

Polymorphism rs1800795 of the *IL6* gene in samples of Siberian indigenous ethnic groups

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Motivation and Aim: Investigation of the frequencies of functionally significant gene variants in the context of medical biology and gene geography is a relevant issue in studying of the population genetic structure of indigenous Siberian peoples. Genetic polymorphism of the interleukin *IL6* gene is of great importance for understanding the pathogenesis of cardiovascular, bronchopulmonary, oncological, autoimmune, non-infectious and infectious diseases.

Methods and Algorithms: The study was performed on the monoethnic samples of Buryats, Teleuts, Dolgans and Yakuts. Real-time PCR was used to determine the alleles of the single nucleotide polymorphism C-174G, *rs1800795*, in gene *IL6*.

Results: The highest frequency of the *-174G* allele was recorded in Yakuts (97.3%), slightly less in Buryats, followed by Dolgans and Teleuts (87.1%) (Table 1). The differences between Teleuts and Yakut populations are statistically significant (Table 2). The results obtained were compared to frequencies identified for Russians from Eastern Siberia (57.4%) and the values in other Caucasian groups (54-65%) described in literature [1]. The studied samples of the indigenous populations showed a significantly higher occurrence of the *-174G* allele compared to Caucasians. At the same time, they were significantly lower than in several East Asian populations described in literature (Chinese, Japanese and Vietnamese). Those Asian populations have this value close to 100%.

Table 1. Genotype distribution for *C-174G*, *rs1800795* of gene *IL6* in samples indigenous Siberian peoples and Russians

Population		Buryats	Teleuts	Yakuts	Doligans	Russians
Genotype distribution, <i>n</i> (%)	<i>G/G</i>	241 (88.3)	87 (75.0)	88 (94.6)	143 (82.6)	54 (34.8)
	<i>G/C</i>	30 (11.0)	28 (24.1)	5 (5.4)	29 (16.8)	70 (45.2)
	<i>C/C</i>	2 (0.7)	1 (0.9)	0	1 (0.6)	31 (20.0)
<i>N</i> , ppl		273	116	93	173	155
<i>p</i> (H-W)		0.861	0.839	0.975	0.937	0.621
<i>-174G</i> frequency, %		93.8	87.1	97.3	91	57.4

Note. *N* is the sample size; *n* is the quantity; *p* (H–W) is the probability of Hardy–Weinberg equilibrium deviation.

Table 2. *IL6 -174G* allele frequency in some populations (ethnic groups) and comparison of populations (p-value)

Population/ ethnic group	N, ppl	<i>IL6 -174G</i> frequency, %	Population comparison (p-value)				
			Buryats	Teleuts	Yakuts	Dolgans	Russians
Buryats*	273	93.8		0.003	0.099	0.151	<i>p</i> < 0.001
Teleuts*	116	87.1	0.003		<i>p</i> < 0.001	0.175	<i>p</i> < 0.001
Yakuts*	93	97.3	0.099	<i>p</i> < 0.001		0.010	<i>p</i> < 0.001
Dolgans*	173	91	0.151	0.175	0.010		<i>p</i> < 0.001
Russians *	155	57.4	<i>p</i> < 0.001	<i>p</i> < 0.001	<i>p</i> < 0.001	<i>p</i> < 0.001	
Chinese Dai in Xishuangbanna, China**	93	100	0.001	<i>p</i> < 0.001	0.071	<i>p</i> < 0.001	<i>p</i> < 0.001
Japanese, Tokyo, Japan**	104	100	<i>p</i> < 0.001	<i>p</i> < 0.001	0.053	<i>p</i> < 0.001	<i>p</i> < 0.001
Kinh (Viet), Ho Chi Minh City, Vietnam**	99	99.5	0.002	<i>p</i> < 0.001	0.185	<i>p</i> < 0.001	<i>p</i> < 0.001
Finns, Finland**	99	54.5	<i>p</i> < 0.001	<i>p</i> < 0.001	<i>p</i> < 0.001	<i>p</i> < 0.001	0.740
English people and Scots**	91	58.8	<i>p</i> < 0.001	<i>p</i> < 0.001	<i>p</i> < 0.001	<i>p</i> < 0.001	0.701
Toscani, Italy**	107	64.5	<i>p</i> < 0.001	<i>p</i> < 0.001	<i>p</i> < 0.001	<i>p</i> < 0.001	0.094

Note. * marks the data obtained by the authors, ** marks the data from the literature (The 1000 Genomes..., 2012); $p < 0.025$, at which differences were considered statistically significant are marked in bold.

Conclusion: Thus, an increase in the frequency of the *-174G* variant of the *IL6* gene in human populations is shown when moving along the Eurasian map from west to east. This in-between position of indigenous Siberian populations, as exemplified by Buryats and Teleuts, had been demonstrated earlier in the polymorphism frequencies of some other genes [2]. Carriage of the *-174G* variant was shown to be associated with a high level of *IL6* expression and, as a consequence, with an enhanced inflammatory response to the pathogen encounter. The increased concentration of this allele in Siberian populations compared to Caucasian populations may indicate a selection pressure with increasing pathogen load due to environmental features.

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References

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