Section: Original Investigation

Article Title: *ACTN3* R577X Polymorphism and Explosive Leg Muscle Power in Elite Basketball Players

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ABSTRACT

Purpose: The purpose of this study was to determine the association of the *ACTN3* R577X polymorphism with leg muscle explosive power in Spanish (Caucasian) elite basketball players and controls. **Methods:** 100 (60 men) elite basketball players (cases) and 283 non-athletic controls participated in the study. We assessed power performance by means of the vertical squat and counter-movement jump tests. **Results:** Genotype distributions did not differ between groups (cases: 37.0% (RR), 42.0% (RX) and 21.0% (XX); controls: 31.8% (RR), 49.8% (RX) and 18.4% (XX); P=0.353). We did not observe any effect of the *ACTN3* R577X polymorphism on study phenotypes in either group, including when we performed the analyses separately in men and women. We found no association between the *ACTN3* R577X polymorphism and the likelihood of being an elite basketball player using the dominant or the recessive model, and the results remained unaltered when the analyses were adjusted for sex, weight, height and age or when performed for men and women separately. **Conclusions:** Although the *ACTN3* R577X is associated with 'explosive' muscle performance and this phenotype is important in the sport of basketball (i.e. during jumps), we found no association with leg 'explosive' power in elite basket players or with the status of being such type of athlete.

Key words: basketball, jump, power, α-actinin-3, genotype.

INTRODUCTION

α-Actinin-3 is a structural protein that is the predominant component of the sarcomeric Z-discs, and it confers type II fibers, where it is predominantly expressed, with a higher capacity for the absorption/transmission of force at the Z line during rapid contractions compared with type I fibers.[1] The R577X polymorphism (rs1815739) of the gene (*ACTN3*) encoding α-actinin-3, which results from a C-to-T transition at position 1,747 in exon 16 that substitutes an arginine residue at codon 577 for a premature stop codon,[2] may be associated with muscle phenotypes, particularly with the ability to produce powerful muscle contractions.[3]

The effect of the *ACTN3* genotype has mainly been studied in elite athletes, based on the hypothesis that its influence on muscle function would be most readily observable at the extremes of human performance. The α-actinin-3-deficient XX genotype (with a frequency of ~18% among European Caucasians) is believed to preclude top-level athletic performance in 'pure' power and sprint sports (sprinting, jumping, throwing events),[4] especially in women,[5] whereas, compared with the general population, the X allele tends to be overrepresented in those humans with an 'extreme endurance phenotype', i.e. elite endurance athletes.[4-6]

Basketball has a remarkable level of popularity worldwide. The game is physically demanding, requiring players to participate in repeated bouts of intense actions (sprinting, shuffling, and jumping) separated by short bouts of low-intensity activity (walking, jogging) and recovery.[7, 8] As a result, basketball players must draw on many fitness components including particularly muscular power.[8, 9] Besides technical and tactical skills, muscular strength and 'explosive' leg power are indeed the most important factors contributing to successful performance during elite basketball competitions.[10] Since α-actinin-3 is a determinant factor for producing high-power, high-velocity muscle contractions,[11] the

ACTN3 R577X polymorphism is a candidate to explain, at least partly, individual variations in basketball performance.

To our knowledge, there are no genetic association studies directly linking muscle phenotypes with specific polymorphisms in basketball. In order to examine the possible influence of the *ACTN3* R577X polymorphism on the ability to produce peak ('explosive') power across the human performance continuum, we studied the association of the *ACTN3* R577X polymorphism with leg muscle 'explosive' power performance in a group of elite basketball players (cases, men and women) and in a group of nonathletic young adults (controls, also men and women). A secondary goal was to determine whether the genotypic frequencies of the *ACTN3* R577X genotypes differed between cases and controls. We hypothesized that (i) the *ACTN3* R577X polymorphism is associated with jumping performance in elite basketball players, with the RR/RX genotype having a favorable influence compared with the XX (i.e. α-actinin-3 deficient) genotype; and (ii) the XX genotype is less frequent in cases compared with controls.

METHODS

Subjects

Written informed consent was obtained from each subject. The study protocol was approved by the institutional ethics committee [*Universidad Europea de Madrid* (UEM), Spain] and was in accordance with the Declaration of Helsinki for Human Research of 1974 (last modified in 2008).

The present study comprised 283 (216 men, 67 women) healthy young adults (University students) and 102 elite basketball players (61 men, 41 women). All of the basketball players were participants of the major Spanish Leagues, i.e. *ACB/LEB oro* (men) or *Liga Femenina LFB/LF2* (women). All controls and cases were of the same Spanish (Caucasian) ancestry for at least 3 generations. Controls were physically active (Physical

Education students), but were not engaged in competitive sports and performed less than 1 (power) or 3 (endurance) structured weekly training sessions within the last year.

Methodology

Genotype assessment: we extracted genomic DNA from saliva samples of all participants during 2008-2011. Our study followed the recommendations for the human genotype–phenotype association.[12]

Genotyping was performed during 2011 in the genetics laboratory of *Universidad Europea de Madrid* (Spain) following the methodology reported elsewhere.[6] In brief, the polymerase chain reaction (PCR) was performed in order to amplify the sequence containing the mutation. A fragment of 303 bp was amplified using the following primers: forward CTGTTGCCTGTGGTAAGTG GG, with 50 VIC labeling, and reverse TGGTCACAGTATG CAGGAGGG. The PCR conditions were as follows: initial denaturing at 95 1C for 5 min; 35 cycles at 95 1C for 30 s, at 60 1C for 30 s, at 72 1C for 30 s and a final extension at 72 1C for 10 min.

ACTN3 genotypes (rs1815739) were established by enzymatic digestion of amplicons with *Dde I*.[6] The R577X change creates a restriction site resulting in fragments of 108, 97 and 86 bp. Digestion of R577 allele results in fragments of 205 and 86 bp. Digestion products (108 bp for 577X and 205 bp for R577) were detected by capillary electrophoresis in an ABI Prism 310 genetic analyzer (Applied Biosystems, Foster City, California, USA).

Phenotype assessment: we assessed the squat (SJ) and counter-movement jump (CMJ) tests using an infrared contact timing platform (Globus Ergo Tester, Codognè, Italy) to evaluate leg muscles' ability to produce 'explosive' power.[13] Details on the methodology used to assess jump performance can be found elsewhere.[14] In brief, the SJ tests were performed without rebound or previous counter movement. Before the jumps, subjects reached 90º of knee flexion angle for ~1 s while keeping both hands on the hips and the trunk

straight.[15, 16] For the CMJ tests, subjects started from a standing position, with trunk straight, legs extended and both hands on hips and performed a vertical jump with a prior fast counter movement allowing 90º knee flexion.[15, 16] During all jumps, subjects kept both hands on hips, trunk straight and they could not perform hip or knee flexions.[15, 16]

Both tests were performed thrice (each separated by a 2-min rest period) and the best score was recorded. These tests have shown to be reliable.[14] The subjects were encouraged to do their best when performing the tests and were advised not to perform any strenuous physical activity within the previous 48 h. One-week before the tests, all the participants received comprehensive instructions on the tests, after which a familiarization session took place.

Statistical analysis

We performed the analyses with the IBM SPSS package (v. 18.0 for WINDOWS, Chicago, USA), and the level of significance was set at a $P \leq 0.05$. We compared mean differences in the studied phenotypes between controls and cases using 1-way analysis of variance. We analyzed the differences in the studied phenotypes among *ACTN3* R577X genotypes within control and cases' group by regression analysis and 1-way analysis of covariance after adjusting for weight, height and age.

We compared genotype frequencies between groups as wells as between men and women using the χ^2 test. We used logistic regression to calculate the odds ratio (OR) for being an elite basketball player using both dominant (RR vs. RX+XX) and recessive (RR+RX vs. XX) models.

RESULTS

There were no failures in sample collection, DNA acquisition or genotyping procedures, except in 2 elite basketball players (1 woman, 1 man), due to an insufficient

amount of DNA gathered from saliva. The distribution of *ACTN3* R755X genotypes met Hardy–Weinberg equilibrium in both cases and controls (P=0.167 and 0.803 respectively).

Characteristics of study participants by sex and group are shown in Table 1. Cases (both as a group or when examining men and women separately) were significantly older, heavier and taller than their controls (P<0.001). Cases showed better jumping performances as a whole group than controls (P<0.01). While there was no significant differences between men in cases' and control groups (all P>0.1), control women presented better jumping performances that women in the cases' group (P<0.05).

The association between the *ACTN3* R577X polymorphism and studied phenotypes within each group is presented in Tables 2 and 3 for men and women respectively. We did not observe any effect of the *ACTN3* R577X polymorphism on study phenotypes in either group or sex.

Table 4 shows the genotype distribution of the *ACTN3* R577X polymorphism in controls and cases, and the OR and 95% confidence intervals (CI) for being a basketball player depending on *ACTN3* R577X genotypes. Genotype and allele frequencies did not differ between the 2 groups (P=0.403 and P=0.752 respectively). We did not observe genotype differences between sexes in the control (χ^2=2.109, P=0.348) or cases' group (χ^2=0.997, P=0.607). We found no association between the *ACTN3* R577X polymorphism and the likelihood of being an elite basketball player using the dominant or the recessive model. The results remained unaltered when the analyses were adjusted for sex, weight, height and age (data not shown) or when performed for men and women separately (Table 4).

DISCUSSION

The present study indicates that the *ACTN3* R577X polymorphism is not associated significantly with the ability to produce peak ('explosive') power in elite basketball players or in non-athletic people. We also observed that genotype frequencies were similar between

basketball and control groups. Taken together, these findings suggest that the *ACTN3* R577X polymorphism does not influence performance in an explosive power-oriented sport such as basketball.

These results were theoretically unexpected, given the role of α-actinin-3 on skeletal muscle phenotypes, particularly muscle power [11] and the importance of this phenotype for basketball performance.[17] Recent findings from association and case control studies are contradictory. Whereas several studies showed no effect of the *ACTN3* R577X polymorphism on muscular strength and power phenotypes,[14, 18-21] others reported an advantageous effect of the RR[22, 23] or the XX genotype.[24, 25] We found no higher prevalence of the RR genotype in basketball players, although a recent meta-analysis of results from the published literature has provided evidence for a higher frequency of the *ACTN3* R577X RR genotype in European sprint/power athletes compared with their non-athletic peers.[26]

Vertical jump is one of the most prevalent acts performed by basketball players. Jumping acts are part of various defensive (blocking, rebounding, stealing) and offensive maneuvers (passing, rebounding, shooting) performed by basketball players in practices and games.[17] Given that basketball requires players to be fast and explosive and the key role of α-actinin-3 on muscular strength and power,[11] theoretically, the *ACTN3* R577X polymorphism might help explaining, at least partly, individual variations in basketball performance-related phenotypes. The absence of significant ORs for being a basketball player could be partly explained because the possibility of becoming an elite athlete depends on numerous influential factors (including simply social or economic possibility), among which possessing a favourable genetic endowment cannot be discarded but is not necessarily the main one.[27, 28] It is likely that epigenetic factors, environment and the complex gene–gene and gene–environment interactions are also important determinants. Further, there might be other genetic variants not yet explored that could help to explain inter-individual variability in muscle phenotypes.

We believe that the results of our study are overall valid, as all of the following criteria were met:[29] cases clearly presented the main study phenotype (i.e. being an elite basketball player), as we indeed studied some of the best Spanish elite basketball players, with Spanish basketball leagues, especially in the men's category, being recognized as one of most competitive worldwide; genetic assessment was accurate and unbiased with genotype distributions meeting HWE in both groups; and muscle-power phenotype was consistently and reliably assessed. However, our results should be taken with caution due to the relatively

low number of cases we studied. Indeed, although the sample size for detecting associations between disease/health related phenotypes and polymorphic markers is known to be highly affected by factors such as allele frequency, degree of linkage disequilibrium, inheritance models, or the effect size of the genetic variants (e.g. OR), it could be estimated that, for instance, testing a single polymorphism as here would require ~250 cases to obtain a statistical power of 80%.[30] We believe this limitation could be partly overcome by the fact that several valid power-related phenotypes were consistently and reliably assessed by the same researchers. The lack of data from a 'replication' cohort of a different ethnic background is also to be kept in mind. Thus, more research is needed using our model on larger basketball players' cohorts of different ethnic backgrounds in order to replicate the present findings.

PRACTICAL APPLICATIONS

Data suggest that the *ACTN3* R577X polymorphism is not related to 'explosive' power performance in elite basketball players, so this fact should be taken into account when training programs were designed by basketball coaches and trainers.

Because of ORs for being a basketball player based on *ACTN3* R577X genotypes were not significant, the possibility of becoming an elite athlete depends on numerous influential factors. Basketball coaches could consider possessing a favourable genetic endowment but is not necessarily the main one.

CONCLUSIONS

In summary, we did not observe an association between the *ACTN3* R577X polymorphism and 'explosive' power performance in elite basketball players or in non-athletic physically young adults of both genders. Moreover, the genotypic frequencies of the *ACTN3* R577X genotypes were similar between groups, largely discarding, at least in our cohort, a major role of this polymorphism on the status of being an elite Spanish Caucasian basketball player.

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Table 1. Characteristics of study participants by sex and group.

	Controls			**Cases**			*P** all	*P** men	*P** women
	All (n=283)	Men (n=216)	Women (n=67)	All (n=100)	Men (n=60)	Women (n=40)			
Age (years)	21.1 (2.0)	21.25 (2.1)	20.8 (1.6)	25.0 (5.4)	25.5 (5.2)	24.4 (5.8)	<0.001	<0.001	<0.001
Weight (kg)	69.6 (10.6)	73.6 (8.8)	57.8 (6.3)	83.8 (15.1)	91.8 (11.6)	71.8 (11.3)	<0.001	<0.001	<0.001
Height (cm)	173.3 (7.8)	176.3 (5.8)	163.7 (5.5)	189.6(11.9)	195.9 (8.9)	180.1 (9.3)	<0.001	<0.001	<0.001
SJ									
Vertical displacement of CG (cm)	36.7 (6.4)	38.6 (5.5)	30.4 (5.2)	33.4 (7.1)	37.4 (5.4)	27.4 (4.9)	<0.001	0.120	<0.01
Flight time (ms)	544.9 (47.9)	559.5 (41.3)	498.2 (36.8)	520.1(53.4)	550.1 (40.1)	475.2 (36.8)	<0.001	0.120	<0.01
CMJ									
Vertical displacement of CG (cm)	38.0 (6.7)	40.1 (5.8)	31.3 (5.2)	35.4 (7.4)	39.7 (5.1)	28.9 (5.1)	<0.01	0.668	<0.05
Flight time (ms)	555.1 (50.6)	570.7 (43.2)	504.7 (38.7)	535.6(55.1)	568.0 (38.4)	487.0 (37.9)	<0.01	0.668	<0.05

Values are means (standard deviation). Abbreviations: CG, center of gravity; CMJ, counter-movement jump; SJ, squat jump

Table 2. Mean (standard deviation) estimates of study phenotypes by genotypes of the *ACTN3* R577X (rs1815739) polymorphism in men of the control and cases' groups.

	Controls			P add.	P dom.	P recess.	**Cases**			P add.	P dom.	P recess.
	RR (n=70)	RX (n=103)	XX (n=43)				RR (n=20)	RX (n=26)	XX (n=14)			
SJ												
Vertical displacement of CG (cm)	38.9 (5.2)	38.8 (5.8)	37.7 (5.3)	0.614	0.688	0.324	38.35 (5.4)	37.1 (4.9)	36.2 (6.1)	0.551	0.275	0.605
Flight time (ms)	561.6 (39.4)	560.8 (43.4)	552.7 (39.6)				558.8 (40.8)	548.2 (36.5)	541.2 (45.9)			
CMJ												
Vertical displacement of CG (cm)	39.9 (5.9)	40.2 (5.8)	40.0 (5.4)	0.95	0.776	0.966	40.6 (5.4)	39.7 (4.6)	38.6 (5.7)	0.737	0.434	0.720
Flight time (ms)	569.4 (44.7)	571.8 (43.6)	570.0 (40.4)				574.2 (40.4)	567.7 (34.9)	559.8 (42.7)			

Analyses were adjusted for weight, height, and age.

Abbreviations: add., additive; CG, center of gravity; CMJ, counter-movement jump; dom., dominant (RR vs. RX + XX); recess., recessive (RR + RX vs. XX); SJ. squat jump
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Table 3. Mean (standard deviation) estimates of study phenotypes by genotypes of the *ACTN3* R577X (rs1815739) polymorphism in women of the control and the cases' group.

	Controls			P add.	P dom.	P recess.	**Cases**			P add.	P dom.	P recess.
	RR (n=20)	RX (n=38)	XX (n=9)				RR (n=17)	RX (n=16)	XX (n=7)			
SJ												
Vertical displacement of CG (cm)	30.0 (4.7)	31.1 (5.0)	28.3 (4.6)	0.266	0.945	0.116	27.9 (4.2)	27.8 (5.9)	25.1 (3.8)	0.367	0.434	0.159
Flight time (ms)	495.0 (35.7)	503.6 (37.5)	482.5 (34.3)				479.2 (31.8)	478.4 (44.1)	458.2 (28.4)			
CMJ												
Vertical displacement of CG (cm)	31.7 (4.8)	31.5 (5.4)	29.5 (4.74)	0.595	0.785	0.307	29.9 (4.7)	28.6 (5.5)	27.3 (4.9)	0.289	0.128	0.308
Flight time (ms)	507.6 (36.4)	506.4 (40.8)	491 (35.6)				494.6 (35.5)	484.2 (41.4)	474.8 (36.8)			

Analyses were adjusted for weight, height and age.
Abbreviations: add., additive; CG, center of gravity; CMJ, counter-movement jump; dom., dominant (RR vs. RX + XX); recess., recessive (RR + RX vs. XX); SJ. squat jump.

Table 4. Allele and genotype distribution in control and cases.

		Control		Cases		χ^2 (*P*-value)	OR (95% CI) dom.	OR (95% CI) recess.
		n	%	n	%			
All								
	RR	90	31.8	37	37.0	1.816 (0.403)	1.259 (0.782-2.029)	0.847 (0.480-1.494)
	RX	141	49.8	42	42.0			
	XX	52	18.4	21	21.0			
	p (R)	321	56.7	116	58.0	0.100 (0.752)		
	q (X)	245	43.2	84	42.0			
Men								
	RR	70	32.4	20	33.3	0.469 (0.791)	1.043 (0.568-1.915)	0.817 (0.412-1.620)
	RX	103	47.7	26	43.3			
	XX	43	19.9	14	23.3			
	p (R)	243	56.3	66	55.0	0.060 (0.807)		
	q (X)	189	43.7	54	45.0			
Women								
	RR	20	29.9	17	42.5	2.823 (0.244)	1.737 (0.768-3.930)	0.732 (0.249-2.146)
	RX	38	56.7	16	40.0			
	XX	9	13.4	7	17.5			
	p (R)	78	58.2	50	62.5	0.384 (0.536)		
	q (X)	56	41.8	30	37.5			

Abbreviations: CI, confidence interval; dom., dominant (RR vs. RX+XX); OR, odds ratio; recess., recessive (RR + RX vs. XX).