Gene polymorphisms determining physical performance in Ukrainian power-oriented events of track and field athletics

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Summary
Research background and hypothesis. Modern research on the molecular genetics of physical activity shows that increased physical performance is determined by a set of specific genes. However, the systematic genetic studies of high performance athletes for selection and training purposes do not exist to our knowledge and there are no studies for specific sports, which could have made the specialization choices easier.

Research aim was to analyse important genetic variations that may underlie differences in the potential to be an elite athlete of power-oriented events of track and field athletics.

Research methods. In total, DNA of 73 athletes involved in power-oriented events of track and field athletics and 283 persons not involved in sports (sedentary control) were examined. In this paper, we consider ten candidate-genes and carry out statistical analysis to determine their most favourable polymorphism for power-oriented events of track and field athletics. Using the polymerase chain reaction method I/D (ACE), R577X (ACTN3), T-786→C (eNOS), Pro/Ala (PPARG), G/C (PPARA), Pro582→Ser (HIF-1α), Ala203Pro (PPARGC1B), C-1306T (MMP2), Ser422Gly (ELN), Tag1A (DRD2) genes polymorphisms were detected in athletes of different abilities.

Research results. The significance of differences between group of power-oriented events of track and field athletics and the control group on the distribution of allele NO-synthase gene polymorphism and distribution of genotype of metalloproteinase 2 gene polymorphism was found. The frequency of T allele T-786→C polymorphism eNOS prevails in the group of athletes by 11.5% (p = 0.02). The frequency of T/T genotype C-1306T (MMP2) is 11.8% (p = 0.003) higher than in the control group. The athletes specializing in different events are characterized by genetic heterogeneity. Favourable alleles for high athletic performance in power-oriented events of athletics were established.

Discussion and conclusions. Sports genetics can be used to design better selection and training processes in order to allow athletes to achieve their full potential. Our study provides evidence for the association between gene polymorphisms and elite power athlete status. However, athletes belonging to the same group of sports differ in the distribution of genotypes and allele of gene that associate with elite performance. Only after detection of favourable genotype combination for the particular sport potential of genetic testing in sport will be increased.

Keywords: sport selection, sports genetics, gene polymorphism, power-oriented events, track and field athletics

Introduction
Modern research on the molecular genetics of physical activity shows that increased physical abilities are determined by a set of specific genes (Ahmetov et al., 2012; Bouchard et al., 1997; Bray et al., 2009). However, the systematic genetic studies of high performance athletes for selection and training purposes do not exist at the moment (Rankinen et al., 2010; Hagberg et al., 2011; Wang et al., 2013).

To our knowledge, there are no studies for specific sports, which could have made the specialisation choices easier. We propose to use sports genetics to design better selection and training processes in order to allow athletes to achieve their full potential.

The efficiency of sports training and results of athletes’ performances largely depend on the selection process. One of the most important factors for effective selection is the knowing and realizing the requirements of the particular sport. Athletic abilities can be forecast only regarding a selected sport or group of sports with similar features of power supply mechanisms. And primarily, we should pay attention to those features which are stable or less variable and determine success in the future sports activity.

In sports, the notion that there are individuals who are more endowed than others in terms of basic skills and traits, indicator of special training and their growth rates has been developed. The criteria by which the sports talent of a child is usually detected are the data of body mass, stature, maturity level, body composition and motor abilities. These criteria are commonly included in talent-detection test batteries, though the most efficient selection
is the one made by the complex of pedagogical, medical and biological, psychological and social criteria (Платонов, 2004; Сергиенко, Лигиевская., 2011; Шинкарук, 2011). This approach is already used in track and field athletics (Захарова, 2003; Бобровник, 2005). However, in Ukraine, there are no research results on the selection based on hereditary, molecular and genetic characteristics. It is believed that such a new area in science as sports genetics will help improve the initial selection for sports trainings, the determination of the reserve abilities of high-class athletes and the selection of athletes for teams. The modern methods of sports genetics will allow to avoid many mistakes in the training process by determining the molecular genetic markers which reflect the hereditary abilities of individual athletes.

The contemporary researchers of the molecular genetics of muscle activity have proved that the physical properties are determined polygenetically, i.e. the hereditary disposition to sports depends on number of genes. Therefore, in order to achieve high athletic results in some sport, a combination of genes is required (Bouchard et al., 1997; Рогозкин и др., 2005; Ahmetov et al., 2012).

Having applied the method of meta-analysis to scientific publications, we managed to identify the candidate genes that most likely influence the results of performance in the power-oriented types of track and field athletics. Taking into account that in these types, the results depend on the contraction velocity of motor units and intramuscular coordination, on the reserve of energy sources (Платонов, 2004), and the fact that the muscular activity is carried out mainly owing to anaerobic ways of resynthesis, we have assumed that the greatest influence on the athletes’ abilities in these sports will be made by the gene polymorphisms that affect the condition of the neuromuscular system (ACE, ACTN3) (Williams et al., 2005; Druzhkevskaya et al, 2008; Berman, North., 2010; Puthuacheary, 2011); metabolism in muscles (PPARG, PPARA, PPARGC1B) (Ahmetov et al., 2006; Arany, 2007; Дроздовская и др., 2012); determine adequate adaptation to hypoxic states (eNOS, HIF1A) (Semenza, 2004; Wolfarth B et al., 2008; Drozdovska et al., 2009; Cięszczyk et al., 2011), condition of the connective tissue (ELN, MMP2) (Hanon, 2001; Price, 2001) and properties of the nervous system (DRD2) (Malyuchenko et al, 2010).

The purpose of the study was to analyse important genetic variations that may underlie differences in the potential to be an elite athlete of power-oriented events of track and field athletics.

**Methods**

For this study, we have examined 73 athletes who were recruited from power-oriented types of track and field athletics (60 males and 13 females; age 18-31) and 283 healthy unrelated Ukrainians without any competitive sport experience (187 males and 96 females; age 18-34). The athletes and control groups were all Caucasians.

The athletes were divided into the following three groups: 18 athletes specializing in athletics throwing (hereinafter referred to as ‘throwing’); 21 athletes specializing in sprint (hereinafter referred to as ‘sprint’); 34 athletes specializing in track and field athletics jumping (hereinafter referred to as ‘jumping’). There were 47 elite athletes, 26 sub-elite athletes.

The molecular and genetic analysis was performed in the laboratory of the Department of General and Molecular Pathophysiology, O.O. Bohomolets Institute of Physiology, National Academy of Sciences of Ukraine). The DNA was extracted from the buccal epithelium using a set of reagents Diatom™ DNAPrep (Biokom, Russia). The method of polymerase chain reaction (PCR) followed by a restriction analysis was used to determine the following polymorphisms: I/D gene polymorphism of angiotensin-converting enzyme (ACE), R577X (C/T) gene polymorphism of α-actinin-3 (ACTN3), T→C polymorphism of the promoter of the gene of endothelial NO-synthase (eNOS), Pro/Ala gene polymorphism of γ-receptor which activates peroxisome proliferation (PPARG), G/C polymorphism of the 7th intron of the gene of α-receptor which activates peroxisome proliferation (PPARA), Pro582→Ser (C/T) polymorphism of the gene factor which is induced by hypoxia (HIF-1α), Ala203Pro gene polymorphism of the gene of β-coactivator PPARγ (PPARGC1B), C-1306T gene polymorphism of matrix metalloproteinase 2 (MMP2), Ser422Gly gene of elastin (ELN), Tag1A gene polymorphism of dopamine receptor of type II (DRD2).

Statistical analyses were conducted using SPSS ver.17.0 software package. Genotype distribution and allele frequencies between each of the three groups of athletes and controls were compared using χ² tests. P values < 0.05 were considered statistically significant.

The Regional Ethics Committee (Kiev, Ukraine) approved the study and written informed consent
was obtained from each participant. All experiments were performed in accordance with the ethical standards of the Helsinki Declaration. All genotyping analyses were conducted blind to subject identity.

**Results and discussion**

According to the results of genotyping, the distribution of *ACE, ACTN3, HIF1A, PPARA, PPARG, PPARC1B, DRD2* polymorphisms genotype and allele frequencies in the groups of athletes and controls, weren’t significantly different and were in line with Hardy–Weinberg equilibrium ($p^2_{\text{ACE}} = 0.51; p^2_{\text{HIF1A}} = 0.65; p^2_{\text{PPARA}} = 1; p^2_{\text{PPARG}} = 0.63; p^2_{\text{PPARC1B}} = 0.3; p^2_{\text{DRD2}} = 0.26$).

According to the literature, D allele of *ACE* gene is associated with the development of speed, strength and muscle mass (Puthuacheary, 2011). In our work, the likely difference by this allele was not identified in the group of athletes of power-oriented events, though there is a slight (5.5%) prevalence of representatives with D/D genotype in the group of athletes. D allele was most frequent in the subgroup of athletes specializing in sprint (13.5% higher than the control group).

The distribution of genotypes by T→C polymorphism of *eNOS* gene in the group of athletes meets the Hardy–Weinberg equilibrium ($p = 0.05$). In the group of athletes, the frequency of T/T genotype is 15.6% higher and the frequency of T/C and C/C genotypes is 8.3% and 7.3% lower respectively. The allele frequency distribution by this polymorphism in the group of athletes differs definitely from the control group ($p = 0.02$) (Fig. 1) by increased frequency of T allele and decreased frequency of C allele (11.5%).

**Fig. 1.** Distribution of the frequency of the alleles of *eNOS* gene in the group of athletes specializing in power-oriented events of track and field athletics and in the control group (%): A – athletes; B – control group

Since T→C polymorphism of *eNOS* gene causes the reduced production of NO in human blood (Dosenko, Zagoriy, 2005), which plays an important role in ensuring the long-term adaptation of the organism to a significant amount and high intensity of exercise load, it is obvious that C allele is not favorable for these types of sports.

Ser 582 allele (T) of *HIF1A* gene in scientific literature is considered a marker of disposition to the sports aimed at developing speed and strength (Cięszczyk et al., 2011; Gabbasov et al., 2013). We have not determined in our study the importance of this marker for the predisposition to exercises with power-oriented types of track and field athletics. When the athletes were classified by the types of sports, there was a slight (4.2%) predominance of the number of persons with T allele in the group of jumpers. Attempt to analyse the influence of the interaction between *HIF1A* Pro582Ser and *ACTN3* R577X genotypes on sprint performance showed that *HIF1A* Pro582Ser polymorphism by itself is not critical in determining sprint performance. Israelian researchers emphasized that sprinter performance is determined by the interaction between the wild-type *HIF1A* Pro/Pro genotype and *ACTN3* RR genotype (Eynon et al., 2010).

The distribution of genotypes by *MMP2* gene in the group of athletes differed significantly from the control group ($p = 0.003$) and was characterized by increased frequency of T/T genotype (11.8%) (Fig. 2). Given that this polymorphism influences the changes of the expression of *MNR2* gene, which results in a reduced amount of enzyme that degrades the connective-tissue components of the muscles, we can assume that the decreased splitting of the intercellular matrix of the muscle fibres, which retain the structural integrity and composition of a cell, causes higher results in the group of athletes who do power-oriented kinds of sports. I.e. during intense power exercise loads, the reduction of the intensity of the destructive processes caused by T/T genotype of *MNR2* gene increases the physical performance in these sports.

**Fig. 2.** Distribution of the frequency of genotypes by C/T polymorphism of *MMP2* of the athletes specializing in power-oriented events of track and field athletics and of the control group: A – athletes; B – control group
We know from the published data that the polymorphism of ACTN3 gene is considered to be one of the most influential and important genes for the performance in power-oriented kinds of sports. According to our results, XX genotype is found 9.4% more rarely in the group of athletes than in the control group, though there was no definite difference in the distribution of the genotype and allele frequencies between the control group and the group of athletes with ACTN3 gene.

G/G genotype by G/C polymorphism of PPARA gene is found more frequently in the group of athletes doing power-oriented events of track and field athletics then in the control group by 10.1%, though the frequency of encountering alleles in these groups is the same.

The distribution by PPARA gene polymorphism in the group of athletes is characterized by slightly more frequent Ala/Ala genotype and Ala allele.

The analysis of the frequency of genotypes in the groups divided by events show that by ACE gene the samples of the athletes doing different sports did not differ significantly. The most special genotype distribution was in the group of athletes specializing in sprint. This group was characterized by a low number of persons with I/I genotype and a high number of persons with D/D genotype.

Differences of distribution of eNOS gene polymorphism genotypes in the jumpers were the most significant. The frequency of T and C alleles differed significantly from the control group by 14.2% (p = 0.03) (Fig. 3). Analysis of PPARA gene polymorphism showed the samples of the athletes who specialized in sprint differed significantly by a high frequency of Ala allele (p = 0.04) compared to the control group. The jumpers’ frequency of Ala allele is the lowest among all the groups. The ‘sprint’ and ‘jumping’ groups differed significantly by the frequency of Ala allele by 16.3% (p = 0.04). The highest frequency of Pro allele of PPARA gene polymorphism was in the group of jumping (Fig. 4).

According to the PPARA gene polymorphism, the group of throwers significantly differed from the control group (p = 0.04) and from the jumpers (p = 0.02), with high frequency of G allele (Fig. 5). According to the ACTN3 gene polymorphism, the athletes who specialized in jumping were characterized by the more frequency of R allele. In this subgroup, the frequency of this allele was 15.2% with significant difference between the control group (p = 0.04) and from the jumpers (p = 0.04). Throwers had also high frequency of R allele than the control group – by 15.5% (p = 0.04). Throwers had also high frequency of R allele than the control group (by 13.7%). The lowest frequency was in the group of athletes who specialized in sprint. The difference of frequency of the rare allele between the sprint athletes and jumpers was 15.2% with significant difference between the genotype frequencies (p = 0.04) (Fig. 6).
3. The group of the athletes who specialize in sprint differs significantly from the control group by high frequency of Ala allele and lower frequency of Pro allele by PPARG gene (p = 0.04); and from the group of athletes engaged in jumping, with lower rate of R allele of ACTN3 gene.

4. The group of the jumpers is significantly different from the control group by T(-786)→C of eNOS gene polymorphism with high frequency of T allele (p = 0.03); from the athletes engaged in sprint with higher frequency of Pro allele by PPARG gene (p = 0.04); by higher frequency of G allele by PPARA gene compared to the athletes who specialize in throwing; by higher frequency of R allele by ACTN3 gene compared to the control group and athletes engaged in sprint.

5. Using the molecular and genetic markers will allow improving the selection to the power-oriented kinds of track and field athletics, supplementing the results of pedagogical testing and morphological criteria.

REFERENCES


23. Бобровник, В. И. (2013). Система оценки и прогнозирования физического состояния квалифицированных спортсменов в лёгкой атлетике. "Педагогика, психология и медико-биологические проблемы физического воспитания и спорта", 1, 12–19.


Buvo išmėgta 73 lengvosios atletikos jėgos rungčių sportininkų ir 283 nesportuojančių asmenų DNR. Šiame straipsnyje aptariai dešimt galimų genų ir pateikiami statistinė analizė siekiant išsiaiškinti lengvosios atletikos jėgos rungčių sportininkams tinkamiausius jų polimorfinius darinius. Naudojant polimerų grandinės reakcijos metodą: I/D (ACE), R577X (ACTN3), T-786→C (eNOS), Pro/Ala (PPARG), G/C (PPARA), Pro. →Ser (HIF-1α), Ala203Pro (PPARGC1B), C-1306T (MMP2), Ser422Gly (ELN), Tag1A (DRD2), nustatytas skirtumų reikšmingumas tarp lengvosios atletikos jėgos sporto rungtis kultivuojančių sportininkų ir kontrolinės grupės atstovų alelinio NO-sintazės genų polimorfiškumo ir metaloproteinazės genotipo 2 genų polimorfiškumo pasiskirstymo atvejais. T alelio T-786→C polymorphizmo eNOS dažnis vyrauja sportuojančių asmenų grupėje 11,5 % (p = 0,02). T/T genotipo C-1306T (MMP2) dažnis yra 11,8 % (p = 0.003) didesnis nei kontrolinėje grupėje. Skirtingas sportines veiklas kultivuojantys sportininkai pasižymi genetiniu heterogeniškumu. Nustatyti aleliai, reikalingi norint didelio sportinio meistriškumo su jėga susijusiose lengvosios atletikos rungtyse.

Sporto genetikos tyrimas galima remtis siekiant geriau atrinkti ir ugdyti sportininkus, atskleidžiant jų turimą sportinį potencialą. Mūsų tyrimas pateikia įrodymų dėl ryšio tarp genų polimorfiškumo ir didelio meistriškumo sportininkų lygio. Tačiau tą pačią sportą kultivuojantys sportininkai skiriasi genotipų pasiskirstymu ir genų aleleiais, susijusiais su dideliu meistriškumu. Tik nustačius atitinkamai sporto šakai tinkamą genotipinę kombinaciją, galima padidinti genetinio testavimo transporte potencialą.

Raktažodžiai: sportinė atranka, sporto genetika, genų polimorfiškumas, su jėga susijusios sporto šakos, lengvoji atletika.