

## INFLUENCE OF GENETIC VARIATIONS IN PHYSICAL FITNESS

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Older population has increased and presents reduced physical fitness. On the other hand, it is known that genetic variants can explain these declines. The aim of this study was evaluate the effect of ACE I/D and ACTN3 R/X polymorphisms in body fat and physical capacities in old women. 66 old women were genotyped to ACTN3 and ACE polymorphisms and submitted to anthropometric measurements, physical tests, IPAQ and a questionnaire related to food intake. For the ACTN3 genotype, XX group presented reduced levels of upper limb flexibility and cardiorespiratory capacity. Related to ACE genotype, ID group demonstrated increased cardiorespiratory capacity and decreased body fat percentage. There was no statistical difference in the other variables. In the future, data like these will enable each person have his/her intervention focused in that variables with more genetic potential to decline.

**KEY WORDS:** aging, genetic polymorphisms, strength.

**INTRODUCTION:** According to 2000 Census data, the Brazilian population was composed of 14,5 millions of elderly and in 2010 this number increased to 18 millions. Old age is a phase of decline in physiological measurements and associated to decreased functional performance after a fast improvement during the childhood and a maximum between the end of adolescence and 30 years of age (Martin & Preston, 1994). These hallmarks of aging result of genetic and environmental factors, as physical activities practice and food habits (Gottlie et al., 2007). It is also known that decreased physical fitness and increased body fat may result in decreased physical activity practice, social isolation, increased levels of falls, chronic degenerative diseases and enlarged public health spending (Gomes, 2012).

On the other hand, genetic polymorphisms are variations in a DNA sequence that affect more than 1% of the population (Matos, 2005) and angiotensin-converting enzyme (ACE) and alpha-actinin 3 genes polymorphisms potentially may influence physical fitness in elderly. ACE has a vital role in the renin angiotensin system, converting angiotensin I to angiotensin II, which acts in vasoconstrictor function, cell proliferation, cardiac hypertrophy and aldosterone secretion from adrenals glands (Abdollahi et al., 2005). The possible genetic variations associated to ACE are insertion (I) or deletion (D) alleles and are related to levels of enzyme activity (Shulman, 2000). ACTN3, on the other hand, acts in Z line of the muscle cells, helping in the actin filaments anchorage (Yang et al., 2003). Polymorphisms in its gene have been associated with muscle function, power and strength (Paparini et al., 2007) and the possible variations of the ACNT3 polymorphism are R (functional protein) or X (stop codon) alleles. Studies related to this area are scarce and important to help in the prediction of physical capacities that will decrease in each person, enabling the adoption of strategies to slow down these declines. In this context, the aim of this study was to evaluate the effects of ACE I/D and ACTN3 R/X genetic polymorphisms in body fat, muscle strength, aerobic capacity, flexibility and agility in old woman.

**METHODS:** 66 old women from Ribeirao Preto (Sao Paulo) assigned an independent and informed term of consent and were submitted to health exams to exclude people who presented health problems that could preclude to perform physical tests.

To ACE I/D and ACTN3 R/X genotyping blood was collected from the antecubital fossa by venipuncture and the DNA was extracted through phenol-chloroform method. After that step the sequences of interest were amplified by PCR (polymerase chain reaction). Finally, agarose gels (1,5% and 3%) were submitted to electrophoresis and colored with SYBR

green. In the ACTN3 genotyping was also added the Dde I restriction enzyme before the electrophoresis step (Rodriguez-Romo et al., 2010).

In addition to basics anthropometric measurements (body mass and stature), body composition were analyzed by bioimpedance (Maltron BF-906). Six minutes walking test, chair sit and reach test, back scratch test, arm curl test and AAHPERD agility test (Osness et al., 1990; Rikli & Jones, 2008) were performed in order to measure physical capacities. IPAQ to measure physical activity level and the Food Consumption Markers Questionnaire for analysis of consumption of 10 food groups during a week were also used.

Related to statistical analysis, t Student test for independent samples was performed to compare the groups related to ACTN3 gene and one-way analysis of variance (ANOVA) for ACE genotyping - when appropriated, the Tukey's post-hoc test was done. The significance level adopted was 5%.

**RESULTS/DISCUSSION:** In Table 1 no differences were observed for the ACTN3 and ACE genes among the groups related to age, stature, body mass and body mass index. However, the ID group presented lower values for body fat when compared with the other two groups.

**Table 1**  
**Sample characterization. Data are presented as mean ± standard error of mean and the sample number for each group in parentheses. BMI: body composition; BM: body mass index. \*: p<0.05 versus the others two groups.**

Groups (n)	ACTN3		ACE		
	XX (n)	RR/RX (n)	DD (n)	ID (n)	II (n)
Age (years)	63,9 ± 2,9 (13)	60,1 ± 1,1 (52)	63,2 ± 1,9 (26)	61,2 ± 1,8 (23)	59,2 ± 1,9 (16)
Stature (m)	1,60 ± 0,01 (13)	1,60 ± 0,01 (52)	1,59 ± 0,01 (26)	1,60 ± 0,01 (23)	1,60 ± 0,02 (16)
BM (kg)	72,7 ± 5,5 (13)	70,4 ± 1,2 (52)	69,6 ± 1,9 (26)	67,3 ± 2,8 (23)	78,2 ± 5,8 (16)
BMI (kg/m <sup>2</sup> )	28,3 ± 1,9 (13)	27,5 ± 0,7 (51)	27,4 ± 0,6 (26)	26,0 ± 1 (23)	30,0 ± 2 (16)
Body fat (%)	31,8 ± 3,7 (12)	31,3 ± 1,4 (51)	33,4 ± 1,5 (25)	26,6 ± 2,2* (23)	35,6 ± 3,1 (15)

In Table 2 is possible to observe that XX group presented lower performance in the six minutes walking test and in the back scratch test. For the ACTN3 gene the ID group presented a better performance than II group in the six minutes walking test.

**Table 2**  
**Physical capacities. Data are presented as mean  $\pm$  standard error of mean and the sample number for each group in parentheses. \* $p < 0.05$ ; # $p < 0.05$  versus II.**

Groups (n)	ACTN3		ACE		
	XX (n)	RR/RX (n)	DD (n)	ID (n)	II (n)
Agility (seg)	24,7 $\pm$ 3,3 (12)	23 $\pm$ 1,2 (51)	20,8 $\pm$ 2,1 (51)	22,8 $\pm$ 1,2 (23)	26 $\pm$ 2,9 (15)
Sit and reach (cm)	-1,5 $\pm$ 3,2 (13)	1,2 $\pm$ 1,2 (51)	1 $\pm$ 1,6 (24)	0,6 $\pm$ 1,9 (23)	0,3 $\pm$ 2,8 (15)
6 min (m)	502 $\pm$ 31 (12)	565 $\pm$ 11 * (50)	541 $\pm$ 13 (25)	592 $\pm$ 18 # (23)	509 $\pm$ 25 (14)
Back scratch (cm)	-13,9 $\pm$ 5,6 (11)	-3,3 $\pm$ 1,5 * (51)	-4,4 $\pm$ 2,1 (24)	1,8 $\pm$ 2,4 (23)	11,6 $\pm$ 4,5 (15)
Arm curl (repetitions)	17,4 $\pm$ 1,3 (12)	19,6 $\pm$ 0,7 (52)	18,4 $\pm$ 0,8 (26)	20,4 $\pm$ 1,2 (23)	18,7 $\pm$ 1,2 (15)

The different groups no showed difference in terms of physical activity level and only one variation of the ten food groups. Then, it is possible to suppose that the differences in body composition, flexibility levels and in cardiorespiratory capacity are associated to genetic variations and not to environmental factors.

Data from a study of Lemes et al. (2013) with Brazilian children and teens showed that DD and ID have lower body fat when compared to II group. However, Passaro et al. (2011) and Bonnet et al. (2008), studying European population, did not demonstrate differences for body fat in any group related to ACE gene. These data show the importance of multicenter studies with different populations and age ranges.

It was demonstrated that groups presented similar results in the strength test. This reinforces importance of studies with older people because Ahmetov et al. (2014) performed a study with 209 juvenile Russian athletes, in which testosterone levels were increased in R-allele-carriers compared to XX genotype. Additionally, Norman et al. (2014) concluded that X allele prevalence is reduced among strength/speed athletes, suggesting that ACTN3 is crucial to these capacities in young adults.

Related to other physical fitness variables, the ID group presented better aerobic capacity when compared to II group and this result is not found in other studies from literature. Almeida et al. (2012) and Nazarov et al. (2001) showed better aerobic capacity associated to the II genotype for physically active young people and athletes, respectively. Finally, RR/RX ACTN3 genotype presented better aerobic capacity and flexibility of upper limbs. Pimenta et al. (2013) demonstrated better aerobic capacity associated to X allele. On the other hand, there is no study related to flexibility. Differences in physical tests performed, physical activity level and age may help to explain these different results, suggesting that these

polymorphisms and gene expression related to them can be regulated according to level of physical activity and age.

**CONCLUSION:** Old women with the XX ACTN3 genotype was associated to lower levels of flexibility in the upper members and cardiorespiratory capacity. On the other hand, ID ACE genotype resulted in better cardiorespiratory capacity and lower body fat percentage. Related the professional practice, people that present genetic characteristics that predispose to lower results in any physical capacity should try to improve it before to reach old age.

#### REFERENCES:

- Abdollahi, M.R., Gaunt, R.T., Syddall, H.E., et al. (2005). Angiotensin II type I receptor gene polymorphism: anthropometric and metabolic syndrome traits. *Journal of Medical Genetics*, 42, 396-401.
- Ahmetov, I.I., Donnikov, A.E.; Trofimov, D.Y. (2014). ACTN3 genotype is associated with testosterone levels of athletes. *Biology of Sport*, 31(2), 105-8.
- Almeida, J.A., Boulosa, D.A., Pardono, E., et al. (2012). A influência do genótipo e da ECA sobre a aptidão cardiovascular de jovens do sexo masculino moderadamente ativos. *Arquivos Brasileiros de Cardiologia*, 98, 315-20.
- Bonnet, F., Patel, S., Laville, M., et al. (2008). Influence of the ACE gene insertion/deletion polymorphism on insulin sensitivity and impaired glucose tolerance in healthy subjects. *Diabetes Care*, 31, 789-94.
- Gomes, M. Influência da força e da potência muscular no controle postural de idosas de diferentes faixas etárias. 2012. Dissertação (Mestrado) - Universidade de São Paulo, Ribeirão Preto, 2012.
- Gottlie, M.G.V., Carvalho, D., Schneider, R.H., et al. (2007). Aspectos genéticos do envelhecimento e doenças associadas: uma complexa rede de interações entre genes e ambiente. *Revista Brasileira de Geriatria e Gerontologia*, 10(3), 273-83.
- Lemes, V.A., Neves, A.L., Guazzelli, I.C., et al. (2013). Angiotensin converting enzyme insertion/deletion polymorphism is associated with increased adiposity and blood pressure in obese children and adolescents. *Gene*, 532, 197-202.
- Martin, L. & Preston, S. (1994). *Demography of aging*. Washington: National Academic Press.
- Matos, M.F.D. Polimorfismos dos genes da ECA, angiotensinogênio e do receptor tipo 1 da angiotensina II e pressão arterial. 2005. 145 f. Tese (Doutorado em Cardiologia) - Faculdade de Medicina, Universidade Federal do Rio de Janeiro, Rio de Janeiro, 2005.
- Norman, B., Esbjornsson, M., Rundqvist, H., et al. (2014). ACTN3 genotype and modulation of skeletal muscle response to exercise in human subjects. *Journal of Applied Physiology*, 116, 1197-203.
- Nazarov, I.B., Woods, D.R., Montgomery, H.E., et al. (2001). The angiotensin converting enzyme I/D polymorphism in Russian athletes. *European Journal of Human Genetics*, 9, 797-801.
- Osness, W.H., Adrian, M., Clark, B., et al. (1990). *Functional fitness assessment for adults over 60 years*. Reston: American Alliance for Health, Physical Education, Recreation and Dance.
- Paparini, A., Ripani, M., Giordano, G.G., et al. (2007) ACTN3 genotyping by real-time PCR in the Italian population and athletes. *Medicine & Science in Sports & Exercise*, 39(5), 810-5.
- Rikli, R.E., Jones, C.J. (2008). *Teste de aptidão física para idosos*. 1<sup>o</sup> Ed. Barueri: Manole.
- Passaro, A., Nora, E.D., Marcello, C., et al. (2011). PPAR $\gamma$  Pro12Ala and ACE ID polymorphisms are associated with fat distribution, but not metabolic syndrome. *Cardiovascular Diabetology*, 10, 112.
- Pimenta, E.M., Coelho, D.B., Veneroso, C.E., et al. (2013). Effect of ACTN3 gene on strength and endurance in soccer players. *The Journal of Strength & Conditioning Research*, 27, 3286-92.
- Rodriguez-Romo, G., Ruiz, J.R., Santiago, C., et al. (2010). Does the ACE I/D polymorphism, alone or in combination with the ACTN3 R577X polymorphism, influence muscle power phenotypes in young, non-athletic adults? *European Journal of Applied Physiology*, 110, 1099-106.
- Shulman, G.I. (2000). Cellular mechanisms of insulin resistance. *Journal of Clinical Investigation*, 106(2), 171-6.
- Yang, N., McArthur, D.G., Gulbin, J.P., et al. (2003). ACTN3 genotype is associated with human elite athletic performance. *American Journal of Human Genetics*, 73, 627-31.