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Ficoline-3 and MASP-2 gene variants in Russian Arctic populations

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Motivation and Aim: Ficolin-3 and MASP-2 are among the key participants in the lectin pathway of complement activation. Ficolins are pattern recognition receptors capable of binding to MASP and activating complement via the lectin pathway. Ficolin-3 is the most powerful activator of the lectin pathway, with its serum concentrations being significantly higher than MBL level. The *FCN3* rs28357092 mutation is associated with low ficolin-3 levels in plasma. MASP-2 is a protease that turns out to be complexed with MBL and ficolins, leading to complement activation. Mutation in *MASP2* rs72550870 is associated with impaired protein binding to lectins, and therefore complement activation does not occur. The purpose of the study was to reveal ethnic differences in the distribution of allelic gene variants for the lectin pathway components of complement activation between the indigenous populations of the Arctic territory of Siberia and Caucasoids.
Methods and Algorithms: Newborn children of indigenous Arctic ethnic groups (Nenets, Dolgan-Nganasans, mixed population) (n = 678) and Russians (n = 302) were included in the study. *FCN3* rs28357092 and *MASP2* rs72550870 genotyping was carried out by RT-PCR. Allele and genotype frequencies were compared using an online calculator, the χ^2 test.
Results: As the analysis result of the *FCN3* rs28357092 and *MASP2* rs72550870 genotype frequency, the prevalence of GG and AA homozygotes respectively, in all the studied populations according to the world data has been shown. The heterozygous genotype G/del rs28357092 was found to take place in only one Russian child. The heterozygous AG rs72550870 genotype occurs occasionally in the Arctic populations compared with Russians, i.e. the frequency of the AG genotype in Russians was 6.6 %, in Nenets – 0.3 %, in Dolgan-Nganasans – 1.4 %, and in the mixed population – 1.8 %. None of the groups were found to be homozygous for the minor allele G (Table 1). *Conclusion*: The genetic analysis results showed lower prevalence of genetic markers of ficolin-3 and MASP-2 deficiency in the indigenous populations of the Arctic territories compared to Russians.
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Table 1. The *FCN3* and *MASP2* genotype frequency in newborns of different ethnic populations, *n* (%)

Genotype /allele	Nenets, n = 323 (1)	Dolgan-Nganasans, *n* = 138 (2)	Mixed population, *n* = 217 (3)	Caucasoids, *n* = 302 (4)	*p*
FCN3 rs28357092					
GG	292 (100.0)	128 (99.2)	199 (98.0)	291 (96.4)	1/3 = 0.02 1/4 < 0.001
G/del	0 (0.0)	0 (0.0)	0 (0.0)	1 (0.3)	–
del/del	0 (0.0)	1 (0.8)	4 (2.0)	10 (3.3)	1/3 = 0.02 1/4 = 0.002
del*	0 (0.0)	2 (0.8)	8 (2.0)	21 (3.5)	1/3 < 0.001 1/4 < 0.001 2/3 = 0.02 2/4 = 0.003
MASP2 rs72550870					
AA	322 (99.7)	136 (98.6)	213 (98.2)	226 (93.4)	1/4 < 0.001 2/4 = 0.02 3/4 = 0.01
AG	1 (0.3)	2 (1.4)	4 (1.8)	16 (6.6)	1/4 < 0.001 2/4 = 0.02 3/4 = 0.01
GG	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	-
G*	1 (0.2)	2 (0.7)	4 (0.9)	16 (3.3)	1/4 < 0.001 2/4 = 0.03 3/4 = 0.01