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ALA54THR POLYMORPHISM OF THE *FABP2* GENE AND METABOLIC SYNDROME IN THE YAKUT POPULATION

ABSTRACT
The metabolic syndrome represents complex of the metabolic risk factors connected with increase in prevalence of diabetes and cardiovascular diseases. Each component of metabolic syndrome to some extent has communication with heredity that demonstrates that genetic factors can have significant effect on pathogenesis of this syndrome. In researches among some populations the association of polymorphism Ala54Thr of gene of *FABP2* (rs1799883) with metabolic violations was shown. We analyzed association of allelic rs1799883 options (*FABP2*) with metabolic syndrome and its components at representatives of the Yakut ethnic group (on self-determination) from Berdigestyakh village of the Gorniy region of Republic Sakha (Yakutia). All surveyed people the written informed consent to participation in research. The program of research included screening by uniform technique on detection metabolic risk factors of chronic noncommunicable diseases. Polymerase chain reaction in real time was carried out in the CFX96 system of production Bio-Rad. Tests and primers were developed by means of the Beacon Designer 8 program from PREMIER Biosoft. From 228 participants of 42% had the increased level of blood pressure, 9,7% — the reduced level high density lipoproteins, 5,8% — raised triglycerides, 22,4% — fasted hyperglycemia, 56,8% — abdominal obesity, 16,8% — metabolic syndrome by IDF criteria (2005). G polymorphism of Ala54Thr *FABP2* (71%) are characteristic of the vast majority of representatives Yakut population allele carriage. Frequency of GG genotype was 42,1% (95% CI 35,9-48,6%), AG — 57,9% (95% CI 51,4-64,1%). The association of genotype AG with abdominal obesity was established at OR 1,7 (95% CI 1,01-2,99). Taking into account prevalence in this group of the population of such metabolic disturbances as obesity and the increased level of blood pressure, and also the growing incidence of diabetes type 2, it is necessary to continue search of genes of predisposition to these diseases.

Keywords: *FABP2* gene (rs1799883), Ala54Thr polymorphism of the FABP2 gene, metabolic syndrome, abdominal obesity, Yakut population.

INTRODUCTION

The metabolic syndrome is complex of the metabolic risk factors connected with increase in prevalence of diabetes and cardiovascular diseases [8, 12]. Each component of metabolic syndrome to some extent has communication with heredity that demonstrates that genetic factors can have significant effect on pathogenesis of this syndrome [9, 12, 15]. The gene of the protein connecting fatty acids in intestines (*FABP2*) participates in regulation of capture and transfer of long-chain fatty acids [5]. Polymorphic options of gene *FABP2* (rs 1799883) can exert impact on concentration of lipids in blood plasma and their intracellular transport [6]. The result of researches, conducted among northern populations was shown that lipid metabolism plays key role in effective adaptation to conditions of cold climate [1, 3]. The Republic of Sakha (Yakutia) belongs to terri-

tories with extreme climatic conditions, first of all because of low temperatures. Yakuts whose number according to census of 2010 makes 466 492 people are representatives of the Central Asian type of North Asian race. Now among this region population also as well as around the world, incidence of diabetes type 2 promptly grows. It is promoted by the high frequency of obesity and other metabolic disorders [2]. The researches devoted to studying of communication between genetic factors and health of the person can expand limits of our knowledge of influence of genes and the environment on phenotype. In this regard, studying of communication the polymorphism Ala54Thr *FABP2* gene (rs 1799883) with metabolic syndrome and its components, and also with some biochemical and anthropometrical indicators at Yakuts, representatives of the Central Asian type of North Asian race was research objective.

MATERIALS AND METHODS Groups were created during the epidemiological research among the Berdigestyakh village population of the Gorniy region Republic Sakha (Yakutia) conducted within basic unit of the State task Education and Science Ministry Russian Federation 17.6244.2917/8.9 "Clinical and genetical aspects of the diseases characteristic of Yakutia native population in modern conditions". In total 242 persons, representatives of the Yakut ethnic group participated in research (on self-determination). From them consent to genetic research was received at 228 persons. All participants of research were inspected according to the uniform program including double measurement of the blood pressure (BP), anthropometrical inspection by standard technique, the analysis of composition structure of organism on the bioimpedance Tanita analyzer (Japan) SSC 330, intake of blue blood for laboratory researches. Definition of glucose, general cholesterol (OHS), triglycerides, cholesterol of high density lipoproteins carried out on the Cardiochek PA (USA) express analyzer from the venous blood taken in the morning 10–12 hours later after meal. Concentration of cholesterol low density lipoproteins calculated by formula Fridvald at the content of triglycerides in blood less than 4,5 mmol/l. Levels of maintenance of leptin, CRP, insulin determined by the ELISA method with use of DRG sets. Existence of metabolic syndrome (MS) and its components defined by IDF criteria 2005 [8]. Control groups were created from among persons without certain metabolic disturbances.

228 DNA samples were genotyping by the PCR method. For SNP genotypings used TaqMan of test specific to the sites supporting the interesting SNP. Tests and primers were developed by means of the Beacon Designer 8 program from PREMIER Biosoft. As reporters FAM and R6G dyes, and as quencher – BHQ-1 were used. Polymerase chain reaction in real time was carried out in the CFX96 system of production Bio-Rad. The volume of reactionary mix made 25 mcl. Each reaction was carried out in three repetitions. The stage of activation was carried out at 95 °C within 3 minutes, the course of one cycle consisted of three temperature time spans – 95 °C (30 sec.), 54 °C (20 sec.) and 72 °C (20 sec.). Total quantity of cycles made 40.

Inspection of compliance of distribution of genotypes to the law of balance of equilibrium of Hardy-Weinberg was carried out with use of the online calculator on the website https://wpcalc.com/en/equilibrium-hardy-weinberg/ [7]. The statistical analysis of data was carried out in IBM SPSS STATISTICS 22. When comparing groups depending on data type used Mann-Whitney and Pearson's criteria χ^2. For measure of effect we are calculated the odds ratio (OR). Critical value of the statistical significantly was accepted equal 5%.

The research project was approved by local committee on bioethics of the Yakut scientific center of complex medical problems the Russian Academy of Medical Science (extract from the protocol No. 39 of June 26, 2014). Participation in research was completely voluntary. Obtained clinically useful information was available to participants of research.

RESULTS AND DISCUSSION

The proteins binding fatty acids *(FABPs*) concern family cytoplasmatic lipid - the binding proteins participating in intracellular transport and metabolism of lipids. *FABP2* gene expressing in epithelial cells of small intestine is located in chromosomal area 4q28-4q31, consists of 4 exons and 3 introns and codes the protein containing 131 amino acids. Polymorphism rs1799883 FABP2 gene is caused by replacement of guanine by alanine in the 54th codon that brings to replacement of alanine by threonine (Ala54Thr) in exon 2. The protein containing threonine has big affinity to long-chain fatty acids, than alanine - the containing option [5-6]. *FABP2* is considered as the candidate gene involved in pathogenesis of diabetes 2 types, metabolic syndrome and obesity in different ethnic groups [4, 10, 11, 13, 14, 16]. At the same time the analysis of literature showed ambiguity of researches results about communication between polymorphism of FABP2 gene and metabolic disturbances risk development. So, Zhao T. with coauthors in 2010 meta-analysis of results 13 researches with 13451 participants was carried out. Generalization of results showed weak communication of polymorphism of Ala54Thr of *FABP2*gene with insulin resistance degree, high level of fasted insulin and glucose in 2 hours after loading [16]. Qiu C. and coauthors meta-analysis (2014), including results of 13 researches with 2020 cases of diabetes and 2910 healthy people in control group (6 Asian and 7 European populations), showed existence of association between Ala54Thr polymorphism *FABP2* gene and risk of diabetes types 2 only for Asian populations [13]. Results of meta-analysis of Y. Liu and coauthors (2015) which combined results of 39 researches (24 —types 2 diabetes, 9 — obesity, 6 — metabolic syndrome), showed existence of statistically significant communication between polymorphism of Ala54Thr *FABP2* gene and MS (p =0,031), type 2 Diabetes (p <0,001), but did not find association with risk of obesity (p = 0,367) [11].

In present research for the analysis of association allelic rs1799883-*FABP2* variants with metabolic syndrome and its components data of 228 representatives of the Yakut population are used (on self-determination). Average age of participants made 43,9 (17,5) years. Abdominal obesity is established at 56,8% of participants, the increased level BP – at 42%, fasted hyperglycemia – at 22,4%, reduced HDL cholesterol – at 9,7%, raised triglycerides at – 5,8%, and metabolic syndrome by IDF criteria (2005) is revealed at 16,8% of the inspected population.

G polymorphism of Ala54Thr *FABP2* gene (71%) is characteristic of the vast majority of representatives' Yakut population allele carriage. According to literature the frequency of carriage of allele A in the majority of populations also makes about 30% [11]. Among inspected persons with AA genotype are not revealed. Frequency of alleles and genotypes was similar both among men and women, and in different age groups (table 1). Distribution of genotypes in subgroups will be coordinated with the equilibrium Hardy-Weinberg.

Comparative analysis of lipid and carbohydrate metabolism at the inspected persons showed that polymorphism of this gene does not exert noticeable impact on average concentration of the specified indicators (tab. 2). It should be noted that distinctions of average values of the main anthropometrical indicators (body mass index, waist circumference) depending on *FABP2* genotypes, were close, but did not reach level statistically significant that, perhaps, is caused by the small sample size.

The MS was diagnosed for 36 persons that made 16,8% of total number of inspected. Occurred among women of MS considerably more often than at men (20.9 and 7.6% respectively, p =0.016), and its frequency increased inspected with age (from 3.5% at the age of 20-39 years to 28.9% in 60 years and is more senior, p <0.001). The research of Clinical-genetic associations with use of Pearson χ^2 criteria showed that level of significance when comparing frequency of abdominal obesity in groups with different genotypes was near-critical to value (0.045). At the same time chances to have AG genotype by 1.7 times (95% CI 1.01-2.99) were higher at persons with abdominal obesity (table 3). Frequency of MS and other its components did not depend on *FABP2* genotype.

Thus, results of research showed rather high frequency of minor allele of gene of

FABP2 54Thr in Yakut population (0.29). The analysis of clinical-genetic associations showed possible communication of this polymorphism with abdominal obesity. Taking into account prevalence in this group of the population of such metabolic disorders as obesity and the increased level BP, and also the growing incidence of diabetes type 2, it is necessary to continue search of genes of predisposition to these diseases.

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Table 1

Alleles and genotypes of polymorphism ALA54THR FABP2 gene distribution in Yakut population *

Groups	n	Frequency (95% CI)		p
		Alleles		
		G	A	
Women Men	156 72	70,2 (64,9-75) 72,9 (65,1-79,5)	29,8 (25-35,1) 27 (20,5-34,8)	0,629
Both groups	228	71,1 (66,7-75)	28,9 (25-33,2)	0,548
20-39 years 40-59 years 60 years and older	91 88 49	70,9 (63,9-77) 72,2 (65,1-78,3) 69 (60-77,6)	29,1 (23-36,1) 27,8 (21,8-34,9) 30,6 (22,4-40,3)	
Genotypes				
	GG	AG		
Women Men	156 72	40,4 (33-48,2) 45,8 (34,8-57,3)	59,6 (51,8-67) 54,2 (42,7-65,2)	0,439
Both groups	228	42,1 (35,9-48,6)	57,9 (51,4-64,1)	
20-39 years 40-59 years 60 years and older	91 88 49	41,8 (32,2-52,2) 44,3 (34,4-54,7) 38,8 (26,4-52,8)	58,2 (48-67,8) 55,7 (45,3-65,6) 61,2 (47,2-73,6)	0,817

Note. * — Distribution of genotypes in all groups will be coordinated with the Hardy-Weinberg equilibrium; p — Pearson $\chi 2$ criteria statistical significantly.

Table 2

Anthropometrical and biochemical indicators in Ala54Thr *FABP2* genotypes groups

Indicator	Genotype of n (%)				p
	GG		AG		
	n	Me (Q1; Q3)	n	Me (Q1; Q3)	
Age, years	96	45,7 (25,3; 56,8)	132	47 (27; 59)	0,512
SBP, mm Hg	94	113,2 (106,3; 130,0)	132	120,0 (109,8; 132,4)	0,261
DBP, mm Hg	94	76,5 (69,6; 82,8)	132	77,3 (70,0; 86,9)	0,416
Total cholesterol, mmol/l	96	4,4 (3,5; 5,1)	130	4,4 (3,7; 5,5)	0,199
High density lipoprotein cholesterol, mmol/l	96	1,7 (1,4; 2,1)	130	1,7 (1,4; 2,2)	0,348
Triglycerides, mmol/l	96	0,9 (0,8; 1,0)	130	0,9 (0,8; 1,1)	0,295
Low density lipoprotein cholesterol, mmol/l	96	2,1 (1,4; 2,8)	130	2,2 (1,5; 3,1)	0,281
Fasting plasma glucose, mmol/l	94	5,1 (4,6; 5,5)	129	5,1 (4,7; 5,5)	0,953
Leptin, ng/ml	94	4,8 (4,3; 5,2)	130	4,7 (4,3; 5,2)	0,977
Insulin, mu/ml	94	19,5 (13; 27,5)	130	18 (12,6; 27,7)	0,730
CRP, mg/ml	94	0,2 (0,07; 1,9)	130	0,3 (0,07; 1,2)	0,720
BMI, kg/m2	94	22,6 (20,3; 27,5)	127	24,2 (21,8; 27,8)	0,066
Waist circumference, cm	92	83,8 (76,2; 95,4)	128	88,1 (79,8; 97,0)	0,082
Body fat percent	92	24,2 (16,6; 32,6)	126	29,1 (21,4; 34,6)	0,107
Fat free mass, kg	94	43,6 (40; 52,4)	127	43,1 (39,7; 49,6)	0,677

Note: p — statistical significantly Mann-Whitney criteria; Me (Q1; Q3) - median (25-75%).

Table 3

Associations of Ala54Thr *FABP2* gene polymorphism with metabolic risk factors

Risk factor *	Genotypes, n (%)		OR (95% CI)	p
	GG	AG		
Metabolic syndrome, n=214	13 (14.8)	23 (18.3)	1.57 (0.76-3.2)	0.503
Raised blood pressure, n=226	38 (40.4)	57 (43.2)	1.12 (0.65-1.9)	0.679
Reduced HDL cholesterol, n=226	10 (10.4)	12 (9.2)	0.87 (0.36-2.1)	0.766
Raised triglycerides, n=226	5 (5.2)	8 (6.2)	1.19 (0.38-3.8)	0.763
Raised fasting plasma glucose, n=223	22 (23.4)	28 (21.7)	0.91 (0.48-1.71)	0.764
Abdominal obesity, n=220	45 (48.9)	80 (62.5)	1.74 (1.01-2.99)	0.045

Note: * — by IDF criteria 2005; p — Pearson $\chi 2$ criteria statistical significantly

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GENETIC TESTING AND INFORMED CONSENT FOR SPINOCEREBELLAR ATAXIA TYPE I, THE MOST COMMON HEREDITARY DISEASE IN THE YAKUT POPULATION

ABSTRACT

The article discusses the issues of informed consent for DNA testing at type 1 spinocerebellar ataxia, the most common hereditary disease with late onset of manifestation in the Yakut population. Different stages of obtaining informed consent in medical genetic counseling and in scientific research are described. The expediency of using the bioethical principle of non-disclosure of genetic information for a participant in a scientific study on the research of hereditary diseases with late manifestation is established.

Keywords: DNA testing, hereditary diseases, informed consent, type I spinocerebellar ataxia.

INTRODUCTION

Since 70th years of last century the works about cloning of human DNA developed in high gear and accomplished with the successful Human Genome project of the full interpretation of the DNA nucleotide sequence, and it offered great opportunities for development of new fields of science and practice, including molecular genetics, ethnogenomics, molecular medicine etc.

In the world there are about 7000 nosologies of monogenic diseases, which are detected in 3-6% of newborns, and in structure of the child mortality total rate of under-fives it is 10-14% [13]. There is a conditional separation of monogenic diseases on orphan diseases with a frequency of 1:100000 (lysosomal storage disorders, etc.) and common hereditary diseases - 1:10000 (a mucoviscidosis, a phenylketonuria, etc.). Frequency of monogenic diseases in various populations of the world can differ considerably. It depends on evolutionary features of formation of a genetic pool of the people. In some populations, however, this or that mutation which is the reason of monogenic pathology owing to evolutionary and genetic features becomes frequent and can be called "ethno-specific". For example, so-called "Finnish" hereditary diseases, generally autosomal and recessive which frequency of Finns is much higher, than in any other populations are known