ACTN3 R577X genotype influences 100 and 200 m Olympic sprinting performance in seven cohorts of elite sprinters

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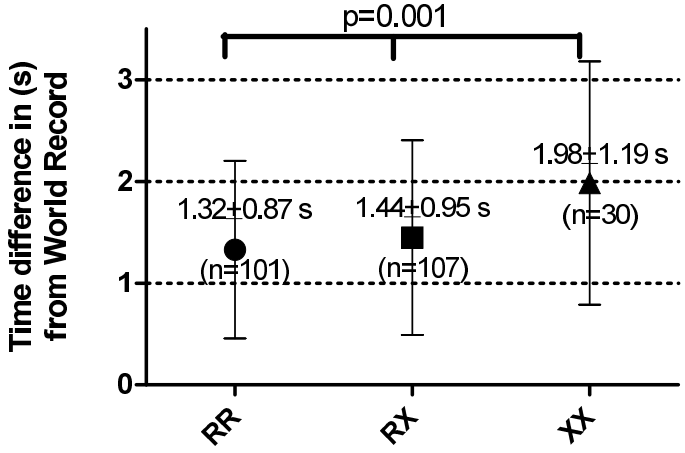
Deficiency of α-actinin-3 protein, as a consequence of the *ACTN3* 577XX genotype, has repeatedly been associated with elite athletic performance (Yang *et al.*, 2003; Niemi & Majamaa, 2005; Papadimitriou *et al.*, 2008; Eynon *et al.*, 2009,2012; Mikami *et al.*, 2014). These association studies suggest that α-actinin-3 deficiency is detrimental to optimal fast muscle function at the extremes of sprint and power performance, possibly through a shift in the physiological and metabolic properties of "fast" glycolytic muscle fibres - as shown in mice (MacArthur *et al.*, 2007,2008). However, these association studies in elite athletes have been limited by small sample size, the inclusion of sprint and power athletes from mixed sport disciplines, and a lack of quantitative measure of performance.

Aim: Therefore, the aim of the present study was to investigate the association between the *ACTN3* R577X polymorphism and 100 m and 200 m personal best times using a new quantitative collaborative approach involving seven cohorts of elite sprinters.

Methods: A total of 238 personal best 100 m and 200 m sprint times for 144 (90 male and 54 female) elite sprinters from Greece, Italy, Poland, Lithuania, Russia, Spain and Brazil were analysed. The sprinters' best personal sprint times, grouped according to sex and event (100 or 200 m), were expressed relative to the relevant current world record. Genotyping of the sprinters was performed using PCR. To compare the sprinters' best times between all genotypes, and the gap between personal best sprint time and the relevant world record, we used one-way Analysis of Variance (ANOVA). To compare between the *ACTN3* 577RR genotype and the 577XX genotype (α-actinin-3 deficiency; hypothesized to be detrimental for sprinters), we used a parametric unpaired two-tailed t-test.

Results: On average, Caucasian sprinters with the *ACTN3* 577RR genotype had faster best 100 m sprint times than their *ACTN3* 577XX counterparts (P=0.02), and only one male, but no female, sprinter with the 577XX genotype had a best time faster than the 2012 London Olympics qualifying time (Table). Male 200 m sprinters with the *ACTN3* 577RR genotype ran faster than their 577XX counterparts (P=0.01), and there were no *ACTN3* 577XX male 200 m sprinters with a best time faster than the 2012 Olympic qualifying time (Table). Furthermore, the best sprint times for both males and females with the *ACTN3* 577RR genotype were closer to the relevant world record than their 577RX and 577XX counterparts (P=0.001) (Figure).



The 100-m and 200-m best sprint times (mean ± SD) according to ACTN3 *genotype distribution and sprinters' ancestry.*

	African Ancestry (n=63)			**Caucasians** (n=175)		
	RR (n=32)	**RX** (n=25)	**XX** (n=6)	**RR** (n=69)	**RX** (n=82)	**XX** (n=24)
100m Male	10.41±0.28 (n=12)	10.33±0.28 (n=7)	10.97±0.00 (n=1)	10.57±0.29* (n=23)	10.67±0.35 (n=31)	10.84±0.31 (n=9)
200m Male	21.10±0.72 (n=9)	20.92±0.67 (n=6)	22.58±0.00 (n=1)	21.39±0.69** (n=20)	21.85±0.86 (n=22)	22.22±0.88 (n=7)
100m Female	11.59±0.41 (n=7)	11.47±0.37 (n=7)	11.68±0.47 (n=2)	11.97±0.42 (n=15)	12.01±0.46 (n=15)	12.11±0.35 (n=3)
200m Female	23.75±0.96 (n=4)	22.94±0.40 (n=5)	23.59±1.09 (n=2)	24.26±0.90 (n=11)	24.38±1.09 (n=14)	25.41±0.57 (n=5)

*P=0.02; **P=0.01

Conclusion: Our large, multi-centre study has enabled us to demonstrate that it is unlikely for athletes with α-actinin-3 deficiency (577XX) to achieve speeds required for victory in Olympic sprint events. This finding could have important applications for identifying and coaching talented 100 and 200 m sprinters.

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