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CARDIOVASCULAR AND MUSCULAR FITNESS IN OVERWEIGHT ADULTS: EFFECTS OF ACTN3 GENOTYPE AND SEX

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INTRODUCTION:

The α -actinin-3, encoded by the ACTN3 gene, is an actin cross-linking protein expressed in type II muscle fibres (MacArthur et al., 2007). A common loss-of-function polymorphism (R577X) results in the 577XX genotype, which is widespread globally (North et al., 1999). The ACTN3 R577X polymorphism has been repeatedly tested for associations with sports performance status and much less so for fitness of the general population. α -actinin-3 expressing genotype (RR genotype) has initially been linked to muscle power, whereas α -actinin-3 deficient genotype (XX genotype) has been associated with superior endurance performance (Yang et al., 2003). However, the role of ACTN3 R577X polymorphism in endurance exercise performance remains controversial, and the association with physical fitness of non-athletic overweight populations is still unclear.

The aim of the present study was to compare muscular and cardiovascular fitness between overweight, untrained men and women with the XX vs. RR genotypes. It was hypothesized that individuals with the XX genotype exhibit lower muscular and cardiovascular fitness than RR subjects.

METHODS:

Adults with a BMI of 25–35 kg·m⁻² were genotyped for ACTN3 and assigned to four groups: XX men, XX women, RR men, and RR women. Each group comprised 19–21 participants matched for age (43 ± 7 years). Knee extensor and flexor torques were assessed using isokinetic dynamometry. Maximal oxygen uptake (VO₂ max) and other cardiopulmonary and metabolic parameters were determined during an incremental cycling test, using continuous gas exchange analysis combined with lactate and heart rate monitoring. Genotype- and sex-related differences in muscular and cardiovascular fitness were compared.

RESULTS:

Peak muscle torque did not differ between the RR and XX groups in either sex, but was lower in women than in men, as expected ($p > 0.05$ for genotype effect; $p < 0.001$ for sex effect). No genotype-related differences were observed in either sex for any of the measured maximal variables, including absolute and body mass–relative VO₂ max, peak power output, pulmonary ventilation, O₂ pulse, fat oxidation, heart rate, and blood lactate ($p > 0.05$ for genotype effect in all cases). Most of these variables were higher in men than in women, except for maximal breathing frequency, maximal fat oxidation, maximal heart rate, and peak lactate, which did not differ between sexes ($p > 0.05$ for sex effect).

CONCLUSION:

In overweight, untrained adults, ACTN3 genotype was not associated with differences in muscular strength or cardiovascular and metabolic exercise responses. As expected, clear sex-related differences were observed, with men generally exhibiting higher performance and physiological values than women.

REFERENCES:

- 1) MacArthur et al., Nat Genet., 2007
- 2) North et al., Nat Genet., 1999
- 3) Yang et al., Am J Hum Genet., 2003

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