S-I-1 Athletic Performance and Genetics in Skeletal Muscles

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At the end of July in 2012, the 30th Olympic Games will be held in London, UK. Then, the superior results by athletes will depend on not only their practice but also their genetic nature of birth. The genetic research has been advanced up for medical treatments like the muscular dystrophy due to the dystrophin loss so far, but recently people paid attention to the relation between the gene polymorphism and the diseases as well as the athletic performance. Particularly, a single nucleotide polymorphism (SNP) that especially one base replaces other bases has been analyzed often with the technique detecting gene polymorphism as a marker of an individual genetic background.

A muscle fiber type composition in skeletal muscles is the most important factor for the exercise performance. It shows a clear relation to the sprint performance, the endurance capacity, and the physical power. We will discuss on the fiber types and the polymorphisms of alpha- actinin-3 (ACTN3) that is topic in sprint performance and angiotensinconverting enzyme (ACE) and mitochondrial DNA (mtDNA) related with endurance capacity. The athletic performance might be explained by the gene polymorphisms.

Keywords: SNPs, ACTN3, ACE and mtDNA

S-I-2 ACTN3 Gene and Skeletal Muscle

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The alpha-actinin is actin-binding protein. In skeletal muscle, alpha-actinins-2 (ACTN2) and alpha-actinins-3 (ACTN3) isoforms are playing an important role as structural component of the Z-lines that anchor the actin-containing thin filaments and maintain the spatial between myofilaments. ACTN3 relationship is expressed only in type II muscle fibers while the ACTN2 is found in all muscle fibers. A common nonsense mutation in ACTN3 (R577X) results in ACTN3 deficiency that appears to be non-pathogenic and it is postulated that ACTN2 compensates for the deficiency. Therefore, it is hypothesized that ACTN3 genotype (RR, RX, XX) may influence variation in muscle function in humans and many studies have confirmed that the presence of the ACTN3 protein, or RR genotype is associated with greater success in activities requiring sprint or power performance. In contrast to the findings in power athletes, it is controversial that a lack of ACTN3 protein, or XX genotype is beneficial to endurance athletes. And associations of ACTN3 genotype with trainability, exercise-induced muscle damage and age-related changes of muscular function have been studied. However, these results are different among the studies because of differences of human race, age, sex, and physical activity level and so on. To clarify roles of ACTN3 genotype on skeletal muscle functions and their mechanisms, further studies with genetically modified animal models would be useful.

Key words: athletic performance, adaptation, KO mouse