

Table S9 Top 10 genomic regions harboring extreme PBS signal in the Ainu in comparison to CHB.

Region	Top SNP	Anc ¹	Der ²	Derived allele frequency		SNP location	GWAS catalogue ⁴ (based on the name of gene)	Empirical <i>P</i> -value ⁵			
				EA ³	Ainu			$N_e=2,219$		$N_e=1,000$	
								$T=800$	$T=1,000$	$T=800$	$T=1,000$
Chr1: 54467753-54967752	rs3753405	T	C	1.1%	54.2%	<i>SSBP3</i>	None	0.004	0.010	0.066	0.112
Chr2: 133465703-133765702	rs16841046	T	C	15.0%	87.5%	<i>NCKAP5</i>	Hypersomnia, glaucoma, and height	0.000	0.001	0.015	0.031
Chr3: 77969244-78719243	rs4680999	G	T	1.0%	70.8%	Upstream of <i>ROBO1</i>	Aspartate transaminase in liver, brain activity in schizophrenia patients	0.000	0.002	0.020	0.046
Chr3: 95219244-96119243	rs9826768	G	A	0.1%	70.8%	Upstream of <i>EPHA6</i>	Blood trace element (Cu and Zn levels), serum free IGF-1 level (obesity related)	0.000	0.001	0.017	0.040
Chr4: 86718911-87218910	rs345326	G	A	10.1%	75.0%	<i>ARHGAP24</i>	PR interval in electrocardiogram	0.002	0.004	0.028	0.050
Chr10: 1604427-2054426	rs17156778	G	A	12.2%	75.0%	<i>ADARB2</i>	Radiation response in LCL, Emphysema, BMI, % body fat	0.002	0.005	0.036	0.063
Chr12: 341619-1091618	rs2305164	C	T	2.8%	83.3%	<i>SLC6A13</i>	Serum metabolite level (pyroglutamine, deoxycarnitine, betaine,), glomerular filtration rate	0.000	0.000	0.008	0.020
Chr12: 52991619-53341618	rs17116681	G	A	18.4%	91.7%	<i>KRT72</i>	None	0.001	0.001	0.013	0.023
Chr15: 53571673-54121672	rs518263	T	C	1.6%	62.5%	<i>WDR72</i>	Blood urea nitrogen level, serum creatinine level, glomerular filtration rate, longevity, cognitive function	0.001	0.005	0.038	0.076
Chr20: 43961795-44611794	rs6032336	A	G	95.4%	20.8%	<i>WFDC8</i>	None	0.000	0.000	0.012	0.030

¹ Ancestral allele; ² Derived allele; ³ 1KG phase 3 East Asians (excluding JPT); ⁴ Last date accessed to the database (<https://www.genome.gov/26525384>) is April 8th, 2015; ⁵ Empirical *p*-values were calculated as the proportion of simulations with their simulated frequency of the derived allele equal to or greater than the observed frequency in Ainu.