GENETIC SUBSTRUCTURE AT THE STR LOCI HUMTH01 AND HUMVWA IN HAN POPULATIONS, CHINA

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Systems and loci: HUMTH01 (11p15-p15.5) and HUMVWA (12p12-pter)
Populations and sample size: Han populations living in 3 different geographic areas in China, Guangzhou(S China) N: 101, Chengdu(SW China) N:121 and Changchun(N China) N: 92.

Methods:
Primers for HUMTH01 and HUMVWA (Kimpton et al 1993).
PCR amplification conditions: Hou et al (1994a).
Electrophoresic methods: native polyacrylamide gels with discontinuous buffer system and silver stain (Hou et al 1994a).Typing for HUMTH01 and HUMVWA was performed by comparison with sequenced allele ladders according to the published ISFH guidelines(1994)

Results:
Table 1. Distribution of HUMTH01 genotypes in three Han populations of China

	Observed				Observed		
Genotype	Guangzhou	Chengdu	Changchun	Genotype	Guangzhou	Chengdu	Changchun
6-6	5	2	-	7-10	-	2	1
6-7	2	9	3	8-9	9	7	5
6-8	-	1	1	8-9.3	-	-	2
6-9	12	15	7	8-10	1	2	-
6-9.3	4	-	-	9-9	23	26	29
6-10	2	3	-	9-9.3	3	4	3
7-7	7	7	5	9-10	3	5	3
7-8	2	2	4	9.3-9.3	1	-	-
7-9	26	32	26	9.3-10	-	1	-
7-9.3	1	3	3				

Table 2. Distribution of allele frequencies for HUMTH01

	Frequency				Frequency		
Allele	Guangzhou	Chengdu	Changchun	Allele	Guangzhou	Chengdu	Changchun
6	0.149	0.132	0.060	9	0.490	0.475	0.554
7	0.223	0.256	0.255	9.3	0.049	0.033	0.044
8	0.059	0.050	0.065	10	0.030	0.054	0.022

Test for Hardy-Weinberg Equilibrium (Hou et al 1994b): each population P>0.05

Table 3. Distribution of HUMVWA genotypes in three Han populations of China

	Observed				Observed		
Genotype	Guangzhou	Chengdu	Changchun	Genotype	Guangzhou	Chengdu	Changchun
13-18	-	1	-	16-16	1	1	3
14-14	5	6	5	16-17	6	11	14
14-15	5	2	4	16-18	10	8	5
14-16	8	11	9	16-19	3	4	3
14-17	15	18	9	16-20	1	1	-
14-18	11	7	6	17-17	6	12	3
14-19	3	5	4	17-18	8	13	7
14-20	1	1	-	17-19	5	6	1
15-15	1	-	-	17-20	2	1	2
15-16	-	1	4	18-18	2	4	2
15-17	1	-	4	18-19	7	7	4
15-18	-	-	1	19-19	-	1	1
15-19	-	-	1				

Table 4. Distribution of allele frequencies for HUMVWA

	Frequency				Frequency		
Allele	Guangzhou	Chengdu	Changchun	Allele	Guangzhou	Chengdu	Changchun
13	-	0.004	-	17	0.243	0.302	0.234
14	0.262	0.232	0.228	18	0.198	0.182	0.147
15	0.040	0.012	0.076	19	0.089	0.099	0.081
16	0.148	0.157	0.223	20	0.020	0.012	0.011

Test for Hardy-Weinberg Equilibrium(Hou et al 1994b): each population P>0.05

Comments:

In order to assess the genetic substructure in Chinese populations and analyse human evolution on the basis of DNA sequences, three populations of the Chinese Han nationality from northern and southern as well as from southwestern China, respectively, were directly sampled. The distributions of genotype and allele frequencies at two STR loci HUMTH01 and HUMVWA in these population samples are shown in tables 1-4. The most common allele at the HUMTH01 locus in the Chinese Han population is allele 9, while at the HUMVWA locus the most frequently observed alleles are 14 and 17. No evidence of deviation from Hardy-Weinberg equilibrium was observed in these population samples using the modified statistical approach (Hou et al 1994b). In search of the relationship between the Chinese Han nationality and major racial groups, a phylogenetic tree of 11 populations was constructed on the basis of allele frequencies at the loci HUMTH01 and HUMVWA (figure 1). As shown in figure 1, at the level of a genetic distance of 0.136, the 11 populations were clustered in three major groups, corresponding to Mongoloids, Negroids and Caucasoids. At the level of a genetic distance of 0.023, the Chinese Han nationality was divided into two subgroups, north and south. The comparison of the data showed that the distribution of the frequency of allele 6 at HUMTH01 locus between northern Han and southern Han was significantly different (Changchun:Guangzhou, $x^2=7.36$, df=2, P<0.05. Changchun: Chengdu, $x^2=6.15$, df=2,

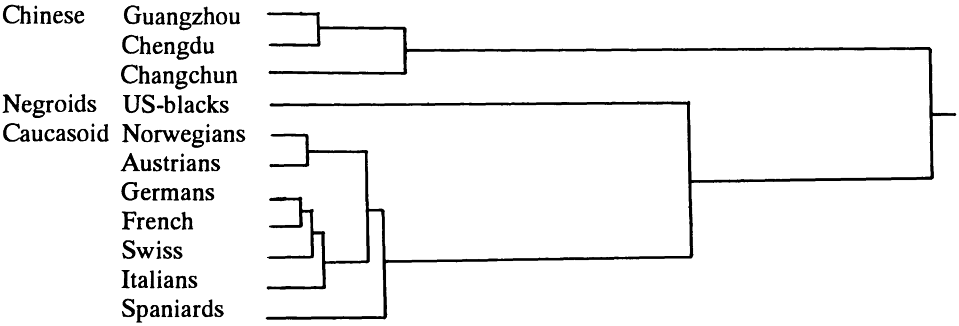

Fig 1. Phylogenetic tree of 11 populations

P<0.05), but when x^2 and P values were calculated using a 2-way R*C contingency table, there were no statistical difference in the distributions of allele frequencies for the loci HUMTH01 and HUMVWA within the Chinese Han population samples. In contrast, the ethnic distributions of allele frequencies for the two loci between the compared populations were always significantly different. The implication of these results is that the greatest genetic differences of allele frequencies for the loci HUMTH01 and HUMVWA appear between major racial groups and that there is some substructure in the Chinese Han population at HUMTH01 locus, but the statistical effect of such subdivision on the match probability may be minor. More effort should be put into further clarification of allele frequency distribution for various Chinese nationalities in order to evaluate the necessity to include a ceiling principle approach for forensic application of the two STR loci.

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