

ACTN3 R577X Polymorphism in Asian Indian Athletes

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ABSTRACT Replacement of Arginine (R) by a premature stop codon (X) (R577X, rs1815739) in Alpha-actinin 3 gene (*ACTN3*) has been commonly reported. The R allele has been hypothesized to be associated with power-based performance, while the X allele may be linked with endurance abilities. To evaluate if this theory holds true in Indian subjects, we studied the *ACTN3* genotype distribution in 305 Asian Indians, comprising of 155 athletes and 150 normal sedentary individuals. We did not observe any significant difference in the allelic frequencies between athletes ($R/X=0.42/0.58$) and normal subjects ($R/X=0.39/0.61$). But, while categorizing the athletes on the basis of their competitive level (National/International vs. Regional); the genotype distribution pattern became more prominent ($\chi^2 = 4.9$, d.f=2, $p=0.08$), though not statistically significant. Additionally, in the National/International athletes, R allele was seen to be associated with power based performance ($\chi^2 = 6.6$, d.f=1, $p<0.01$). In the same group, a high frequency of X allele was observed with endurance performance ($\chi^2 = 3.14$, d.f=1, $p=0.07$), but fell short of significance. Thus, XX genotype could be an important factor in determining endurance performance. However, the study needs to be expanded to a larger cohort of National/International athletes. To our information, this is the first Indian study on *ACTN3* genotype and its association with sporting performance.