Association between Dermatoglyphic Configuration and the ACTN3 Genotype in Juvenile Male Athletes

Ramon Cunha-Montenegro[1,2], Carlos Renato Paz[3], Eric de Lucena Barbosa[2],
Asdrúbal Nóbrega Montenegro-Neto[2], Michelle Salles de Oliveira[2], Vanduir Soares de
Araújo Filho[2], Fernando Policarpo Barbosa[4], Paula Roquetti Fernandes[4] and
José Fernandes Filho[5]

[1]*Autonomous University of Asunción - UAA/PY;*
[2]*Physical Evaluation Laboratory – UNIPÊ-SANNY;*
[3]*Federal Institute of Education, Science and Technology of Paraíba – IFPB.*
[4]*Center of Excellence in Physical Evaluation – CEAF/RJ/BR;*
[5]*Federal University of Rio de Janeiro – UFRJ-EEFD/RJ/BR*

Abstract

This study examined whether dermatoglyphic characteristics are associated with the α-actinin isoform 3 (ACTN3) R577X sequence variant, employing descriptive and comparative approaches. Boys (N=82) were classified according to the dermatoglyphic configuration of their digital impressions into the following groups: Anaerobic Power (AP=8); Speed Resistance (SR=44); Pure Force (PF=5), and Aerobic Resistance (AR=25). All of the AP group subjects (9.76% of the subjects) had a genetic predisposition for anaerobic power performance, with 37.5% being RR homozygotes and 62.5% being RX heterozygotes. The dermatoglyphic profiles, correlated with the ALW fingerprint formulas, classified the SR group (53.66% of the subjects) as having speed resistance. The PF group (6.09% of the subjects) notably all had AL fingerprints and an absence of W. Finally, the AR group (30.49% of the subjects) had a predisposition for aerobic capacity. In conclusion, dermatoglyphic features can be associated with the R577X **allelic variant** *of the ACTN3 gene, as much through anaerobic muscle power profiling as through the ACTN3 genotype.*

Key words: *alpha-actinin; fingerprint; genetic polymorphism.*

Introduction

Dermopapillary impressions have diverse uses from medical and sports science research to legal identification; physical qualities have been associated with dermopapillary patterns and numbers of lines, and have been related to fingerprint formulas (Zarya et al., 2010; Cunha, Cunha, Schneider, Dantas & Fernandes-Filho, 2006; Reed, Viken & Rinehart, 2006; Kücken & Newell, 2005). The study of dermal papillae, which are formed during fetal development, is based on the scientific principles of permanency, immutability, variation, and universality. These properties provide advantages over other physical and physiological methods of identifying people. In addition, the capacity for fingerprint patterns to be classified and archived renders dermatoglyphics highly suitable for research (Sharma & Bakshi, 2005; Martín, Fañanás, Gutiérrez, Chow, & Bassett, 2004; Burton et al., 2003; Pechenkina, Benfer, Vershoubskaya, & Kozlov, 2000; Cummins & Midlo, 1961).

Sports science examines the mechanisms that facilitate adaptations over the course of time related to the process of human evolution. In many forms of sport, anaerobic power is critical for good performance, and for this reason, studies have focused on this physical quality and how it is manifested in different populations using various techniques. The physical performance of athletes is also dependent on other variables, such as technical, tactical, and psychological properties (Asano, Bartholomeu, Ribeiro, Barbosa, & Sousa, 2009; Cronin, Jones, & Frost, 2007; Bencke, Damsgaard, Saekmose, Jorgensen, & Klausen, 2002; Izquierdo, Hakkinen, Gonzalez-Badillo, Ibanez, & Gorostiaga, 2002).

Studies in the field of genetics have come to occupy a fundamental role in identifying factors associated with physical qualities, especially qualities related to motor coordination and muscular force (McCauley, Mastana, & Folland, 2010; Scott et al., 2010; Tsianos et al., 2010; Zempo et al., 2010; Bray et al., 2009; Missitzi et al., 2004; Yang et al., 2003) presented a classification of more than 200 genetic characteristics associated with athletic performance, among them the actin-binding protein α-actinin isoform 3 (ACTN3).

ACTN3 is selectively expressed in the sarcomere Z-lines of rapid-contraction (type II) muscle fibers. A single nucleotide polymorphism in the gene that encodes human ACTN3 at position 577 has resulted in the existence of two ACTN3 allele types, the normal functional protein (produced by the normal R gene) and an inactive mutant form (produced by the X gene mutation). The R allele of ACTN3 is associated with a predisposition for anaerobic power; RR homozygotes and RX heterozygotes can exhibit greater anaerobic power than XX homozygotes. Several recent studies have demonstrated an association between ACTN3 and the muscle condition needed to execute elite-level rapid and strong movements (Ahmetov et al., 2008; Papadimitriou, Papadopoulos, Kouvatsi, & Triantaphyllidis, 2008; Roth et al., 2008; Niemi & Majamaa, 2005).

Identifying the dermatoglyphic configuration as additional tool for ACTN3 polymorphism could facilitate the process of sports selection and provide some guidance for prescribing training attuned to individual needs. Therefore, the objective of this study was to determine whether there are dermatoglyphic characteristics that

associate with the RR, RX, and XX ACTN3 genotypes that could be used to provide profiles for high performance muscle power in young children and adolescents.

Methods

Study Design and Subjects

This study was an *ex post facto* descriptive and comparative one. It encompassed boys, 7 – 17 years of age (N=82), who played football in the state of Paraíba in Brazil. In accordance with the previously described classifications (Serhiyenko & Lishevskaya, 2010; Dantas, Alonso, & Fernandes-Filho, 2004; Abramova, Nikitina, & Chafranova, 1995), the participants were classified into the following four groups according to their dermatoglyphic patterns: Anaerobic Power (AP=8), Speed Resistance (SR=44), Pure Force (PF=5), and Aerobic Resistance (AR=25). Pure force was considered to be the product of the force that a body segment must produce to achieve its maximum speed i.e., power = force × velocity.

All procedures followed the regulations of National Health Council CNS 196/96. The procedures also followed guidelines for human research, in accordance with protocol 369/10 of the Research Ethics Committee of the Lauro Wanderley University Hospital of the UFPB. Parents and guardians of the subjects were informed of the procedures involved in the investigation and asked to sign a free and clarified consent form, which authorized the minors' participation in the study.

Dermatoglyphic Analysis

A Cross Match® Verifier 320 LC scanner was used to collect digital impressions. Each of the volunteer's fingers was positioned and pressed against the scanner, and then the fingertip was rolled from one side to the other, always toward the index finger. To analyze the images, we used the protocol described by Cummins and Midlo (1961), which classifies pattern types as: "A" Arches (no delta pattern), "L" Loops (delta pattern), and/or "W" Whorls (pattern with two deltas). The indices of the sum of the total quantity of lines (STQL) and the sum of the number of patterns on the fingers (D10) were identified, and the number of lines on the fingers of the left and right hands (LHSTQL and RHSTQL) was verified. Dissociating these factors leads to an incorrect analysis of dermatoglyphic data. This method has been used with elite athletes by linking the character of dermatoglyphic indices to the somato-functional classifications described by various studies (Serhiyenko & Lishevskaya, 2010; Dantas et al., 2004; Abramova et al., 1995).

A minimum value of "0" and a maximum value of "10" were assigned to each of the pattern types (A, L, and W). After the classifications were established, the indexes of digital impressions that corresponded to the STQL and D10 data were calculated, and these variables were treated as quantitative characteristics. The D10 data were ascribed a minimum value of "0" and a maximum of "20," where "0" indicated the presence of an arch (A) in all the fingers, with the absence of deltas. The D10 values were obtained using the following formula: $D10 = \Sigma L + 2(\Sigma W)$. The different representations of

the classifications of digital formulas were: AL, AW, ALW, 10W, L>W, W>L, 10L, and L=W defined as follows: AL, the presence of A and L in any combination; ALW (the presence of A, L, and W in any combination); 10W (10 whorls); W>L (W and L with more than 5 W); L>W (L and W with more than 5 L); 10L (ten loops); L = W (an equal number of L and W).

Genotyping of Residue 577 of ACTN3

DNA Collection

Genetic material was collected using individual sterile cotton buds (swabs) on plastic stems with hydrophillic cotton at one end. The bud was rubbed in the cheek mucosa, then re-placed in its plastic wrapping and sent to the Forensic DNA Laboratory of the Institute for Scientific Policing of Paraíba–SPI/PB/BR.

Genotyping

The collected samples were subjected to DNA extraction according to the procedures outlined by Walsh et al. (1991). Real-time PCR was performed using an IQ5 Thermal Cycler (Biorad) with a kit for determining the R577X polymorphism (Assay Id C_590093_1, from Applied Biosystems) according to the manufacturer's instructions. All procedures for data collection took place at the UNIPÊ/SANNY Physical Evaluation Laboratory of the João Pessoa University Center–UNIPÊ/PB/BR. Genotyping of residue 577 of the ACTN3 locus was performed at the Forensics Laboratory of the Institute for Scientific Policing of the State of Paraíba (SPI/PB).

Statistical Methods

Inferential statistics were applied to interpret the data so as to identify the levels of association and differences between the sample subgroups. The Kolmogorov-Smirnov test was applied to analyze the distribution of the quantitative data. A comparison of variables was initially performed using one-way analyses of variance (ANOVAs) and Scheffé's post hoc test was applied when ANOVAs revealed significant effects. A significance level of $p < 0.05$ was adopted. All statistics were performed using SPSS® (version 14.0) software.

Results

Dermatoglyphic Analysis

Individual subjects' physical aptitudes for anaerobic power, speed resistance, pure force, and aerobic resistance were designated in accordance with the characteristics of their digital impressions based on the numbers of lines, types of STQL digital patterns, and D10 indices (Table 1). The relative frequency results for the formulas and patterns (Table 2) indicated that the AP group showed a clear characteristic of the "ALW" digital formula, whereas the PF groups showed a clear characteristic of the AL digital formula. The SR group showed a frequency relation to nearly all digital formula types, and the AR group had a total absence of arches and high frequencies of L>W and W>L.

Table 1. *Dermatoglyphic profile for anaerobic power, speed resistance, pure force, and aerobic resistance of boys (N = 82) living in the state of Paraíba, Brazil.*

Groups		Dermatographic Profile				
	N	Arch (A)	Loop (L)	Whorl (W)	STQL	D10
AP (Anaerobic Power)	08	1 ≤ **A** ≤ 2	7 ≤ **L** ≤ 8	1 ≤ **W** ≤ 2	60 ≤ **STQL** ≤ 118	9 ≤ **D10** ≤ 11
SR (Speed Resistance)	44	0 ≤ **A** ≤ 4	3 ≤ **L** ≤ 10	0 ≤ **W** ≤ 7	35 ≤ **STQL** ≤ 133	6 ≤ **D10** ≤ 17
PF (Pure Force)	05	1 ≤ **A** ≤ 5	5 ≤ **L** ≤ 9	0 ≤ **W** ≤ 1	24 ≤ **STQL** ≤ 94	5 ≤ **D10** ≤ 9
AR (Aerobic Resistance)	25	0 ≤ **A** ≤ 0	0 ≤ **L** ≤ 0	1 ≤ **W** ≤ 10	135 ≤ **STQL** ≤ 206	11 ≤ **D10** ≤ 20

STQL = Sum of the Total Quantity of Lines; **D10** = Number of Patterns on the Fingers of Both Hands.

Table 2. *Frequency of patterns and of digital formulas in boys living in the state of Paraíba, Brazil.*

Groups	Patterns				Digital Formulas						
	A	L	W		AL	ALW	10W	L>W	W>L	10L	L=W
AP (n = 08)	13%	73%	15%		0%	100%	0%	0%	0%	0%	0%
SR (n = 44)	8%	80%	12%		25%	11%	0%	46%	2%	16%	0%
PF (n = 05)	30%	70%	0%		100%	0%	0%	0%	0%	0%	0%
AR (n = 25)	0%	46%	54%		0%	0%	8%	44%	44%	0%	4%

A= Arch; **L**= Loop; **W**= Whorl; **AP** = Anaerobic Power ; **SR** = Speed Resistance; **PF** = Pure Force; **AR** = Aerobic Resistance.

An analysis of the data dispersion curve showed a normal distribution for all of the quantitative variables (*p* values < 0.05). As shown in Table 3, ANOVAs revealed significant group effects for D10 and STQL (both *p* values < 0.001) as well as significant group effects for the LHSTQL and RHSTQL values for every digit (*p* values ≤ 0.01).

Table 3. *Comparison of the D10, STQL, LHSTQL, and RHSTQL mean values of boys in the state of Paraíba/BR.*

Variables	AP (n = 08)		SR (n = 44)		PF (n = 05)		AR (n = 25)		ANOVA
	Mean ± SD	Min_Max	Mean ± SD	Min_Max	Mean ± SD	Min_Max	Mean ± SD	Min-Max	*p*
D10	10.25 ± 0.89	9-11	10.43 ± 2.18	6-17	7.20 ± 1.79	5-9	15.40 ± 2.66	11-20	0.000
SQTL	94.13 ± 21.0	60-118	101.70 ± 26.1	35-133	66.40 ± 26.81	24-94	155.60 ± 16.43	135-206	0.000
LHSTQL1	11.86 ± 3.18	6-15	12.56 ± 3.98	6-20	16.00 ± 3.61	13-20	15.88 ± 4.35	7-26	0.007
LHSTQL2	7.80 ± 1.92	5-10	9.37 ± 3.60	3-15	12.00 ± 7.07	7-17	14.56 ± 2.62	9-20	0.000
LHSTQL3	9.40 ± 3.51	4-13	9.66 ± 3.55	2-16	9.00 ± 1.41	8-10	14.76 ± 2.44	10-19	0.000
LHSTQL4	11.38 ± 3.70	5-14	12.56 ± 3.69	4-19	6.40 ± 4.98	3-15	16.36 ± 2.93	7-21	0.000
LHSTQL5	12.14 ± 2.04	9-15	10.28 ± 3.56	3-17	10.40 ± 4.67	4-15	15.48 ± 2.84	9-22	0.000
RHSTQL1	11.75 ± 5.60	3-20	13.60 ± 4.76	5-23	11.40 ± 5.59	5-17	16.96 ± 4.72	10-29	0.010
RHSTQL2	9.83 ± 2.04	7-13	9.03 ± 3.13	3-15	4.50 ± 2.12	3-6	14.84 ± 3.16	6-20	0.000
RHSTQL3	8.38 ± 3.11	3-13	9.95 ± 3.61	4-18	6.50 ± 0.71	6-7	14.52 ± 3.39	3-20	0.000
RHSTQL4	12.50 ± 3.46	6-18	12.49 ± 3.43	3-20	9.00 ± 2.16	7-12	17.12 ± 2.65	12-21	0.000
RHSTQL5	11.00 ± 3.16	6-16	10.26 ± 2.99	5-17	8.60 ± 5.13	3-16	15.12 ± 3.37	11-24	0.000

D10 = Number of Patterns on the Fingers of Both Hands; **STQL** = Sum of the Total Quantity of Lines; **LHSTQL** = Sum of the Total Quantity of Lines on the Left Hand; **RHSTQL** = Sum of the Total Quantity of Lines on the Right Hand; SD = Standard Deviation; **Min_Max** = Minimum and Maximum Values.

ACTN3 Genotyping

The AP and AR groups were found to be made up exclusively of subjects with RR and RX genotypes, with the majority being heterozygotes (Table 4). The SR group had individuals of all three genotypes, with slightly more than one-third having the RR genotype, almost one-third having the RX genotypes, and about one-fifth having the XX genotype, which is unfavorable for anaerobic power. Finally, the PF group was genetically disadvantaged in terms of power quality as all subjects in the PF group were XX homozygotes.

Table 4. *Genotype and allele frequencies of the R577X allelic variant of the ACTN3 gene in boys in the state of Paraíba/BR.*

Groups	Genotype Frequency			Allele Frequency	
	RR (%)	RX (%)	XX (%)	R (%)	X (%)
AP (n = 08)	3 (37.5)	5 (62.5)	0 (0)	11 (68.8)	5 (31.2)
SR (n = 44)	18 (35.3)	20 (31.7)	6 (20.6)	56 (63.6)	32 (36.4)
PF (n = 05)	0 (0)	0 (0)	5 (100)	0 (0)	10 (100)
AR (n = 25)	8 (32.0)	17 (68.0)	0 (0)	33 (66)	17 (34)

AP = Anaerobic Power; **SR** = Speed Resistance; **PF** = Pure Force; **AR** = Aerobic Resistance; **RR** = Homozygotes with a genetic predisposition for power; **RX** = Heterozygotes with a genetic predisposition for power; **XX** = Homozygote (absence of ACTN3 in the rapid contraction fibers).

Association of Dermatoglyphic Classification and ACTN3 Genotype

The AP group, which included 9.76% of the total study cohort, included only RR and RX genotypes. They had average values of the D10 (10) and STQL (94) dermatoglyphic indices for the study cohort, with representation of each pattern type: A(~1), L(~7), W(~2).

Slightly more than a half (53.66%) of the study participants were classified into the SR group. The SR group had D10 and STQL mean values (~10 and ~102, respectively) near average for the group and exhibited A(~1), L(~8), W(~1) patterns together with the main digital formulas, AL, L>W, and W>L. Slightly more than two-thirds of the subjects in the SR group were RX heterozygotes and close to one-third were RR homozygotes, and none were XX homozygotes.

The PF group included 6.09% of the participants. They had relatively low D10 and STQL mean values (~7 and ~66, respectively), exhibited A (3) and L (7) patterns at frequencies outside of the AL digital formula, and had an absence of W. All of the participants in the PF group were XX homozygotes.

Finally, 30.49% of the participants were assigned to the AR group. They had relatively high mean D10 (~15) and STQL (~156) values and exhibited L(~5) and W(~5) patterns. Similar to the SR group, slightly more than two-thirds of the subjects in the AR group were RX heterozygotes and close to one-third were RR homozygotes, resulting in a 34% frequency of the X allele.

Discussion

This study examined whether athletic classifications based on dermatoglyphic characteristics are associated with the ACTN3 genotype at the R577X polymorphism. We found that all of the AP group subjects had a genetic predisposition for anaerobic power performance: all carried at least one copy of the active protein allele and their dermatoglyphic profiles were consistent with a predisposition for a high yield of anaerobic power. Similar to the AP group, all of the subjects in the AR group carried at least one R577 allele in the ACTN3 gene, indicating that they too had a genetic predisposition for anaerobic muscular power. The SR group was the only group that included individuals with all three genotypes, though only about 20% had the XX genotype. The PF group was distinct in that all of the subjects in this group had the XX genotype; the PF group also had a notable absence of W in their dermatoglyphic profiles.

Anaerobic Power

Individuals that fit the AP dermatoglyphic classification are considered to be predisposed for achieving intense anaerobic power (Yang et al., 2003; Abramova et al., 1995). The absence of XX genotype individuals in our AP group is consistent with other studies in indicating a performance disadvantage for activities dependent on anaerobic power among individuals with the XX genotype (Alfred et al., 2011; Bray et al., 2009; Montenegro, Barbosa, Dantas, Fernandes-Filho, & Knackfuss, 2008; Cunha et al., 2006; Dantas et al., 2004; Abramova et al., 1995). For example, in comparing sprinters versus a non-athlete control group, Yang et al. (2003) found that sprinters had a higher frequency of the RR genotype than controls (50% vs. 30%), and lower frequencies of the RX (45% vs. 52%) and XX (6% vs. 18%) genotypes than controls. Likewise, in a study comparing the ACTN3 genotypes of sprinters versus endurance runners, Papadimitriou et al. (2008) found that, relative to endurance runners, sprinters had a higher frequency of the RR genotype (73.53% vs. 50%) and a lower frequency of the RX (17.65% vs. 25%) and XX (8.82% vs. 25%) genotypes. Such results indicate that the R allele of ACTN3 favors athletic achievement in fundamentally anaerobic forms of sport. Furthermore, the XX genotype frequencies found among sprinters (Eynon et al., 2009; Yang et al., 2007; Niemi & Majamaa, 2005) are notably lower that those found in the general population. For example, in a study of 154 healthy individuals, including African Americans, Clarkson et al. (2005) found the XX genotype frequency of 26.6%. On the other hand, since all members of the PF group in our study cohort had the XX genotype, it can be deduced that they have a genetic limitation in terms of their ability to produce muscle power due to the premature stop signal in the 577X polymorphism in both of their ACTN3 genes (Eynon et al., 2009; Roth et al., 2008).

Endurance

While individuals of the XX genotype may not be good candidates for sports that require anaerobic power, there is some evidence suggesting that they may be well

suited for endurance sports. Eynon et al. (2009) and Papadimitriou et al. (2008), who describe the R allele of the ACTN3 gene as being advantageous for sports in which sprinting is required, also describe the X allele, and the XX genotype, as potentially beneficial for endurance activities. Consistent with this notion, Yang et al. (2003) found that the XX genotype was more common in endurance athletes (24%) than in the control non-athlete subjects (18%), indicating that there was an increased likelihood of the XX genotype among endurance athletes. In a study examining different kinds of Finnish athletes, Niemi et al. (2005) similarly found that endurance athletes were less likely to have the RR genotype and more likely to have the XX genotype than speed athletes, whereas the speed athlete subgroup did not have any XX genotype individuals. On the other hand, in a study examining the association between ACTN3 genotype and the athletic performance of 992 Greek adolescents, Moran et al. (2005) did not find any evidence that having the X allele was associated with aerobic resistance. Thus, more work is needed to elucidate whether and to what extent the 577X allele may be associated with benefits for athletes engaged in sports with high aerobic resistance demands.

Pure Force

Although the XX genotype was characteristic of all members of the PF group in this study, the relationship between pure force and the ACTN3 gene is also not yet clear. Pure force is related to an individual's capacity to realize maximal dynamic voluntary muscle contraction aided by intra- and inter-muscular coordination. McCauley et al. (2010), who studied the muscular function and muscularity of non-athletic men, found that the ACTN3 polymorphism was not associated with contractile properties, either in absolute terms or in terms of the relative dynamic force of the knee extensor muscles. And Roth et al. (2008) found that, relative to a control group, bodybuilder/ weightlifter athletes—who place high pure force demands on their bodies—had a higher frequency of the RX genotype (62.6% vs. 45.6%), but lower frequencies of *both* the RR genotype (30.7% vs. 38.1%) and the XX genotype (6.7% vs. 16.3%).

Conclusion

Dermatoglyphics was confirmed to be a rational approach for the R577X polymorphism of the ACTN3 gene to predict profile of anaerobic muscle power. As a complement to ACTN3 genotyping, dermatoglyphic analysis has great potential to help in sports selection, especially in terms of flagging those individuals with a characteristic predisposition for anaerobic muscular power versus the individuals who do not have such a predisposition. Finally, our findings confirm the importance of using these two methods to provide coherence and complementarity in evaluating the physical qualities of a population sample.

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Ramon Cunha-Montenegro
Autonomous University of Asunción - UAA/PY
Jejuí 667 st., C.P. 1255, Asunción, Paraguay
Physical Evaluation Laboratory – UNIPÊ-SANNY
BR 230, Km 22, Água Fria, 58000-000 João Pessoa, Paraíba, Brazil
proframon_cunha@hotmail.com

Carlos Renato Paz
Federal Institute of Education, Science and Technology of Paraíba – IFPB.
1º de Maio Av., 720, Jaguaribe, 58.015-430, João Pessoa, Paraíba, Brazil
renatopaz@hotmail.com

Eric de Lucena Barbosa
Physical Evaluation Laboratory – UNIPÊ-SANNY
BR 230, Km 22, Água Fria, 58000-000 João Pessoa, Paraíba, Brazil
eric.luba@hotmail.com

Asdrúbal Nóbrega Montenegro-Neto
Physical Evaluation Laboratory – UNIPÊ-SANNY
BR 230, Km 22, Água Fria, 58000-000 João Pessoa, Paraíba, Brazil
netotraducao@hotmail.com

Michelle Salles de Oliveira
Physical Evaluation Laboratory – UNIPÊ-SANNY
BR 230, Km 22, Água Fria, 58000-000 João Pessoa, Paraíba, Brazil
michelleestatistica@gmail.com

Vanduir Soares de Araújo Filho
Physical Evaluation Laboratory – UNIPÊ-SANNY
BR 230, Km 22, Água Fria, 58000-000 João Pessoa, Paraíba, Brazil
contato@vanduir.com.br

Fernando Policarpo Barbosa
Center of Excellence in Physical Evaluation – CEAF/RJ/BR
http://www.ceafbr.com.br/, Rio de Janeiro, Brazil
fernandopolicar@hotmail.com

Paula Roquetti Fernandes
Center of Excellence in Physical Evaluation – CEAF/RJ/BR
http://www.ceafbr.com.br/, Rio de Janeiro, Brazil
prf@ceafbr.com.br

José Fernandes Filho
Federal University of Rio de Janeiro – UFRJ-EEFD/RJ/BR
http://www.eefd.ufrj.br/, Rio de Janeiro, Brazil
jff@eefd.ufrj.br

Odnos između dermatoglifske konfiguracije i ACTN3 genotipa u maloljetnih muških atletičara

Sažetak

Cilj ovoga istraživanja bio je istražiti postoji li odnos između dermatoglifskih karakteristika i α-aktinin izomorf 3 (ACTN3) R577X varijante. U istraživanju su korišteni deskriptivni i komparativni pristup. Dječaci (N = 82) su klasificirani u skupine na temelju dermatoglifske konfiguracije njihovih digitalnih otisaka na sljedeći način: anaerobna snaga (AP = 8), otpornost pri brzini (SR = 44), čista snaga (PF = 5) i aerobna otpornost (AR = 25). Svi sudionici iz skupine AP (9,76% sudionika) imali su gensku predispoziciju za izvedbu za koju je potrebna anaerobna snaga. Od toga je 37 (5%) bilo homozigotna i 62.5% heterozigotna genotipa. Dermatoglifski profili u korelaciji s formulama otiska prstiju iznjedrili su klasifikaciju SR skupine (53.66% sudionika) kao skupinu s otpornošću pri brzini. Svi sudionici PF skupine (6.09% svih sudionika) imali su Al otiske prstiju i odsutnost W. Na kraju, AR skupina (30.49% svih sudionika) je imala predispoziciju za aerobni kapacitet. Zaključak je da dermatoglifske karakteristike mogu biti povezane s R577X aleličkom varijantom ACTN3 gena, profiliranjem anaerobne mišićne mase, kao i ACTN3 genotipa.

Ključne riječi: *α-aktinin; genski polimorfizam; otisak prsta.*

Uvod

Dermopapilarni se otisci u znanstvenim istraživanjima koriste na različite načine i u različitim područjima, počevši od istraživanja u medicini i sportu do istraživanja osobne identifikacije. Tjelesne kvalitete povezuju se s dermopapilarnim uzorcima i brojevima linija te se dovode u vezu s formulama otisaka prstiju (Zarya i dr., 2010; Cunha, Cunha, Schneider, Dantas i Fernandes-Filho, 2006; Reed, Viken i Rinehart, 2006; Kücken i Newell, 2005). Istraživanje dermalne papile, koja se formira tijekom razvoja fetusa, utemeljena je na znanstvenim principima trajnosti, nepromjenjivosti, varijacije i univerzalnosti. Te karakteristike omogućuju prednost nad ostalim tjelesnim i fiziološkim metodama identifikacije ljudi. Osim toga, kapacitet za uzorke otisaka

prstiju koji se trebaju klasificirati i arhivirati čini dermatoglifiju vrlo pogodnom za provođenje istraživanja (Sharma i Bakshi, 2005; Martín, Fañanás, Gutiérrez, Chow i Bassett, 2004; Burton i dr., 2003; Pechenkina, Benfer, Vershoubskaya i Kozlov, 2000; Cummins i Midlo, 1961).

Znanost o sportu istražuje mehanizme koji olakšavaju prilagodbu vezanu uz procese evolucije ljudi tijekom određenoga razdoblja. U mnogo oblika sportova anaerobna snaga kritična je za dobru izvedbu. Zbog tog su se studije bavile tom tjelesnom kvalitetom i njezinim očitovanjem u različitim populacijama i za to su upotrebljavane različite tehnike. Tjelesna izvedba atletičara također ovisi o različitim varijablama kao što su njihove tehničke, taktičke i psihološke karakteristike (Asano, Bartholomeu, Ribeiro, Barbosa i Sousa, 2009; Cronin, Jones i Frost, 2007; Bencke, Damsgaard, Saekmose, Jorgensen i Klausen, 2002; Izquierdo, Hakkinen, Gonzalez-Badillo, Ibanez i Gorostiaga, 2002).

Istraživanja koja se provode unutar područja genetike imaju temeljnu ulogu u identifikaciji čimbenika povezanih s tjelesnim kvalitetama, a osobito kvalitetama koje se odnose na motoričku koordinaciju i mišićnu silu (McCauley, Mastana i Folli, 2010; Scott i dr., 2010; Tsianos i dr., 2010; Zempo i dr., 2010; Bray i dr., 2009; Missitzi i dr., 2004; Yang i dr., 2003). Navedeni su autori predstavili klasifikaciju više od 200 genskih karakteristika povezanih s atletskom izvedbom, među kojima je i aktin-vezujući protein, α-aktinin izomorf (ACTN3).

ACTN3 je selektivno izražen u sakromer Z-linijama brze kontrakcije (tip II) mišićnih vlakana. Pojedinačan nukleotidni polimorfizam u genu koji inkodira ljudski ACTN3 na poziciji 577 omogućio je postojanje dvaju alela ACTN3 genotipa, normalnog funkcionalnog proteina (koji proizvodi normalni R gen) i neaktivne mutirane forme (koju proizvodi mutacija X gena). R alela ACTN3 gena povezuje se s anaerobnom moći; RR homozigote i RX heterozigote mogu postići veću anaerobnu moć od XX homozigota. Nekoliko recentnih istraživanja pokazalo je povezanost između ACTN3 i mišićne spremnosti potrebne za izvođenje brzih i snažnih pokreta na elitnoj razini (Ahmetov i dr., 2008; Papadimitriou, Papadopoulos, Kouvatsi i Triantaphyllidis, 2008; Roth i dr., 2008; Niemi i Majamaa, 2005).

Identifikacija dermatoglifske konfiguracije kao dodatnog sredstva za ACTN3 polimorfizam može olakšati proces sportske selekcije i pružiti usmjerenje pri izradi plana treninga namijenjena specifičnim potrebama pojedinca. Stoga je cilj ovoga istraživanja bio utvrditi postoje li dermatoglifske karakteristike koje se mogu povezati s RR, RX i XX ACTN3 genotipima, a koje se mogu upotrijebiti s ciljem izrade profila mišićne snage koja obećava visokouspješnu izvedbu u djece i adolescenata.

Metode

Dizajn istraživanja i sudionici

Ovo je bilo *ex post facto* deskriptivno i komparativno istraživanje. Obuhvaćalo je dječake između 7 i 17 godina starosti (N = 82) koji su trenirali vaterpolo u brazilskoj

saveznoj državi Paraíba. U skladu s prethodno opisanim klasifikacijama (Serhiyenko i Lishevskaya, 2010; Dantas, Alonso i Fernandes-Filho, 2004; Abramova, Nikitina i Chafranova, 1995), sudionici su, s obzirom na svoje dermatoglifske uzorke, svrstani u četiri skupine: anaerobna snaga (AP = 8), otpornost pri brzini (SR = 44), čista snaga (PF = 5) i aerobna otpornost (AR = 25). Čistom se snagom smatrao produkt sile koju tjelesni segment mora proizvesti kako bi postigao svoju maksimalnu brzinu, odnosno moć = snaga x brzina..

Sve su procedure provedene u skladu s regulacijama Nacionalnog zdravstvenog vijeća CNS 196/96. Procedure su također bile u skladu s uputama za istraživanja koja se provode s ljudima, s obzirom na protokol 369/10 Odbora za istraživačku etiku Lauro Wanderley sveučilišne bolnice pri UFPB. Roditelji i skrbnici sudionika upućeni su u procedure istraživanja te su zamoljeni da potpišu besplatan i pojašnjen obrazac u kojem su dali svoju suglasnost da njihova maloljetna djeca/štićenici sudjeluju u istraživanju.

Dermatoglifska analiza

Za skupljanje digitalnih otisaka upotrijebljen je Cross Match® Verifier 320 LC skener. Svaki prst ispitanika namješten je i pritisnut na skener, a zatim je vrh prsta zavaljan po skeneru s jedne na drugu stranu, krećući se uvijek u smjeru kažiprsta. Za analizu slika upotrijebili smo protokol koji su opisale Cummins i Midlo (1961), a prema kojem se tipovi uzoraka klasificiraju kao: „A" lukovi (bez delta uzorka), „L" petlje (delta uzorak), i/ili „W" spirale (uzorak s dvije delte). Nađeni su indeksi zbroja ukupne količine linija (STQL) i zbroj broja uzoraka prstiju (D10) pa je verificiran broj linija prstiju lijeve i desne ruke (LHSTQL i RHSTQL). Odjeljivanje tih dvaju čimbenika uzrokuje netočnu analizu dermatoglifskih podataka. Ta se metoda koristi s elitnim atletičarima tako što se povezuje karakter dermatoglifskih linija i somato-funkcionalne klasifikacije opisane u raznim studijama (Serhiyenko i Lishevskaya, 2010; Dantas i dr., 2004; Abramova i dr., 1995).

Minimalna vrijednost „0" i maksimalna vrijednost „10" dodijeljena je svakom tipu uzorka (A, L i W). Nakon što su utvrđene klasifikacije, izračunati su indeksi digitalnih otisaka koji su odgovarali STQL i D10 podatcima pa su te vrijednosti tretirane kao kvantitativne karakteristike. D10 podatcima pripisana je minimalna vrijednost „0" i maksimalna vrijednost „20", pri čemu je „0" označavalo prisutnost luka (A) na svim prstima i odsutnost delta uzorka. Vrijednosti D10 dobivene su upotrebom formule: $D10 = \Sigma L + 2(\Sigma W)$. Različiti prikazi klasifikacije digitalnih formula bili su: AL, AW, ALW, 10W, L>W, W>L, 10L, i L=W. Oni su definirani na sljedeći način: AL (prisutnost A i L u bilo kojoj kombinaciji); ALW (prisutnost A, L, i W u bilo kojoj kombinaciji); 10W (10 spirala); W>L (W i L s više od 5 W); L>W (L i W s više od 5 L); 10L (deset petlji); L = W (jednak broj L i W).

Izrada genotipa ostatka 577 ACTN3 gena

Skupljanje DNA

Genski je materijal prikupljen uporabom individualnih sterilnih pamučnih nastavaka na plastičnim štapićima s hidrofilnim pamukom na jednom kraju. Pamučni dio protrljan je o sluznicu unutrašnje strane obraza nakon čega je vraćen u svoj plastični omot i poslan u forenzički DNA laboratorij instituta Institute for Scientific Policing države Paraíba–SPI/PB/BR.

Izrada genotipa

Iz prikupljenih uzoraka izdvojeni su uzorci DNA, što je učinjeno u skladu s procedurama koje su dali Walsh i dr. (1991). PCR u stvarnom vremenu izveden je s pomoću IQ5 Thermal Cycler (Biorad) priborom za utvrđivanje R577X polimorfizma (Assay Id C_590093_1, Applied Biosystems) prema uputama proizvođača. Sve procedure prikupljanja podatakâ odvijale su se u laboratoriju UNIPÊ/SANNY Physical Evaluation Laboratory Sveučilišnog centra João Pessoa–UNIPÊ/PB/BR. Izrada genotipa ostatka 577 ACTN3 lokusa provedena je u forenzičnom laboratoriju instituta Scientific Policing države Paraíba (SPI/PB).

Statističke metode

Za interpretaciju podataka te s ciljem identifikacije stupnjeva povezanosti i razlika između podskupina sudionika upotrijebljena je inferencijalna statistika. Kolmogorov-Smirnov test upotrijebljen je kako bi se utvrdilo kakva je distribucija kvantitativnih podataka. Usporedba varijabli inicijalno je provedena uz pomoć jednosmjernih analiza varijanci (ANOVA). Scheffêov post hoc test je upotrijebljen nakon što su analize varijanci pokazale statistički značajne efekte. Korištena je $p < 0,05$ razina značajnosti. Statistička je analiza izrađena uz pomoć SPSS® statističkog softvera (inačica 14,0).

Rezultati

Dermatoglifska analiza

Tjelesne sposobnosti svakog sudionika pojedinačno, njihova anaerobna moć, otpornost pri brzini, čista snaga i anaerobna otpornost određene su u skladu s karakteristikama njihovih digitalnih otisaka utemeljenih na broju linija, tipu STQL digitalnih uzoraka i D10 indeksa (Tablica 1). Rezultati relativne frekvencije za formule i uzorke (Tablica 2) pokazali su da je AP skupina imala jasnu karakteristiku „ALW" digitalne formule, dok su PF skupine pokazale jasnu karakteristiku AL digitalne formule. SR skupina imala je frekvencijski odnos prema gotovo svim tipovima digitalnih formula, a AR skupina imala je potpunu odsutnost lukova i visokih frekvencija L>W i W>L.

Tablica 1. i 2.

Analiza krivulje disperzije rezultata pokazala je normalnu distribuciju u slučaju svih kvantitativnih varijabli (p vrijednosti < 0,05). Kao što je prikazano u Tablici 3, analize

varijance otkrile su značajne skupne efekte za D10 i STQL (obje *p* vrijednosti < 0,001) i značajne skupne efekte za LHSTQL i RHSTQL vrijednosti za svaku znamenku (*p* vrijednosti ≤ 0,01).

Tablica 3.

Izrada genotipa za ACTN3

AP i AR skupine okupile su isključivo sudionike s RR i RX genotipima, a većina su bili heterozigote (Tablica 4). U SR skupini bili su sudionici svih genotipa, od kojih je nešto više od jedne trećine imalo RR genotip, gotovo jedna trećina sudionika imala je RX genotipe, a otprilike jedna petina imala je XX genotip, koji ne podržava anaerobnu snagu. PF skupina nije imala gensku predispoziciju u smislu kvalitete snage jer su svi sudionici u toj skupini bili XX homozigote.

Tablica 4.

Odnos između dermatoglifske klasifikacije i ACTN3 genotipa

AP skupina, koja je obuhvaćala 9,76% ukupne kohorte, sadržavala je samo RR i RX genotipe. Oni su imali srednje vrijednosti za D10 (10) i STQL (94) dermatoglifskih indeksa kohorte, s prikazom svakog tipa uzorka: A(~1), L(~7), W(~2).

Nešto više od polovine (53,66%) sudionika ovoga istraživanja bili su svrstani u SR skupinu. SR skupina imala je srednje vrijednosti za D10 i STQL (~10 i ~102, tim redoslijedom) blizu srednje vrijednosti za skupinu s prikazom uzoraka A(~1), L(~8), W(~1), zajedno s glavnim digitalnim formulama AL, L>W i W>L. Malo više od dvije trećine sudionika u SR skupini bili su RX heterozigote, a blizu jedne trećine bili su RR homozigote. Ni jedan nije bio XX homozigota.

PF skupina obuhvaćala je 6,09% sudionika. Oni su imali prilično niske srednje vrijednosti D10 i STQL, (~7 i ~66, tim redoslijedom), s uzorcima A (3) i L (7) pri frekvencijama izvan AL digitalne formule i nisu sadržavali W. Svi sudionici PF skupine bili su XX homozigote.

Na kraju, 30,49% sudionika pripali su AR skupini. Oni su imali relativno visoke srednje vrijednosti D10 (~15) i STQL (~156) pa su imali L(~5) i W(~5) uzorke. Slično kao i SR skupina, nešto više od dvije trećine sudionika u AR skupini bili su RX heterozigote, a gotovo jedna trećina RR homozigote, što je dalo frekvenciju X alele od 34%.

Diskusija

U ovome smo radu istražili postoji li odnos između atletskih klasifikacija utemeljenih u dermatoglifskim karakteristikama i ACTN3 genotipa u R577X polimorfizmu. Pokazalo se da su svi sudionici AP skupine imali gensku predispoziciju za izvedbu za koju je potrebna anaerobna moć: svi su nosili barem jednu kopiju alele aktivnog proteina pa su njihovi dermatoglifski profili bili u skladu s predispozicijom za visoko

oslobađanje anaerobne moći. Slično AP skupini, sudionici u AR skupini nosili su barem jednu R577 alelu u ACTN3 genu, što je upućivalo na to da su imali gensku predispoziciju za anaerobnu mišićnu snagu. SR skupina je bila jedina skupina koja je okupila sudionike sa sva tri genotipa, iako je samo oko 20% njih imalo XX genotip. PF skupina bila je različita u tome što su svi sudionici u toj skupini imali XX genotip. PF skupina također je imala vidljiv nedostatak W u dermatoglifskim profilima.

Anaerobna snaga

Smatra se da pojedinci koji se uklapaju u AP dermatoglifsku klasifikaciju imaju predispoziciju za postizanje intenzivne anaerobne snage (Yang i dr., 2003; Abramova i dr., 1995). Odsustvo pojedinaca XX genotipa u našoj AP skupini u skladu je s ostalim istraživanjima koja pokazuju nedostatak izvedbe za aktivnosti koje ovise o anaerobnoj snazi među pojedincima s XX genotipom (Alfred i dr., 2011; Bray i dr., 2009; Montenegro, Barbosa, Dantas, Fernandes-Filho i Knackfuss, 2008; Cunha i dr., 2006; Dantas i dr., 2004; Abramova i dr., 1995). Primjerice, usporedivši skupinu trkača sprintera i kontrolnu skupinu sudionika koji nisu bili atletičari, Yang i dr. (2003) utvrdili su da su trkači imali višu frekvenciju RR genotipa od sudionika u kontrolnoj skupini (50% nasuprot 30%), i niže frekvencije RX (45% nasuprot 52%) te XX (6% nasuprot 18%) genotipa od sudionika kontrolne skupine. Papadimitriou i dr. (2008) usporedili su ACTN3 genotipe trkača sprintera s trkačima na duge staze i utvrdili da su sprinteri imali višu frekvenciju RR genotipa (73,53% nasuprot 50%) i nižu frekvenciju RX (17,65 nasuprot 25%) i XX (8,82% nasuprot 25%) genotipa. Takvi rezultati upućuju na to da je R alela ACTN3 gena povoljna za atletski uspjeh u fundamentalno anaerobnim oblicima sporta. Nadalje, frekvencije XX genotipa pronađene u sprintera (Eynon i dr., 2009; Yang i dr., 2007; Niemi i Majamaa, 2005) značajno su niže od onih koje se pronalaze u općoj populaciji. Primjerice, u istraživanju sa 154 zdrava pojedinca, uključujući afroamerikance, Clarkson i dr. (2005) pronašli su frekvenciju XX genotipa od 26%. S druge strane, s obzirom na to da su svi sudionici PF skupine u našoj kohorti imali XX genotip, može se zaključiti da imaju gensku limitaciju u smislu njihove sposobnosti da postignu mišićnu snagu zbog preranog signala za zaustavljanje u 577X polimorfizmu u oba njihova ACTN3 gena (Eynon i dr., 2009; Roth i dr., 2008).

Izdržljivost

Dok pojedinci XX genotipa možda nisu dobri kandidati za sportove u kojima je potrebna anaerobna moć, postoje dokazi o tome da imaju potrebne sposobnosti za sportove u kojima je potrebna izdržljivost. Eynon i dr. (2009) i Papadimitrou i dr. (2008), koji opisuju R alelu ACTN3 gena kao pogodnu za sportove u kojima je potrebno brzo trčanje (sprint), također opisuju X alelu i XX genotip kao podobnu za aktivnosti u kojima je potrebna izdržljivost. U skladu s navedenim, Yang i dr. (2003) pronašli su da je XX genotip češći u atletičara koji trče na duge staze (24%) nego

u kontrolnih sudionika koji nisu atletičari. U istraživanju koje se bavilo različitim tipovima finskih atletičara, Niemi i dr. (2005) također su pronašli da su atletičari na duge staze rjeđe imali RR genotip, a češće XX genotip od atletičara sprintera. U isto vrijeme, atletičari sprinteri nisu imali ni jednog člana s XX genotipom. S druge strane, u istraživanju koje se bavilo odnosom između ACTN3 genotipa i atletske izvedbe 992 grčka adolescenta, Moran i dr. (2005) nisu pronašli dokaze o tome da je činjenica da su ispitanici imali X alelu bila povezana s atletskom otpornošću. Stoga je potrbno provesti još istraživanja kako bi se utvrdilo je li i do koje mjere posjedovanje X alele povezano s dobrim predispozicijama u atletičara koji se bave sportovima s visokim zahtjevima aerobne otpornosti.

Čista snaga

Iako je XX genotip bio karakterističan za sve članove PF skupine u ovome istraživanju, odnos između čiste snage i ACTN3 gena još nije jasan. Čista je snaga vezana uz sposobnost pojedinca da realizira maksimalnu dinamičku hotimičnu kontrakciju mišića potpomognutu unutar i međumišićnom koordinacijom. McCauley i dr. (2010) proučavali su mišićnu funkciju i mišićnu strukturu muškaraca koji nisu atletičari pa su utvrdili da ACTN3 polimorfizam nije povezan s kontraktivnim karakteristikama, ni apsolutno, ni u smislu relativne dinamičke snage mišića koljena. Roth i dr. (2008) utvrdili su da su body builderi / dizači utega, koji od svojega tijela zahtijevaju čistu snagu, imali višu frekvenciju RX genotipa (62,6% nasuprot 45,6%), ali niže frekvencije *oba* RR genotipa (30,7% nasuprot 38,1%) i XX genotipa (6,7% nasuprot 16,3%).

Zaključak

Pokazalo se da je dermatoglifija racionalan pristup R577X polimorfizmu ACTN3 gena s ciljem predviđanja profila anaerobne mišićne mase. Kao dopuna izradi genotipa ACTN3 gena, dermatoglifska analiza ima velik potencijal u sportskoj selekciji, osobito u smislu označavanja pojedinaca koji imaju gensku predispoziciju za anaerobnu mišićnu moć nasuprot pojedincima koji nemaju takvu predispoziciju. Na kraju, rezultati potvrđuju važnost upotrebe tih dviju metoda kako bi se postigla koherentnost i komplementarnost u evaluaciji kvalitete tjelesnih karakteristika danog uzorka populacije.