

## Supplementary data:

### Analysis of single nucleotide polymorphisms of *PRNP* gene in twenty-four ethnic groups of India

Mainak Sengupta, Amrita Chakraborty, Indian Genome Variation Consortium and Kunal Ray

*J. Genet.* **89**, 247–251

**Table 1.** Allele and genotype frequency of intergenic SNP rs2756271.

Populations ( <i>n</i> )	Allele frequency (T)	Genotype frequency			HW <i>P</i> values	Standard errors of the allele frequency estimates
		TT	TC	CC		
IE-E-IP1 (22)	0.43	0.23	0.41	0.36	0.419	0.074
IE-E-LP2 (21)	0.57	0.33	0.48	0.19	1	0.076
IE-E-LP4 (21)	0.38	0.14	0.48	0.38	1	0.074
IE-NE-IP1 (21)	0.55	0.33	0.43	0.24	0.66	0.076
IE-NE-LP1 (22)	0.52	0.27	0.50	0.23	1	0.075
IE-N-IP2 (20)	0.48	0.15	0.65	0.20	0.367	0.078
IE-N-LP1 (19)	0.39	0.26	0.26	0.47	0.06	0.079
IE-N-LP5 (23)	0.57	0.35	0.43	0.22	0.674	0.072
IE-N-LP9 (22)	0.52	0.23	0.59	0.18	0.668	0.075
IE-N-SP4 (21)	0.40	0.10	0.62	0.29	0.367	0.075
IE-W-LP1 (23)	0.52	0.30	0.43	0.26	0.676	0.073
IE-W-LP2 (23)	0.50	0.22	0.57	0.22	0.689	0.073
IE-W-LP3 (23)	0.35	0.09	0.52	0.39	0.662	0.07
IE-W-LP4 (23)	0.57	0.30	0.52	0.17	1	0.072
DR-C-IP2 (19)	0.61	0.32	0.58	0.11	0.633	0.079
DR-S-IP4 (21)	0.36	0.10	0.52	0.38	0.664	0.074
DR-S-LP2 (21)	0.60	0.29	0.62	0.10	0.367	0.076
DR-S-LP3 (22)	0.43	0.23	0.41	0.36	0.418	0.074
AA-C-IP5 (15)	0.33	0.07	0.33	0.60	1	0.077
AA-E-IP3 (21)	0.55	0.29	0.52	0.19	1	0.077
TB-NE-LP1 (20)	0.68	0.55	0.25	0.20	0.113	0.074
TB-N-IP1 (23)	0.61	0.39	0.43	0.17	0.675	0.072
TB-N-SP1 (19)	0.50	0.32	0.37	0.32	0.356	0.081
OG-W-IP (22)	0.59	0.45	0.27	0.27	0.071	0.074
ASW (53)	0.27	0.075	0.396	0.528	1	0.043
CEU (113)	0.38	0.124	0.513	0.363	0.426	0.032
CHB (84)	0.55	0.298	0.500	0.202	1	0.038
CHD (85)	0.66	0.447	0.424	0.129	0.630	0.036
GIH (88)	0.49	0.227	0.511	0.261	1	0.038
JPT (86)	0.61	0.407	0.407	0.186	0.179	0.037
LWK (90)	0.38	0.144	0.467	0.389	1	0.036
MEX (50)	0.28	0.1	0.360	0.540	0.485	0.045
MKK (143)	0.49	0.287	0.392	0.322	0.011*	0.03
TSI (88)	0.45	0.227	0.455	0.318	0.518	0.036
YRI (113)	0.34	0.088	0.496	0.416	0.296	0.031

*n*, Number of individuals where genotyping was successful for this SNP. Originally each Indian ethnic consisted of 23 individuals. The frequency of the allele majorly represented (i.e., major allele) in maximum Indian population is furnished. \*, The HapMap population MKK is not in HWE for this SNP.

**Table 2.** Allele and genotype frequencies of intronic SNP rs6116471.

Populations ( <i>n</i> )	Allele frequency (A)	Genotype frequency			HW <i>P</i> values	Standard errors of the allele frequency estimates
		AA	AG	GG		
IE-E-IP1 (22)	0.80	0.64	0.32	0.05	1	0.06
IE-E-LP2 (19)	0.76	0.58	0.37	0.05	1	0.069
IE-E-LP4 (22)	0.80	0.64	0.32	0.04	1	0.06
IE-NE-IP1 (23)	0.80	0.61	0.39	0.00	0.544	0.059
IE-NE-LP1 (23)	0.83	0.65	0.35	0.00	1	0.055
IE-N-IP2 (23)	0.78	0.61	0.35	0.04	1	0.061
IE-N-LP1 (21)	0.81	0.71	0.19	0.10	0.117	0.06
IE-N-LP5 (23)	0.78	0.65	0.26	0.09	0.25	0.061
IE-N-LP9 (21)	0.76	0.57	0.38	0.05	1	0.066
IE-N-SP4 (23)	0.67	0.43	0.48	0.09	1	0.069
IE-W-LP1 (20)	0.85	0.75	0.20	0.05	0.352	0.056
IE-W-LP2 (21)	0.86	0.71	0.29	0.00	1	0.053
IE-W-LP3 (15)	0.70	0.47	0.47	0.07	1	0.084
IE-W-LP4 (22)	0.77	0.55	0.45	0.00	0.538	0.063
DR-C-IP2 (13)	0.96	0.92	0.08	0.00	1	0.038
DR-S-IP4 (20)	0.77	0.65	0.25	0.10	0.213	0.067
DR-S-LP2 (15)	0.90	0.80	0.20	0.00	1	0.055
DR-S-LP3 (18)	0.81	0.61	0.39	0.00	1	0.065
AA-C-IP5 (9)	0.72	0.56	0.33	0.11	1	0.105
AA-E-IP3 (15)	0.67	0.47	0.40	0.13	1	0.086
TB-NE-LP1 (23)	0.87	0.78	0.17	0.04	0.31	0.05
TB-N-IP1 (22)	0.82	0.77	0.09	0.14	0.006*	0.058
TB-N-SP1 (17)	0.82	0.70	0.24	0.06	0.411	0.066
OG-W-IP (20)	0.92	0.90	0.05	0.05	0.077	0.043
ASW (53)	0.528	0.302	0.453	0.245	0.581	0.048
CEU (113)	0.699	0.513	0.372	0.115	0.261	0.03
CHB (84)	0.94	0.881	0.119	0.00	1	0.018
CHD (85)	0.982	0.965	0.035	0.00	1	0.01
GIH (88)	0.733	0.534	0.398	0.068	1	0.033
JPT (86)	0.948	0.895	0.105	0.00	1	0.017
LWK (90)	0.672	0.433	0.478	0.089	0.483	0.034
MEX (50)	0.690	0.480	0.420	0.1	1	0.046
MKK (143)	0.633	0.469	0.329	0.203	0.0005*	0.029
TSI (88)	0.682	0.455	0.455	0.091	0.807	0.035
YRI (113)	0.646	0.398	0.496	0.106	0.419	0.031

*n*, Number of individuals where genotyping was successful for this SNP. Originally each Indian ethnic group consisted of 23 individuals. The frequency of the allele majorly represented (i.e., major allele) in maximum Indian populations is furnished. \*, TB-N-IP1 from India and the HapMap population MKK are not in HWE for this SNP.

**Table 3.** Allele and genotype frequencies of intronic SNP rs116474.

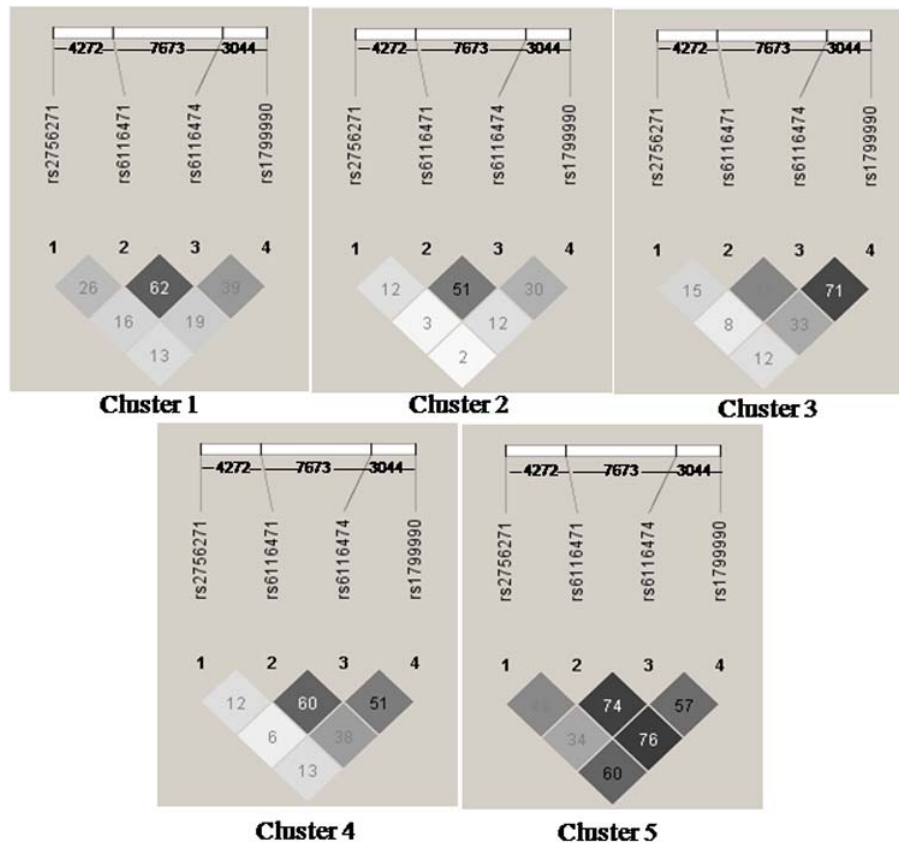
Populations ( <i>n</i> )	Allele frequency (T)	Genotype frequency			HW <i>P</i> values	Standard errors of the allele frequency estimates
		TT	TC	CC		
IE-E-IP1 (23)	0.91	0.87	0.09	0.04	0.132	0.042
IE-E-LP2 (20)	0.85	0.70	0.30	0.00	1	0.056
IE-E-LP4 (23)	0.91	0.83	0.17	0.00	1	0.042
IE-NE-IP1 (21)	0.86	0.71	0.29	0.00	1	0.053
IE-NE-LP1 (22)	0.84	0.68	0.32	0.00	1	0.055
IE-N-IP2 (21)	0.90	0.81	0.19	0.00	1	0.046
IE-N-LP1 (17)	0.79	0.71	0.18	0.11	0.096	0.069
IE-N-LP5 (21)	0.86	0.76	0.19	0.05	0.338	0.053
IE-N-LP9 (13)	0.88	0.77	0.23	0.00	1	0.063
IE-N-SP4 (21)	0.79	0.62	0.33	0.05	1	0.062
IE-W-LP1 (20)	0.90	0.80	0.20	0.00	1	0.047
IE-W-LP2 (21)	0.90	0.81	0.19	0.00	1	0.046
IE-W-LP3 (17)	0.82	0.71	0.24	0.06	0.411	0.065
IE-W-LP4 (19)	0.82	0.63	0.37	0.00	1	0.062
DR-C-IP2 (11)	0.86	0.73	0.27	0.00	1	0.073
DR-S-IP4 (22)	0.82	0.73	0.18	0.09	0.107	0.057
DR-S-LP2 (19)	0.87	0.74	0.26	0.00	1	0.055
DR-S-LP3 (17)	0.85	0.71	0.29	0.00	1	0.061
AA-C-IP5 (5)	0.90	0.80	0.20	0.00	1	0.095
AA-E-IP3 (19)	0.63	0.42	0.42	0.16	0.648	0.078
TB-NE-LP1 (20)	0.90	0.85	0.10	0.05	0.152	0.047
TB-N-IP1 (18)	0.89	0.83	0.11	0.06	0.169	0.052
TB-N-SP1 (16)	0.81	0.69	0.25	0.06	0.434	0.069
OG-W-IP (20)	0.92	0.85	0.15	0.00	1	0.043
CEU (59)	0.746	0.610	0.271	0.119	0.037*	0.04
CHB (45)	0.967	0.933	0.067	0.00	1	0.019
JPT (45)	0.989	0.978	0.022	0.00	1	0.01
YRI (60)	0.808	0.650	0.317	0.033	1	0.036

*n*, Number of individuals where genotyping was successful for this SNP. Originally each Indian ethnic group consisted of 23 individuals. The frequency of the allele majorly represented (i.e. major allele) in maximum Indian populations is furnished. \*, The HapMap population CEU is not in HWE for this SNP.

**Table 4.** Haplotype frequencies of four *PRNP* SNPs in 24 ethnic groups of India.

Ethnic groups	Haplotypes											
	TGTA	CGTA	CGTG	CACG	CATA	CATG	CACA	TACG	TGTG	TATA	CGCG	CGCA
IE-E-IP1	0.452	0.262	0.095	0.095	0.095							
IE-E-LP2	0.632	0.135	0.025	0.067	0.132				0.035			
IE-E-LP4	0.357	0.357		0.071	0.143	0.071						
IE-NE-IP1	0.517	0.283	0.05	0.092	0.025		0.033					
IE-NE-LP1	0.523	0.273	0.045	0.159	0.088			0.01	0.062			
IE-N-IP2	0.403	0.322	0.04	0.075	0.088							
IE-N-LP1	0.469	0.184	0.129	0.184		0.035						
IE-N-LP5	0.487	0.251	0.048	0.034		0.024		0.085				
IE-N-LP9	0.334	0.25	0.042	0.042	0.083	0.042		0.25				
IE-N-SP4	0.403	0.287	0.023	0.214	0.047	0.024						
IE-W-LP1	0.559	0.353	0.029	0.029	0.059							
IE-W-LP2	0.5	0.322	0.047	0.047	0.078				0.028			
IE-W-LP3	0.417	0.25	0.083	0.083	0.167							
IE-W-LP4	0.544		0.095	0.167				0.067				
DR-C-IP2	0.667	0.167	0.083		0.083							
DR-S-IP4	0.323	0.427		0.132	0.081							
DR-S-LP2	0.692	0.192		0.077	0.038			0.035				
DR-S-LP3	0.267	0.4	0.133						0.067			
AA-C-IP5	0.167	0.222	0.222	0.056		0.222	0.056					0.056
AA-E-IP3	0.538	0.058	0.058	0.308	0.019	0.019						
TB-NE-LP1	0.596	0.22	0.026	0.044	0.053			0.062				
TB-N-IP1	0.444	0.087	0.101		0.062	0.031	0.094		0.181			
TB-N-SP1	0.611	0.167	0.056	0.056	0.056			0.056				
OG-W-IP	0.647	0.294		0.029							0.029	
Cluster 1	0.4	0.245	0.058	0.145	0.074		0.018		0.059			
Cluster 2	0.506	0.224	0.051	0.052	0.063		0.002	0.03	0.04			
Cluster 3	0.501	0.293	0.034	0.086	0.07							
Cluster 4	0.514	0.249	0.053	0.088	0.034	0.016		0.022	0.019			
Cluster 5	0.469	0.125	0.06	0.281		0.063						

SNPs used for construction of haplotypes (in the following order): rs2756271(C/T); rs6116471 (A/G); rs6116474 (T/C); rs1799990 (A/G) respectively. For clusters refer to Indian Genome Consortium (2008).



**Figure 1.** Estimate of pair-wise linkage disequilibrium (LD) between the selected PRNP SNPs in the five population ‘clusters’ as described previously by Indian Genome Variation Consortium (2008). There is no LD between any of the SNPs ( $r^2 < 80$ ) in any of the clusters. The distance between each SNP (in bp) is shown on the top of the LD blocks in each panel.