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Collagen type I alpha 1 (COL1A1) rs1800012 polymorphism in cyclists

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Abstract

Purpose: We suggested that polymorphic differences affect the performance of cyclists. In this study we designed the experiment to identify collagen type I alpha 1 (COL1A1) rs1800012 polymorphism in cyclists.

Methods: We isolated genomic DNA from buccal epithelial cells of 38 cyclists. We followed COL1A1 rs1800012 single nucleotide polymorphism genotyping technology protocol. We reported allele frequencies for rs1800012 gene variants of cyclists in our kohort.

Results: Our results indicated the differences of the distribution of COL1A1 rs1800012 SNP between cyclists. The numbers and percentages of CC, AC and AA genotypes for rs1800012 polymorphism that we obtained were 24 (63%), 13 (34%) and 1 (3%), respectively. The distributions of C and A alleles were counted as 61 (80%) and 15 (20%).

Conclusions: There was difference in the genotype and also allele distribution between cyclists. Carriers of a C allele as compared to carriers of the allele A are more prone to cyclism. Larger homogenous cohorts may help clarify the association between COL1A1 rs1800012 polymorphism and physical performance.

Keywords: collagen type I alpha I, polymorphism, cycling, allelic distribution

INTRODUCTION

Fibroblasts of ligament generate a amount of extracellular matrix (ECM) components, including fibromodulin, decorin and collagen types I and III (Nordin et al., 2001).Decorin and fibromoduline are two abundant proteoglycans in the ligament. These proteoglycans bind type I collagen and control the fibrillogenesis of collagen. Especially, type 1, collagen is the most abundant form of collagen protein in the tendon tissue (Chamberlain et al., 2011). Collagen type I is a triple helical protein consisting of two alpha chains and one alpha 2 chain. The gene that encodes for the alpha 1 chain of type I collagen (COL1A1) is located on chromosome 17q21 and is indicative of a relationship between tendons and ligaments (Wang et al., 2017).

It has been shown that mutations in this gene lead to connective tissue problems like imperfected osteogenesis (Marini and Blissett, 2013), osteoarthritis, systemic diseases with scleral thinning and myopia. Collagen were

associated also with the variable response to exercise-induced muscle damage possibly by affecting RNA stability and hence collagen abundance.

Single nucleotide polymorphisms (SNPs), occuring in the COL1A1 gene region, may alter the the COL1A1 expression and affect the functions of COL1A1. Among various polymorphisms within the COL1A1 gene, the most frequently studied polymorphism has been the +1245G/T polymorphism (rs1800012, Sp1), described originially in 1996. COL1A1 polymorphism has also been related soft tissue injuries such as Achilles tendon rupture, anterior cruciate ligament (ACL) rupture and shoulder dislocation (Collins et al., 2010).

Aim of this study is to understand the distribution of COL1A1 rs1800012 polymorphism in Turkish cyclists. We hypothesized the dominance of C allele in cyclist for better performance. Therefore, this first report including Turkish athletes will help us to understand the distribution of COL1A1 rs1800012 polymorphsim.

METHOD

Study subjects

A total of 38 male cyclists were recruited for this study. We have especially selected professionals who fulfill the following requirements for this research: (1) each cyclist had a professional cycling team registered at one moment, (2) had any symptoms of cardiovascular or infectious diseases and diabetes, (3) had any genetically transmitted diseases (4) had any hereditary diseases. The Uskudar University Natural Sciences Enstitute Ethics Committee approved the study and written informed consent was obtained from each participant.

COL1A1 genotypes

Genomic DNA was extracted from oral epithelium by using commercially available Invitrogen DNA Isolation Kit (Invitrogen, USA) according to the manufacturer's protocol. A mean total of 20 ng of DNA were isolated from each samples, and purify of the isolates were checked by OD 260/280 spectrophotometric ratios. Genotypes of COL1A1 gene single nucleotide polymorphisms (rs1800012) were performed and all samples were genotyped using real-time PCR (RT-PCR) method (Thermofisher Quantstudio 3, USA) and Taqman Genotyping Assays (Catalog no: 4362691 Thermofisher, USA), including primers and fluorescently labelled (FAM and VIC) MBG probes for the detection of alleles. The 10 µl reaction volume included 5 µl Genotyping Master Mix (Applied Biosystems, Foster City, CA), 0.5 µl genotyping assay (Applied Biosystems), 3.5 µl nuclease-free H_2O (Thermofisher, USA) and 1 µl DNA. Thermal cycler conditions were as follows: denaturation at 92°C for 15 s and followed by anneal/extend at 60℃ for 1 min (40 cycles).

RESULTS

To determine the genotype distribitution we made the calculation CC, AC and AA. In the cyclists the numbers and percentages of CC, AC and AA genotypes for rs1800012 polymorphism were 24 (63%), 13 (34%) and 1 (3%), respectively. The distributions of C and A alleles were as 61 (80%) and 15 (20%).

DISCUSSION and CONCLUSION

Genetic background assignes significant features such as endurance, strength, muscle coordination and motivation which is crucial for sports performance (Ulucan et al., 2014). Genetics of sport examinations include a thorough inquiry of genes with an impact on athletic performance. The development of suitable training and sustenance programmes, depending on the genetic resources, is increasingly important for improving athletic efficiency, both in individual and team sports (Ulucan, 2016).

The recent growth number of genetics experiments expands the knowledge about the individual genes in human metabolic process which can be used identification of tendency of sportive performance and protection from injuries associated with the sports activity such as cycling, skiing and tennis. Definite assessment of performance efficiency genotypes and sport-related risk for acute or chronic diseases are likely to enable changes in training plans to reduce the risk of injury significantly and to optimize performance (Slodkowska et al., 2013).

The studies including human subjects and COL1A1 polymorphism in sport predisposition are not sufficient. We intended to obtain data about effects of COL1A1 Sp1 polymorphism (rs 1800012) on performance in Turkish cyclists study cohort. We examined 38 young cyclists. Analysis revealed us that 24 of the cyclists were CC and 1 of the players had AA genotype. 13 of the cyclists were AC, which is regarded as intermediate genotype. C allele was found to be superior to A allele for examined polymorphism.

In order to gain insight, study groups are seeking to understand in more detail the relations between the sports performance, injuries and gene polymorphisms. Ficek et al. (2013) examined COL1A1-1997G/T (rs1107946) and COL1A1 Sp1+1245G/T (rs1800012) polymorphims in professional soccer players. They found that higher COL1A1 G-T haplotype was correlated with lower danger of preceding risk of anterior cruaciate ligament injury. Studies have also shown that a significantly lower risk for cruciate ligament rupture was recorded as the higher frequency distribution of the COL1A1 +1245TT genotype in South African and Swedish players (Posthumus et al., 2009; Khoschnau et al., 2008).

Previous meta analysis in 2016, Chen et al. reported that COL1A1 G-T haplotype can provide ACL protection against injury (Chen et al., 2016). In addition to these results, Slodkowska et al. investigated COL1A1 rs1800012 polymorphism in Polish skiers. They identified that carriers of minor allele G had lower risk of ACL rupture (Slodkowska et al., 2013). In contrast to these studies Erduran et al. (2014) found no association between rs1800012 polymorphism and tennis elbow in their study group (Erduran et al., 2014). Our results are in agreement with the previous ones, indicating the dominancy of CC genotypes and C alleles in the examined cohort. In our study we found that COL1A1 C-allele carriers (rs1800012) are major and A-allele carriers are minor in professional cyclists cohort study.

Further work is required to validate the results in a larger independent cohort and investigate SNPs of other genes to precisely map the causal variants and genes influencing the phenotypic outcome. However, inclination of sportive activities is complex, multifactorial and may be polygenic trait. We should also consider other factors such as environmental subjects (stress, diet, smoking, alcohol, lifestyle) and epigenetic patterns. With all point of view, genetic polymorphism experiments may leads the generation of new genetic tests to determine appropriate types of sport for individuals.

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