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PS 348

STUDY OF THE COMMON SOUTHERN CHINESE GLUCOSE-6-PHOSPHATE DEHYDROGENASE MUTATIONS IN SINGAPORE AND TAIWAN

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Glucose-6-phosphate dehydrogenase (G6PD) deficiency is a very common X-linked genetic disorder in Southeast Asia. Recently, the mutations of nucleotide 1388(G->A), 1376(G->T), 1024(C->T), 493(A->G), 487(G->A), and 95(A-> G) have been reported to account for approximate 80~90% of G6PD mutant alleles in Taiwan. Because the majority of the Singapore populations are southern Chinese , we were interested in finding out the incidence of those common southern Chinese G6PD mutations in Singapore.

The peripheral blood of 127 male G6PD deficient patients, 100 Chinese, 25 Malay, and 2 Indian, were collected and immediately spotted on filter paper (Toyo Roshi Kaisha Ltd., Tokyo) at Singapore General Hospital. The blood samples were air-dried and mailed to Veterans General Hospital-Taipei for analysis. For comparison, 166 dried blood neonatal screening samples of confirmed male G6PD deficient patient in Taiwan were collected and tested. A non-radioactive method by analyzing the restriction fragments of DNA amplified directly from dried blood spot by polymerase chain reaction was used to detect the six common southern Chinese G6PD mutations. We found that:

		1388	1376	1024	493	487	95	Unknown
Singapore Chinese								
	Hokkien	8	20	1	0	0	1	4
	Cantonese	14	18	2	0	0	7	5
	Hakka	1	6	1	0	0	1	0
	other	3	6	0	0	0	0	2
	total (n=100)	26(26%)	50(50%)	4(4%)	0	0	9(9%)	11(11%)
	Malay (n=25)	1	0	1	0	0	0	23
	Indian (n=2)	0	0	0	0	0	0	2
Taiwan	Hokkien	20	69	7	10	1	12	18
	Cantonese	1	0	0	0	0	0	1
	Hakka	4	6	1	2	0	3	6
	other	1	0	1	1	0	1	1
	total (n=166)	26(16%)	75(45%)	9(5%)	13(8%)	1	16(10%)	26(16%)

The incidence of 1376, 1024, and 95 mutations in these two Chinese population were similar. There were apparent difference between the incidence of 1388 in Singapore and in Taiwan which may be caused by the higher proportion of Cantonese, who have higher incident rate of 1388, in Singaporan population. No case with 487 or 493 were found in Singapore, but 7.8% of the G6PD deficiency in Taiwan were caused by 493 mutation. None of those common Chinese G6PD mutations were found in the Malay and Indian assayed except two cases (1388 & 1024) who were found to have maternal Chinese origin.