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ACTN3 Genotype in Professional Sport Climbers

Michał Ginszt,[1] Małgorzata Michalak-Wojnowska,[2] Piotr Gawda,[1]
Magdalena Wojcierowska-Litwin,[2] Iwona Korszeń-Pilecka,[2] Małgorzata Kusztelak,[3]
Rafał Muda,[4] Agata A. Filip,[2] and Piotr Majcher[1]

[1]*Chair and Department of Rehabilitation, Physiotherapy, and Balneotherapy, Medical University of Lublin, Lublin, Poland;* [2]*Department of Cancer Genetics, Medical University of Lublin, Lublin, Poland;* [3]*Mazowieckie Rehabilitation Center "STOCER", Warsaw, Poland; and* [4]*Department of Banking, Maria Curie-Skłodowska University of Lublin, Lublin, Poland*

Abstract

Ginszt, M, Michalak-Wojnowska, M, Gawda, P, Wojcierowska-Litwin, M, Korszeń-Pilecka, I, Kusztelak, M, Muda, R, Filip, AA, and Majcher, P. ACTN3 genotype in professional sport climbers. *J Strength Cond Res* 32(5): 1311–1315, 2018—The functional RR genotype of the alpha-actinin-3 (*ACTN3*) gene has been reported to be associated with elite sprint/power athlete status. Although large and rapidly increasing number of studies have investigated the associations between the *ACTN3* genotypes and athletic performance in various sport disciplines, there is a lack of studies on the genetic predisposition in sport climbing, which was selected to be part of the next Summer Olympic Games in Tokyo 2020 with three subdisciplines ("lead climbing," "speed climbing," and "bouldering"). The aim of the study is to determine the frequency distribution of *ACTN3* genotypes and alleles in professional lead climbers and boulderers. 100 professional sport climbers from Poland, Russia, and Austria were divided into 2 equal groups: professional boulderers and professional lead climbers were involved in the study. *ACTN3* allele frequencies and genotypes were compared with 100 sedentary controls. Genotypes were determined using polymerase chain reaction–restriction fragment length polymorphism method. The percent distribution of RR genotype in the boulderers was significantly higher than in lead climbers and controls (62 vs. 26%; 33%, respectively; $\chi^2 = 17.230$, $p = 0.0017$). The frequencies of *ACTN3* R allele in boulderers differed significantly from lead climbers and controls (77 vs. 51%; 58%, respectively; $\chi^2 = 15.721$, $p = 0.0004$). The proportion of the *ACTN3* RR genotype is significantly higher in boulderers than in lead climbers and may be related to the specific type of predisposition to this subdiscipline.

Key Words genetics, sport climbing, rock climbing, sport performance

Introduction

Sport climbing, a popular sport worldwide, was selected to be part of the next Summer Olympic Games in Tokyo 2020 with three subdisciplines ("lead climbing," "speed climbing," and "bouldering") (16). Both lead climbing and bouldering, the 2 most practiced climbing styles, can be performed either indoors in climbing gyms or at the natural rock (8,27). In bouldering, the climber ascends short technical routes on low walls up to 4–5 meters using crash mats instead of ropes (8,12). In lead climbing, routes are typically up to 30 m high, where the climber is attached to a rope clipped into permanent bolts using "quickdraws," spaced intermittently from the bottom up (29). To help climbers locate a route or boulder problem of an appropriate difficulty, each route or boulder problem has an associated difficulty rating, based on the size of the hand-holds and foot-holds, the distance between hand-holds or foot-holds, the degree of overhang, and frictional coefficient of the rock (25). In Europe, the most widely used climbing scales to describe the difficulty of a route or boulder problem are the French/sport scale for lead climbing and Font (Fontainebleau) scale for bouldering (15,19). Moreover, the International Rock Climbing Research Association (IRCRA) Reporting Scale is to be used for climbing classification and statistical analyses studies, as one that matches the number of grade steps in the most commonly used climbing scales, according to the recommendation of the IRCRA in 2016 (6).

The associations between morphological and functional variables (body composition, hand grip strength, finger grip endurance, and aerobic and anaerobic metabolism) and performance in sport climbing have been consistently described in the literature (4,10,15). Based on the nature of the effort and strength

Address correspondence to Michał Ginszt, michal.ginszt@umlub.pl.

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Table 1. Genotype frequency and allelic distribution for the R577X polymorphism of the *ACTN3* gene in professional climbers and controls.

	Professional climbers (n = 100)		Controls (n = 100)
Genotype frequency			
RR	44%		33%
RX	40%		50%
XX	16%		17%
χ^2		2.713	
p		0.258	
Allele frequency			
X	36%		42%
R	64%		58%
χ^2		0.757	
p		0.384	

and endurance parameters of climbers, bouldering can be classified as a strength/power discipline, in comparison with lead climbing, which requires higher endurance level (8,20). Several studies have revealed an explosive power profile for boulderers, who have a higher rate of climbing-specific strength than lead climbers (8,15,19). Lead climbers are more capable of longer routes than boulderers, where the duration of the activity is shorter (30 seconds for bouldering vs. 2–7 minutes for lead climbing) (14,15,28). Moreover, the finger grip endurance and the level of grip force are more dependent on the subdiscipline rather than the level of experience (15). Thus, muscular strength and endurance are considered as important determinants of sport climbing. These physiological parameters are highly dependent on muscle fiber composition, which is strongly influenced by genetic factors.

Table 2. Genotype frequency and allelic distribution for the R577X polymorphism of the *ACTN3* gene in boulderers (B), lead climbers (LC), and controls (C).*

	B (n = 50)	LC (n = 50)	C (n = 100)
Genotype frequency			
RR	62%	26%	33%
RX	30%	50%	50%
XX	8%	24%	17%
χ^2 (B vs. LC + C)	17.230	NA	NA
p (B vs. LC + C)	**0.0017**	NA	NA
χ^2 (LC vs. B + C)	NA	1.047	NA
p (LC vs. B + C)	NA	0.306	NA
Allele frequency			
X	23%	49%	42%
R	77%	51%	58%
χ^2 (B vs. LC + C)	15.721	NA	NA
p (B vs. LC + C)	**0.0004**	NA	NA
χ^2 (LC vs. B + C)	NA	2.182	NA
p (LC vs. B + C)	NA	0.140	NA

*B = boulderers; LC = lead climbers; C = controls.

ACTN3, the gene encoding for the synthesis of α-actinin-3 in skeletal muscle fibers, is the first structural skeletal-muscle gene associated with athletic performance (18). α-actinin-3 is specifically expressed in fast-twitch myofibers responsible for generating force at high velocity (30). A common genetic single nucleotide polymorphism at codon 577 of the *ACTN3* results in the replacement of a codon encoding an arginine (R) with a premature stop codon (X), creating 2 alleles (R and X) and three possible genotypes (RR, XX, RX) (26). The R allele encodes normal functional protein, as opposed to the X allele, which encodes a shorter and nonfunctional version of α-actinin-3 (21). For that reason, XX homozygotes are α-actinin-3 deficient (3).

Yang et al. (30) discovered for the first time a significant association between the *ACTN3* genotype and athletic performance, reporting that the RR genotype was overrepresented in the elite sprint/power athletes. Since that time, the *ACTN3* RR genotype has been correlated with elite sprint/power athletic performance in several replication studies (7,13,22,23,31). These findings suggest that *ACTN3* RR genotype is associated with power performance in comparison with XX genotype, which might be postulated to contribute to endurance performance (1,2,11,24). Moreover, the cross-sectional area of type IIa and IIx fibers was larger in *ACTN3* RR genotypes compared with XX individuals (5). Although the large and rapidly increasing number of studies have investigated the associations between the *ACTN3* RR genotype and athletic performance in various sport disciplines (running, jumping, rugby, football, swimming, and cycling), there is a lack of studies on the genetic predisposition in sport climbing (17).

We hypothesized, based on the previous genetics studies and strength/power nature of the effort in bouldering, that the *ACTN3* RR genotype will be more frequent among boulderers in comparison with lead climbers and controls.

Methods

Experimental Approach to the Problem

To determine the frequency distribution of *ACTN3* genotypes and alleles in professional boulderers and lead climbers, genomic DNA was extracted and the genetic data were compared among the groups.

Subjects

All sportsmen and controls provided their written informed consent to participate in the study. The study protocol was approved by the institutional ethics committee of the Medical University of Lublin, Poland (KE-0254/331/2015) and was in accordance with the Declaration of Helsinki for Human Research.

The sample comprised 100 professional climbers (84 males and 16 females; age 18–37 years) from Poland (82%), Russia (12%), and Austria (6%) divided into 2 equal groups: 50 professional boulderers (45 males and 5 females; age 18–35 years) and 50 professional lead climbers (39 males and 11 females; age 18–37 years). Both boulderers and lead climbers were classified into 2 groups: higher elite and elite athletes, based on the self-reported most difficult boulder problem/route ever climbed graded in Font scale for bouldering and French/sport scale for lead climbing. Both scales were converted to IRCRA Reporting Scale (IRS) based on IRCRA standards for the International Rock Climbing Association (6).

Classification in bouldering comprised higher elite males (IRS: ≥28, $n = 20$) and females (IRS: ≥27, $n = 1$), and elite males (IRS: 24–27, $n = 25$) and females (IRS: 21–26, $n = 4$), whereas classification in lead climbing comprised higher elite males (IRS: ≥28, $n = 23$) and females (IRS: ≥27, $n = 1$), and elite males (IRS: 24–27, $n = 16$) and females (IRS: 21–26, $n = 10$). A group of 100 healthy, sedentary Poles (87 males and 13 females; age 23–44 years) served as controls.

Procedures

Genomic DNA was extracted from buccal swabs with GeneJet Kit (Life Technologies). Genotypes were determined using polymerase chain reaction–restriction fragment length polymorphism method, according to the procedure described by Fedotovskaya et al. (9). All samples were successfully genotyped.

Statistical Analyses

Genotype frequencies of the R577X polymorphism in the *ACTN3* gene and the allele frequencies were compared among the groups using the χ^2 test, p-value <0.05 was considered as statistically significant, p-value <0.01 was considered as extremely statistically significant. The IBM SPSS version 21 software was used to perform all statistical evaluations.

Results

Genotype frequencies were consistent with Hardy–Weinberg equilibrium (controls: $p = 0.793$; boulderers: $p = 0.279$; and sport climbers: $p = 0.998$). The frequency of the RR, RX, and XX genotypes and the frequency of the R and X allele in the control group (RR = 33%; RX = 50%; XX = 17%; MAF(R) = 0.58; MAF(X) = 0.42) agreed with data reported in the literature (Table 1) (21,30).

The distribution of RR, RX, and XX genotypes and R and X alleles varied between professional climbers and control group, but the differences were not statistically important ($\chi^2 = 2.713$, $p = 0.258$ and $\chi^2 = 0.757$, $p = 0.384$, respectively) (Table 1).

From the perspective of percentage distribution of RR genotype, the general division of the study group into boulderers, lead climbers, and controls may be presented in following way. The % distribution of RR genotype was highest in the boulderers group and was almost 2 times higher than in lead climbers and controls (62 vs. 26%; 33%, respectively). The frequencies of RR, RX, and XX genotypes and R and X alleles in boulderers differed from lead climbers and controls and the differences were extremely statistically significant ($\chi^2 = 17.230$, $p = 0.0017$; $\chi^2 = 15.721$, $p = 0.0004$) (Table 2).

Table 3. Genotype frequency and allelic distribution for the R577X polymorphism of the *ACTN3* gene in elite (E) and higher elite (HE) boulderers (B), E and HE lead climbers (LC), and controls (C).

	B ($n = 50$)		LC ($n = 50$)		
	E ($n = 29$)	HE ($n = 21$)	E ($n = 26$)	HE ($n = 24$)	C ($n = 100$)
Allele frequency					
R	68%	88%	60%	42%	58%
X	32%	12%	40%	58%	42%
χ^2 (E vs. HE)	5.03		3.22		NA
p (E vs. HE)	**0.025**		0.073		NA
χ^2 (E vs. HE vs. C)	14.37		4.58		NA
p (E vs. HE vs. C)	**0.0008**		0.101		NA
Genotype frequency					
RR	48.3%	81%	30.8%	20.8%	33%
RX	41.4%	14.2%	57.7%	41.7%	50%
XX	10.3%	4.8%	11.5%	37.5%	17%
χ^2 (E vs. HE)	5.55		4.62		NA
p (E vs. HE)	0.063		0.099		NA
χ^2 (E vs. HE vs. C)	16.96		6.771		NA
p (E vs. HE vs. C)	**0.002**		0.148		NA

Bolded differences are statistically significant.

The % distribution of XX genotype and X allele was highest in lead climbers, but comparing with controls, statistically important difference was observed neither in the genotype frequency nor in the allele frequency ($\chi^2 = 1.047$, $p = 0.306$ and $\chi^2 = 2.182$, $p = 0.140$, respectively) (Table 2).

The subdivision of the study group into higher elite climbers, elite climbers, and controls allows us to present the results in the following way. The % distribution of the R and X alleles in higher elite boulderers differed significantly from elite boulderers (88 vs. 68%, 12 vs. 32%, respectively; [$\chi^2 = 5.03$, $p = 0.025$] and controls [$\chi^2 = 14.37$, $p = 0.0008$]) (Table 3). The % distribution of the R and X alleles in higher elite lead climbers did not differ significantly from elite lead climbers (42 vs. 60%, 58 vs. 40%, respectively; [$\chi^2 = 3.22$, $p = 0.073$] and controls [$\chi^2 = 4.58$, $p = 0.101$]) (Table 3). The frequencies of RR, RX, and XX genotypes in higher elite boulderers did not differ significantly from elite boulderers; however, there is an observable tendency for that to happen ($\chi^2 = 5.55$, $p = 0.063$). Comparing with controls, the frequencies of RR, RX, and XX genotypes in higher elite boulderers statistically differ ($\chi^2 = 16.96$, $p = 0.002$). The frequencies of RR, RX, and XX genotypes in higher elite lead climbers did not differ significantly from elite lead climbers ($\chi^2 = 4.62$, $p = 0.099$) nor from controls ($\chi^2 = 6.771$, $p = 0.148$) (Table 3).

Discussion

The influence of individual gene variants on athletic status is at present a matter of investigation worldwide. *ACTN3* gene has been most widely associated with athletic performance in various sport disciplines of all human genes (17). The aim of the present study is to determine the frequency distribution of *ACTN3* genotypes and alleles in professional boulderers and lead climbers. To the best of our knowledge, this is the first report on *ACTN3* genotype distribution not only in professional climbers, but also throughout themed climbing.

The main finding of our study was that the RR genotype frequency was significantly higher in boulderers than in lead climbers and controls. Thus, *ACTN3* RR genotype, which was frequently associated with power performance in several various studies, may have an influence on determining predisposition to the force development in boulderers. Similar results, which genotype distribution of the power athletes was significantly different from endurance athletes, and *ACTN3* RR genotype was significantly higher in international-level than in the national-level sprint/power athletes were reported by Yang et al. (2016) (31). The study presented by Kikuchi et al. (13) also confirmed the association between the RR genotype and elite sprint/power track and field athlete's status. Hence, the hypothesis that the RR genotype has a great impact on determining muscle strength seems to be justified. Moreover, our study reported that in higher elite boulderers, the frequency distributions of the R allele are higher in comparison with elite boulderers and controls. Therefore, our result suggests an association between *ACTN3* R allele and top-level bouldering performance. The results of our study are in agreement with the systematic review and meta-analysis by Ma et al. (17), which reported, based on 88 articles on *ACTN3* gene, a significant association between *ACTN3* R allele and power sports performance (odds ratio = 1.21; 95% confidence interval = 1.03–1.42). Based on these results and our findings, one can infer that the R allele may be associated with a predisposition necessary for better performance in short sequence of very powerful bouldering moves. The results of our research seem to be consistent with physiological parameters of boulderers. Fanchini et al. (2012) (8), in the study on climbers of national-international level, reported that isometric maximal voluntary contraction and rate of force development capacity of finger flexors were significantly greater in boulderers compared with lead climbers. Similar results were presented by Macdonald and Callender (19), who reported that hand grip strength and strength of fingers characteristic for climbing were greater in highly accomplished boulderers compared with elite lead climbers and nonclimbing controls. However, the relationship between physiological parameters and the *ACTN3* genotype of sport climbers requires further research.

Although differences in frequency of XX genotype were not statistically significant, we have observed an overrepresentation of this genotype in lead climbers compared with the boulderers and controls. The previous results, presented by Ben-Zaken et al. (2), have reported that *ACTN3* XX genotype frequency was significantly higher among long-distance runners (35.4%; $n = 65$) compared with short-distance runners (16.7%; $n = 72$) and controls (18.4%; $n = 217$). In our study, X allele is overrepresented in higher elite lead climbers in comparison with elite lead climbers, which could indicate the existence of X allele's predisposition necessary for better performance in the subdiscipline. Having in mind the above mentioned results, one may presuppose the endurance nature of lead climbing. These suppositions are consistent with the study results presented by Grealy et al. (11), supporting our observations for an association between X allele and high-level endurance performance in Ironman World Championship athletes. However, these are only suppositions that need to be confirmed in further studies in larger groups of climbing athletes.

In summary, we have shown that proportion of the *ACTN3* RR genotype is significantly higher in boulderers than in lead climbers and may be related to the specific type of predisposition to this climbing subdiscipline. According to our results, we hypothesize that *ACTN3* genotype could be used to find the proper climbing subdiscipline for an athlete to help him succeed in professional sports climbing, which is not only related to individual physiological and genetic variables, but is also the result of interaction between physiological and psychological factors.

Practical Applications

A genetic profile combined with optimal training is an important factor for professional athletic performance. In our study, *ACTN3* RR genotype was significantly higher in boulderers than in lead climbers and might be an important factor for better performance in bouldering.

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M. Ginszt and M. Michalak-Wojnowska have equally contributed to the submitted work and may be considered the "first" authors.

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