by centrifugation. Glycerol was used as the preservative for red cells. All samples were stored at −70°C before use.

The haemolysates were used to type for UMPK and PGP. UMPK typing was as in Giblett *et al.* (1974). For PGP, a horizontal starch-gel electrophoresis was carried out using a continuous tris/EDTA/$MgCl_2$/TEMM buffer system (Barker and Hopkinson 1978). The staining procedure used was that of Blake and Hayes (1980).

Serum specimens were used to type for AMY_2 by agarose-gel electrophoresis using the methods outlined by Kompf *et al.* (1979).

3. Results and discussion

The results for UMPK phenotype and gene frequency distribution are presented in table 1. All three ethnic groups showed polymorphism for this enzyme. Of the three groups, UMPK 2-1 seems to be most prevalent among the Malays, occurring at 25% incidence. Statistically, there is good agreement between the distribution of phenotypes observed and those expected on the basis of Hardy-Weinberg equilibrium for all three races.

Table 1. UMPK phenotype and gene frequencies in Malaysians.

	UMPK types (no. and phenotype freq. (%))				Gene frequencies		
Population	UMPK 1	UMPK 2-1	UMPK 2	Total	$UMPK^1$	$UMPK^2$	SE
Malays	122(72·62)	42(25·00)	4(2·38)	168	0·851	0·149	0·019
Chinese	98(78·40)	24(19·20)	3(2·40)	125	0·880	0·120	0·021
Indians	108(89·26)	12(9·92)	1(0·83)	121	0·942	0·058	0·051

No data are available yet for the UMPK phenotype distribution of the people in the south-east Asian region except for that of Tan, Teng, Ganesan, Lau and Lie-Injo (1979), who typed the Kadazans of North Borneo, and Omoto, Misawa, Harada, Sumpaico, Medado and Ogonuki (1978), who typed Philippine Negritos, the Aetas, for UMPK distribution. The $UMPK^2$ gene frequency of the Kadazans were reported to be 0·041, which is lower than any of the values obtained for the Malays, Chinese and Indians. The $UMPK^2$ frequency of the Aetas was found to be 0·345, which is much higher than any of the values obtained in this investigation. The comparative study is not beyond expectation; although the Kadazans may have migrated to North Borneo from the Malay Peninsular during prehistoric times (Koentjaraningrat 1975), they have become so isolated geographically and linguistically that they would be better regarded as one other population in Borneo. The Aetas on the other hand are postulated to be remnants of the longest-living population in the region whose place of origin could not be ascertained. Physically and genetically they are thought to be related to the aboriginal Negritos of Malaysia and the Mincopies of the Andaman Islands; it is therefore hardly surprising that their gene frequency distributions do not approximate any of the populations investigated in this study.

In our Malaysian sample we did not observe phenotype variants of the rare allele $UMPK^3$, which was reported to have occurred quite commonly in the Cree Indians (Giblett *et al.* 1974) and in the isolated Warao Indians of Venezuela (Gallango and Suinaga 1978).

Giblett *et al.* (1974) reported that the $UMPK^2$ gene frequency of the Caucasians investigated was 0·05, while the $UMPK^2$ gene frequency of the American Orientals (largely of Japanese origin) was 0·07. Harada, Itoh and Misawa (1975) sampled Japanese in Japan and found the $UMPK^2$ gene frequency to be 0·0528.

The PGP phenotype and gene frequency distribution of the Malays, Chinese and Indians is presented in table 2. In this investigation only two phenotypes, PGP 1 and PGP 2-1 were observed in all three populations. Variants were observed for PGP in Malays and Indians. In Indians, the frequency of PGP^2 reach polymorphic proportions. Chinese were found to be monomorphic for PGP^1. No rare variants were detected. On comparing the observed phenotypes to those expected, assuming Hardy-Weinberg equilibrium, there was no statistically significant difference between them.

Table 2. PGP phenotype and gene frequencies in Malaysians.

	PGP types (no. and phenotype freq. (%))			Gene frequencies		
Population	PGP 1	PGP 2-1	Total	PGP^1	PGP^2	SE
Malays	165(98·21)	3(1·97)	168	0·991	0·009	0·005
Chinese	125(100·00)	0(0·00)	121	1·000	0·000	0·000
Indians	112(92·56)	9(7·44)	121	0·962	0·037	0·012

Barker and Hopkinson (1978) reported that the enzyme is very polymorphic in Caucasians. They found that 73·91% of the Asiatic Indians typed to be of phenotype PGP 1. This value is lower than the 92·56% obtained from the present investigation, which on the other hand is in agreement with that of Blake and Hayes (1980) where PGP 1 was found to range from 93 to 97% in the Indian population. In contrast to Caucasian, Indian, Amerindians and Australian Aborigines, PGP is not polymorphic in the Mongoloid Chinese and Malays. It is also not polymorphic in two other south-east Asian populations, the Balinese and Sumatrans (Blake and Hayes 1980).

The phenotype and gene frequency distribution of AMY_2 for the three races of Malaysia are presented in table 3. Polymorphism was observed only in the Indian population, at a level of 3·31% occurrence in the form of phenotype AMY 2-1. The Chinese and Malays are monomorphic.

Table 3. AMY_2 phenotype and gene frequencies in Malaysians.

	AMY_2 types (no. and phenotype freq. (%))			Gene frequencies		
Population	AMY_2 1	AMY_2 2-1	Total	AMY_2^1	AMY_2^2	SE
Malays	168(100·00)	0(0·00)	168	1·000	0·000	0·00
Chinese	125(100·00)	0(0·00)	125	1·000	0·000	0·00
Indians	117(96·69)	4(3·31)	121	0·983	0·017	0·01

In the present studies only the common phenotypes AMY_2 1 and AMY_2 2-1 were observed on the cathodal side of the gels. These phenotypes are similar to the AMY_2 variants observed by Kompf *et al.* (1979).

Our results are in agreement with that of Teng, Tan, Lopez Ng and Lie-Injo (1978), who found the enzyme AMY_2 to be monomorphic in the Malays and Chinese.

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Zusammenfassung. Drei genetische Marker, UMPK der Erythrozyten, PGP und AMY_2 des Serums, wurden bei Malaysiern von malayischem, chinesischem und indischem Ursprung mithilfe von Stärkegel- und Agargel-Elektrophorese untersucht. UMPK war bei allen drei Rassen polymorph. Bei PGP wurden Varianten bei Malayen gefunden; bei Indern ist es ein polymorpher Marker, wogegen es bei Chinesen monomorph ist. AMY_2 war nur bei Indern polymorph. Die $UMPK^1$-Frequenzen von Malayen, Chinesen und Indern waren 0,851, 0,880 und 0,942. Die PGP^1-Frequenzen waren 0,991, 1,000, 0,962 und die AMY_2^1-Frequenzen waren 1,000, 1,000 und 0,983.

Résumé. Trois marqueurs génétiques, la UMPK érythrocytaire, la PGP et la AMY_2 sérique, ont été étudiés chez des Malais d'ascendance malaise, chinoise et indienne par électrophorèse sur gel d'amidon et sur gel d'agarose. La UMPK a été trouvée polymorphique dans les trois races. Des variantes ont été observées pour la PGP chez les Malais; chez les Indiens elle est un marqueur polymorphique tandis qu'elle est monomorphique chez les Chinois. La AMY_2 était polymorphique chez les seuls Indiens. Les fréquences de $UMPK^1$ chez les Malais, Chinois et Indiens sont respectivement de 0,851, 0,880 et 09,942. Les fréquences de PGP^1 sont de 0,991, 1,000 et 0,962 et les fréquences de AMY_2^1 de 1,000, 1,000 et 0,983.