Genetic polymorphisms in alpha-actinin 3 and adrenoceptor beta genes in Austrian elite athletes and healthy controls

Wessner B¹, Stuparits P¹, Fail C¹, Pavic F¹, Tschan H¹, Bachl N¹,²
¹ Centre for Sport Science and University Sports, University of Vienna, Austria
² Austrian Institute of Sports Medicine, Vienna, Austria

Abstract

Background: During the last years many studies were conducted to investigate the contribution of various genetic variants to endurance and sprint/power performance of elite athletes. Data on team sport athletes are missing to a large extent. Therefore, the aim of the study was to investigate the frequency of selected polymorphisms on elite athlete status in Austrian team sport (handball, soccer), endurance and power athletes as well as in healthy control subjects.

Methods: Genotype and allele frequencies of ACTN3 R577X (rs1815739), ADRB1 Ser49Gly (rs1801252), ADRB2 Arg16Gly (rs1042713), ADRB2 Gln27Glu (rs1042714) and the ADRB3 Trp64Arg (rs4994) polymorphisms were determined in 56 Austrian sprint/power athletes, 86 endurance athletes, 143 team sport athletes and 216 healthy non-athletic controls. Genomic DNA was extracted from saliva and genotyping was performed by real time polymerase chain reaction using a standard protocol.

Results: Handball and soccer players had a higher frequency of the ACTN3 RR genotype and a lower frequency of RX + XX genotypes than the control group. Furthermore, the frequency of athletes being homozygous for the ADRB1 Ser49Ser genotype was significantly higher in team sport athletes in comparison to those competing in power/sprint sports. We did not detect any other differences in genotype distributions between the study groups.

Conclusion: This study provides some evidence that the ACTN3 R577X and the ADRB1 Ser49Gly polymorphisms are associated with team sport performance. Larger prospective studies focusing on the genetic influence of performance development are required for confirmation of these findings.

Keywords: soccer, handball, single nucleotide polymorphisms, ACTN3, ADRB1, ADRB2, ADRB3

Zusammenfassung

Hintergrund: In den letzten Jahren wurden zahlreiche Studien durchgeführt, um den Einfluss verschiedener genetischer Varianten auf die sportliche Leistungsfähigkeit von EliteathletInnen sowohl im Ausdauer- als auch im Sprint-/Kraftbereich zu untersuchen, der Bereich der Teamsportarten wurde dabei aber mit wenigen Ausnahmen ausser Acht gelassen. Das Ziel der vorliegenden Studie war es daher, die Häufigkeit ausgewählter Polymorphismen bei österreichischen AthletInnen aus den Bereichen Teamsport (Handball, Fussball), Ausdauersport und Kraftsport sowie in einer gesunden Kontrollgruppe zu erheben und miteinander zu vergleichen.

Methodik: Es wurden Genotyp- und Allelhäufigkeiten der ACTN3 R577X (rs1815739), ADRB1 Ser49Gly (rs1801252), ADRB2 Arg16Gly (rs1042713), ADRB2 Gln27Glu (rs1042714) und ADRB3 Trp64Arg (rs4994) Polymorphismen von 56 österreichischen Kraftsport-, 86 Ausdauer-, 143 TeamsportathletInnen sowie von 216 gesunden Kontrollpersonen erhoben. Genomische DNA wurde aus den Speichelproben isoliert und die genetischen Varianten mittels Polymerasekettenreaktion nach einem standardisierten Protokoll detektiert.


Conclusion: Diese Studie trägt zur Evidenz bei, dass die ACTN3 R577X und die ADRB1 Ser49Gly Polymorphismen mit der Teamporformance assoziiert sind. Es sind jedoch grösse prospektive Studien erforderlich, die den Einfluss der Genetik auf die Leistungsentwicklung untersuchen, um die vorliegenden Ergebnisse zu bestätigen.

Schlagwörter:
Fussball, Handball, Einzelnukleotidpolymorphismus, ACTN3, ADRB1, ADRB2, ADRB3
Introduction

Unlike the vast number of studies investigating the genetic contribution to endurance and sprint/power performance, the genetic contribution to success in team sport performance has received limited attention, and only a few studies have explicitly characterized genetic variants in elite team sport athletes, mainly soccer players [1–6]. While endurance and power athletes represent the physiological endpoints of the sporting continuum, handball and soccer players require a combination of anaerobic and aerobic qualities. As athletes engaged in handball and soccer have to repeatedly produce maximal efforts like sprints, interspersed with short (handball) or longer (soccer) recovery periods over an extended period of time, soccer and handball can be considered as mixed-energy system sports. In both sports, the ability to generate fast muscle actions during the short and highly intensive periods of critical game actions and a high endurance capacity during the periods in between can be considered as performance-limiting factors [7,8]. Having these physiological demands in mind, it seems to be necessary to investigate candidate genes known to influence both, power or sprinting capacity as well as endurance capacity.

The ACTN3 gene encodes for the sarcomeric α-actinin-3 (ACTN3) protein which is nearly exclusively expressed in fast, glycolytic type IIX fibres. Therefore, it plays an important role in the generation of explosive powerful contractions [9]. North et al. identified a common genetic variant (ACTN3 R577X) that results in either expressing a fully functional or a truncated version of ACTN3. Approximately 16% of the world population are predicted to have a congenital deficiency of α-actinin-3, suggesting that other factor(s) likely compensate for its absence at the Z lines of skeletal muscle fast fibres such as α-actinin-2, which is expressed in all muscle fibers [10,11]. Based on these findings, numerous studies have been conducted to investigate the influence of the ACTN3 R577X polymorphism on elite athletic performance and a recent meta-analysis strongly confirmed that ACTN3 deficiency is detrimental for power and sprint activities, but there seems to be no evidence that the X allele is advantageous for endurance sports [12].

Beta adrenergic receptors (ADRB) are members of the G protein-coupled receptor superfamily being targets for the endogenous catecholamines noradrenaline (norepinephrine) and adrenaline (epinephrine). In humans, three subtypes of beta adrenergic receptors are known (beta-1, beta-2 and beta-3). They are expressed in a large variety of cells and play a pivotal role in the regulation of the cardiac, pulmonary, vascular, endocrine and central nervous system [13]. As the cardiovascular response to exercise is highly individual, genetic variations in genes encoding for receptors involved in cardiovascular regulation are supposed to exert an influence on exercise performance [14]. Hence, ADRB genes are candidates of interest since B1-adrenergic receptors influence cardiac function like heart rate and stroke volume, while the B2-adrenergic receptors primarily affect vascular function as they are expressed through many cell types such as airway smooth muscle cells, airway epithelial cells and blood vessels [15]. On the contrary, B3-adrenergic receptors are primarily found in adipose tissue where they stimulate lipolysis and thermogenesis [16]. However, studies on ADRB polymorphisms and elite performance are scarce and in many cases solely endurance athletes are investigated [17–19].

Therefore, the aim of the study was to compare genotype and allele frequencies of the ACTN3 R577X (rs1815739), ADRB1 Ser49Gly (rs1801252), ADRB2 Arg16Gly (rs1042713), ADRB2 Gln27Glu (rs1042714) and the ADRB3 Thr64Arg (rs4994) polymorphisms between Austrian team sport athletes (handball, soccer) with Austrian endurance and power athletes as well as a healthy control group. We hypothesized that team sport athletes would have a higher frequency of the ACTN3 R577R (beneficial for power), ADRB1 Ser49Ser, ADRB2 Arg16Arg, ADRB2 Gln27Glu and ADRB3 Arg64Arg (beneficial for endurance capacity).

Methods

Participants

The study population consisted of 56 power athletes (sprinters and jumpers (n=49), throwers (n=5), weightlifters (n=2)), 86 endurance athletes (middle and long distance runners (n=63), road cyclists (n=17), triathletes (n=5), biathletes (n=1), 143 team sport athletes (soccer players (n=82), handball players (n=61), and 216 healthy non-athletic controls. Participants included both genders, were of Caucasian descent and had an age between 18 and 83 years.

Athletes were included in the study sample only if they are currently active or had participated previously at the international or at least at the highest national level within the respective discipline. The physical activity level (PAL) of healthy controls was determined by dividing the total energy expenditure over a 24 hour period by the basal metabolic rate. While energy expenditure was estimated from the International Physical Activity Questionnaire (IPAQ) using metabolic equivalent (MET) values according to the 2011 compendium of physical activities [20], the basal metabolic rate was predicted as a function of sex, age, weight, and height as suggested previously [21]. Healthy controls were categorized into having a sedentary or light activity lifestyle (PAL 1.40–1.69), an active or moderately active lifestyle (PAL 1.70–1.99), a vigorous or vigorously active lifestyle (PAL 2.00–2.40) according to the cut points provided by the Food and Agriculture Organization of the United Nations [22]. Controls having a PAL above 2.40 were excluded from the study as they could be considered as having a PAL similar to athletes. Further exclusion criteria consisted of diagnosed cardiorespiratory disease (both groups) and the proven intake of performance-enhancing substances (athletes).

Written informed consent was obtained from all participants and the study was conducted according to the Declaration of Helsinki for Human Research of 1974 (last modified in 2000). The study protocol was approved by the ethics committee of the Medical University of Vienna, Austria (EK-Nr. 416/2008).

Genotyping

Genomic DNA was isolated from saliva using a mouthwash protocol as described previously [23]. The subjects were asked not to smoke, drink or eat one hour before sample collection. For saliva collection subjects had to rinse the mouth using 10 ml Listerine (Johnson & Johnson, Vienna, Austria)
for at least 20 seconds and to spit the liquid into a 50 ml centrifuge tube. DNA was extracted from the saliva samples using the QIAamp DNA Mini Kit (Qiagen, Hilden, Germany) according to the manufacturer’s instructions. Quantity and quality of DNA was checked on a NanoDrop spectrophotometer (Peqlab, Erlangen, Germany). Isolated DNA was stored at –80°C until further analysis.

Genotyping was performed using real time PCR (7500 Real-Time PCR System, Applied Biosystems, Foster City, CA) and commercially available TaqMan SNP genotyping assays (ACTN3 R577X (rs1815739, C_590093_1), ADRB1 Ser49Gly (rs1801252, C_8898508_10), ADRB2 Arg16Gly (rs1042714, C_2084764_20), ADRB2 Gln27Glu (rs1042714, C_2084765_20), ADRB3 Trp64Arg (rs4994, C_2215549_20), Applied Biosystems). Each predesigned genotyping assay includes two allele-specific TaqMan MGB probes containing distinct fluorescent dyes (VIC, FAM) and a PCR primer pair to detect the specific SNP targets. Samples were analysed in duplicates and a non-template control was added on each 96 well plate to orient the VIC- and FAM dyes to an origin as well as to enable the detection of DNA contamination. Genotypes were analysed using the SDS 2.1 software (Applied Biosystems).

Statistical analysis

The SPSS statistical package version 21.0 was used to perform all statistical evaluations. Baseline differences between groups in continuous variables were calculated using ANOVA followed by Bonferroni-corrected post hoc analyses. Allele frequencies were determined by gene counting. The Chi-square test was used to prove whether the observed genotype frequencies were in Hardy-Weinberg equilibrium (HWE) and to compare the allele and genotype frequencies between the groups. The level of significance was set at p <0.05.

Results

In total 501 subjects were enrolled in the study. The overall characteristics of these subjects are shown in Table 1. The frequency of female athletes was generally lower as compared to the healthy controls ($\chi^2 = 60.2; p < 0.001$). Furthermore, the age of team sport athletes was about 12 years lower than the mean age of all the other study groups. In addition, both male and female team sport athletes were taller than healthy controls and endurance athletes and had a higher body mass and BMI than endurance athletes. The mean PAL level of both male and female control subjects corresponded to an active lifestyle, whereby 13 (6.0%) people could be considered as inactive, 88 (17.6%) persons had a sedentary or lightly active lifestyle, 74 (40.7%) were categorized as moderately active and 41 (19.0%) as vigorously active.

Asking team-sport athletes (n = 142) to rate the importance of either endurance or strength for their respective discipline on a scale from 0 (= endurance) to 10 (= strength) the mean of all team sport athletes was 4.6 ± 1.5 with similar ratings in both groups (soccer players: 4.6 ± 1.6, handball

<table>
<thead>
<tr>
<th>Healthy controls</th>
<th>Endurance athletes</th>
<th>Team sport athletes</th>
<th>Power athletes</th>
<th>p-value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total number</td>
<td>216</td>
<td>86</td>
<td>143</td>
<td>56</td>
</tr>
<tr>
<td>Gender [m/f]</td>
<td>102 / 114 (52.8% f)</td>
<td>67 / 19 (22.1% f)</td>
<td>119 / 24 (16.8% f)</td>
<td>42 / 14 (25.0% f)</td>
</tr>
<tr>
<td>Age [years]</td>
<td>35.1 ± 12.8 *</td>
<td>33.4 ± 10.6 *</td>
<td>23.5 ± 4.8 *</td>
<td>37.5 ± 14.0 *</td>
</tr>
<tr>
<td>Height [m] – males</td>
<td>1.80 ± 0.07 *</td>
<td>1.79 ± 0.05 *</td>
<td>1.84 ± 0.07 *</td>
<td>1.83 ± 0.06  *</td>
</tr>
<tr>
<td>Body mass [kg] – males</td>
<td>78.3 ± 11.3 *</td>
<td>66.9 ± 7.4 *</td>
<td>82.2 ± 9.4 *</td>
<td>83.1 ± 13.5</td>
</tr>
<tr>
<td>BMI [kg/m²] – males</td>
<td>24.0 ± 2.7</td>
<td>20.8 ± 1.7 *</td>
<td>24.1 ± 1.5</td>
<td>24.8 ± 3.3</td>
</tr>
<tr>
<td>PAL – males</td>
<td>1.72 ± 0.27</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Height [m] – females</td>
<td>1.67 ± 0.06 *</td>
<td>1.67 ± 0.05 *</td>
<td>1.74 ± 0.08 *</td>
<td>1.70 ± 0.06  *</td>
</tr>
<tr>
<td>Body mass [kg] – females</td>
<td>63.8 ± 11.4</td>
<td>52.6 ± 6.8 *</td>
<td>68.3 ± 8.0</td>
<td>58.2 ± 6.0</td>
</tr>
<tr>
<td>BMI [kg/m²] – females</td>
<td>22.9 ± 4.1</td>
<td>18.9 ± 1.8 *</td>
<td>22.4 ± 1.8</td>
<td>20.0 ± 1.1</td>
</tr>
<tr>
<td>PAL – females</td>
<td>1.78 ± 0.29</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
</tbody>
</table>

Data of subject characteristics are expressed as mean ± standard deviation or frequencies; Differences between groups were detected either by Chi-square test or one way ANOVA: * p<0.05 compared to healthy controls and # p<0.05 compared to team sport athletes as determined by Bonferroni post-hoc analyses.

Abbreviations: m [males], f [females], BMI [body mass index], PAL [physical activity level]

Table 1: Subject characteristics
players: 4.6 ± 1.4; p = 0.871) illustrating that team sport athletes consider endurance and strength equally important for achieving success.

Genotype distribution for the ACTN3 genotype was in Hardy-Weinberg equilibrium within each group. We did not observe any differences in either genotype or allele frequencies between groups (p > 0.05) as shown in table 2. However, when the dominant genetic model (RR vs. RX + XX) was applied, team sport athletes had a higher frequency of the RR genotype and a lower frequency of RX + XX than the control group (p = 0.029). A sub-group analysis between soccer and handball players did not reveal any differences.

Genotype distributions were in Hardy Weinberg equilibrium except for the ADRB1 Ser49Gly and ADRB2Gln-27Glu variants in power athletes (ADRB1 Ser49Gly: controls: \( \chi^2 = 1.650; p = 0.199; \) endurance athletes: \( \chi^2 = 0.613; p = 0.688; \) team sport athletes: \( \chi^2 = 0.650; p = 0.420; \) power athletes: \( \chi^2 = 4.275; p = 0.039 \) and ADRB2 Gln27Glu: controls: \( \chi^2 = 1.560; p = 0.212; \) endurance athletes: \( \chi^2 = 0.8496; p = 0.357; \) team sport athletes: \( \chi^2 = 0.001; p = 0.973; \) power athletes: \( \chi^2 = 5.729; p = 0.017 \) and the ADRB3 Trp64Arg genotype in team sport athletes (controls: \( \chi^2 = 0.071; p = 0.789; \) endurance athletes: \( \chi^2 = 0.0108; p = 0.917; \) team sport athletes: \( \chi^2 = 11.012; p = 0.001; \) power athletes: \( \chi^2 = 0.966; p = 0.326). With respect to the adrenergic receptors we did not observe significant overall differences in genotype and allele frequencies among the study groups, but having a closer look to pairwise comparisons between team sport athletes and the other groups, some trends could be detected as summarized in Table 3. In comparison to power athletes, the frequency of the ADRB1 Ser49Ser (GG) genotype was significantly higher in team sport athletes (7.1% versus 0.7%, p = 0.045). Furthermore, team sport athletes differed from controls as the proportion of subjects being heterozygote for the ADRB3 Trp64Arg genotype was significantly lower in team sport athletes than in controls (15.8% versus 9.1%, p = 0.037).

### Discussion

In this study we aimed to investigate whether genetic variants in the ACTN3, ADRB1, ADRB2 and ADRB3 genes would differ between Austrian team sport, endurance and power athletes as well as healthy controls. Our data revealed that handball and soccer players had a higher frequency of the ACTN3 RR genotype and a lower frequency of RX + XX than the control group (p = 0.029). A sub-group analysis between soccer and handball players did not reveal any differences.

Genotype distributions were in Hardy Weinberg equilibrium except for the ADRB1 Ser49Gly and ADRB2Gln-27Glu variants in power athletes (ADRB1 Ser49Gly: controls: \( \chi^2 = 1.650; p = 0.199; \) endurance athletes: \( \chi^2 = 0.613; p = 0.688; \) team sport athletes: \( \chi^2 = 0.650; p = 0.420; \) power athletes: \( \chi^2 = 4.275; p = 0.039 \) and ADRB2 Gln27Glu: controls: \( \chi^2 = 1.560; p = 0.212; \) endurance athletes: \( \chi^2 = 0.8496; p = 0.357; \) team sport athletes: \( \chi^2 = 0.001; p = 0.973; \) power athletes: \( \chi^2 = 5.729; p = 0.017 \) and the ADRB3 Trp64Arg genotype in team sport athletes (controls: \( \chi^2 = 0.071; p = 0.789; \) endurance athletes: \( \chi^2 = 0.0108; p = 0.917; \) team sport athletes: \( \chi^2 = 11.012; p = 0.001; \) power athletes: \( \chi^2 = 0.966; p = 0.326). With respect to the adrenergic receptors we did not observe significant overall differences in genotype and allele frequencies among the study groups, but having a closer look to pairwise comparisons between team sport athletes and the other groups, some trends could be detected as summarized in Table 3. In comparison to power athletes, the frequency of the ADRB1 Ser49Ser (GG) genotype was significantly higher in team sport athletes (7.1% versus 0.7%, p = 0.045). Furthermore, team sport athletes differed from controls as the proportion of subjects being heterozygote for the ADRB3 Trp64Arg genotype was significantly lower in team sport athletes than in controls (15.8% versus 9.1%, p = 0.037).
genotype) would result in higher endurance capacity. A study with 200 professional soccer players of Brazilian first division teams showed that ACTN3/RR individuals spent less time to run 10, 20 and 30 m, presented a higher jump potential, but had a lower VO₂ max than ACTN3/XX individuals [5]. A very recent study from the same group, where the athletes were first ranked in ascending order according to their performance in each test and then divided into performance quartiles, did not reveal any significant differences in genotypic or allelic frequencies between different performance ratings in adult, professional, U-20 and U-17 years Brazilian first-division soccer players [3].

However, Massida et al. [26] demonstrated the importance of the ACTN3 R577X polymorphism in soccer performance by constructing a new genetic model based on genotype scores proposed by Williams and Folland [27]. By analysing the relationship between six genetic polymorphisms (ACE, ACTN3, BDKRB2, VDR-ApaI, VDR-BsmI, and VDR-FokI) and jumping performance, they came to the conclusion that the ACTN3 R577X polymorphism was the most important genetic variation in predicting vertical jump height, even though no significant differences in the total genotype score between athletes and the control group were detected. In summary, it seems to be evident that the ACTN3

<table>
<thead>
<tr>
<th>Polymorphism</th>
<th>Genotype / allele</th>
<th>Healthy controls</th>
<th>Endurance athletes</th>
<th>Team sport athletes</th>
<th>Power athletes</th>
<th>Overall p-value</th>
<th>Team sports vs Control</th>
<th>Team sports vs Endurance</th>
<th>Team sports vs Power</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>ADRB1 Ser49Gly (rs1801252)</strong></td>
<td>AA (%)</td>
<td>71.6</td>
<td>75.6</td>
<td>67.9</td>
<td>75.5</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>AG (%)</td>
<td>27.9</td>
<td>23.3</td>
<td>25.0</td>
<td>23.8</td>
<td>0.076</td>
<td>0.638</td>
<td>1.000</td>
<td><strong>0.045</strong></td>
</tr>
<tr>
<td></td>
<td>GG (%)</td>
<td>0.5</td>
<td>1.2</td>
<td>7.1</td>
<td>0.7</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>ρ (A)</td>
<td>0.86</td>
<td>0.87</td>
<td>0.80</td>
<td>0.87</td>
<td>0.612</td>
<td>0.622</td>
<td>0.964</td>
<td>0.205</td>
</tr>
<tr>
<td></td>
<td>ρ (G)</td>
<td>0.14</td>
<td>0.13</td>
<td>0.20</td>
<td>0.13</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>ADRB2 Gly16Arg (rs1042713)</strong></td>
<td>GG (%)</td>
<td>40.0</td>
<td>41.9</td>
<td>35.7</td>
<td>37.5</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>GA (%)</td>
<td>47.0</td>
<td>44.2</td>
<td>52.4</td>
<td>53.6</td>
<td>0.851</td>
<td>0.597</td>
<td>0.480</td>
<td><strong>0.833</strong></td>
</tr>
<tr>
<td></td>
<td>AA (%)</td>
<td>13.0</td>
<td>14.0</td>
<td>11.9</td>
<td>8.9</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>ρ (G)</td>
<td>0.63</td>
<td>0.64</td>
<td>0.62</td>
<td>0.64</td>
<td>0.983</td>
<td>0.759</td>
<td>0.754</td>
<td>0.753</td>
</tr>
<tr>
<td></td>
<td>ρ (A)</td>
<td>0.37</td>
<td>0.36</td>
<td>0.38</td>
<td>0.36</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>ADRB2 Gln27Glu (rs1042714)</strong></td>
<td>CC (%)</td>
<td>40.9</td>
<td>36.0</td>
<td>32.2</td>
<td>26.8</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>CG (%)</td>
<td>41.4</td>
<td>44.2</td>
<td>49.0</td>
<td>60.7</td>
<td>0.206</td>
<td>0.231</td>
<td>0.771</td>
<td>0.300</td>
</tr>
<tr>
<td></td>
<td>GG (%)</td>
<td>17.7</td>
<td>19.8</td>
<td>18.9</td>
<td>12.5</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>ρ (C)</td>
<td>0.62</td>
<td>0.58</td>
<td>0.47</td>
<td>0.57</td>
<td>0.790</td>
<td>0.346</td>
<td>0.825</td>
<td>0.949</td>
</tr>
<tr>
<td></td>
<td>ρ (G)</td>
<td>0.38</td>
<td>0.42</td>
<td>0.43</td>
<td>0.43</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>ADRB3 Trp64Arg (rs4994)</strong></td>
<td>TT (%)</td>
<td>83.7</td>
<td>80.2</td>
<td>88.1</td>
<td>82.1</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>TC (%)</td>
<td>15.8</td>
<td>18.6</td>
<td>9.1</td>
<td>17.9</td>
<td><strong>0.127</strong></td>
<td><strong>0.037</strong></td>
<td><strong>0.087</strong></td>
<td><strong>0.157</strong></td>
</tr>
<tr>
<td></td>
<td>CC (%)</td>
<td>0.5</td>
<td>1.2</td>
<td>2.8</td>
<td>0.0</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>ρ (T)</td>
<td>0.92</td>
<td>0.90</td>
<td>0.93</td>
<td>0.91</td>
<td><strong>0.875</strong></td>
<td><strong>0.725</strong></td>
<td><strong>0.412</strong></td>
<td><strong>0.707</strong></td>
</tr>
<tr>
<td></td>
<td>ρ (C)</td>
<td>0.08</td>
<td>0.10</td>
<td>0.07</td>
<td>0.09</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Table 3: ADRB genotype and allele frequencies in Austrian controls (n=216), endurance athletes (n=86), team sport athletes (n=143) and power athletes (n=56)
R577X genotype is associated with power and/or speed, but it has to be confirmed in further studies whether this association is relevant for success in team sports such as handball or soccer.

This is the first study investigating the allelic and genotype frequencies of four main variants of adrenoceptor beta genes (ADRB1 Ser49Gly, ADRB2 Arg16Gly, ADRB2 Gln27Glu and ADRB3 Trp64Arg) in endurance, sprint/power and team sport athletes competing in Austria and our data revealed that team sport athletes differed in their ADRB1 Ser49Gly genotype distribution from sprint/power athletes as well as in their ADRB3 Trp64Arg genetic profile from controls. Studies on the association of the ADRB1 Ser49Gly polymorphism and sports performance are scarce and are mainly performed in patients with cardiac problems. Hence, a study in patients with heart failure has shown that subjects homozygous for Gly49 had a better physical endurance and a lower resting hearth rate than those homozygous for Ser49 [28]. Furthermore, a study in Polish athletes observed that the frequency of the 49Gly allele was significantly higher in endurance athletes than in the controls (11% versus 6.4%) [29]. Although the frequency of the Gly49Gly genotype is generally somewhat lower in our study group, we detected the highest proportion of this genotype in the group of team sport athletes (7.1%) which could point to a possible favourable effect of the Ser49 variant for team sports performance.

The ADRB2 Gly16Arg polymorphism was previously associated with endurance capacity with the Arg (A) allele exerting a favourable effect [18]. For the ADRB2 Gln27Glu polymorphism it has been shown that maximal oxygen consumption is lower in women homozygous for ADRB2 Gln27Glu implying a favourable effect of the Gln (C) allele for endurance performance [30]. However, similar to some other studies we could not confirm any association with athletic status and ADRB2 polymorphisms [17,31].

The ADRB3 Arg (C) allele has been shown to be overrepresented in Spanish world-class endurance athletes [17]. Furthermore, a study among Korean controls and athletes from different disciplines revealed that the highest proportion of the subject carrying the Arg allele was detected for volleyball and gymnastics [32]. This corresponds well to our data indicating the highest frequency of the Arg allele in team sport athletes.

Summarising our study results from the genetic point of view, it can be assumed that sprint/power as well as endurance performance seems to be equally important for success in team sport athletes as both the “power” variant of ACTN3 as well as the “endurance” variant of ADRB1 and ADRB3 were slightly overrepresented. This assumption is further supported by the fact that the team sport athletes themselves rated both strength and endurance as being equally for achieving success. However, this study results have to be treated with some cautions as several limitations might have impacted the results.

First of all we have to consider that there is a limited number of elite athletes worldwide and especially in Austria when it comes to summer sports. Hence, we were unable to find a suitable high number of “world class” athletes as defined previously [33]. However, the included athletes are amongst the best within their discipline in Austria and the data obtained could be very useful on the national level. Furthermore, the number of females is clearly underrepresented in our study population, which indicates that the results of this study may be applicable for male athletes rather than for athletes of both sexes. Finally, the cross-sectional design of our study does not allow drawing cause-effect relationships for the assessment of team sport performance. It is obvious that there are other genetic variants and epigenetic mechanisms that influence athletic performance in complex sports such as handball or soccer. Thus, additional large-scale prospective studies should be envisioned eliminating the above mentioned limitations.

Conclusion

In summary, this study provides preliminary evidence that the ACTN3 R577X as well as the ADRB1 Ser49Gly polymorphisms are associated with team sport performance. Larger prospective studies involving performance tests and including world class elite team sport athletes of both genders are required for confirmation of these findings.

Conflict of interest and funding

The study was supported by financial means of the Austrian Federal Chancellery – Section Sports (GZ 703.710/0027-VI/3/2008). The authors declare not to have any conflicts of interest in the manuscript, including financial, institutional and other relationships that might lead to a conflict of interest.

Practical implications

• These study shows that sports performance in team sports is affected by genetics (similar to sheer endurance or power sports).
• Genetic testing of team sport athletes may have limited value without considering the physiological demands of the respective discipline.
• Ethical issues have to be considered (especially when testing young athletes or when personal data are available for third parties).

Corresponding author

Assoc. Prof. DI Dr. Barbara Wessner
Centre for Sport Science and University Sports
University of Vienna
Auf der Schmelz 6,
A-1150 Vienna, Austria
Mail: barbara.wessner@univie.ac.at
Phone: +43-1-4277-48875

References


