universität wien

Magisterarbeit

Titel der Magisterarbeit

"Frequency and distribution of selected performance related genetic variants: a comparison between soccer players, handball players and a control group"

verfasst von

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angestrebter akademischer Grad

Magister der Naturwissenschaften (Mag.rer.nat.)

Wien, 2015

Studienkennzahl lt. Studienblatt:	A 066 826
Studienrichtung lt. Studienblatt:	Magisterstudium Sportwissenschaft
Betreuerin:	Assoz. Prof. Dipl.-Ing. Dr. Barbara Wessner

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Thomas Teynor, Bakk.rer.nat.

Ternitz, im Oktober 2015

Danksagung

Ich möchte die Gelegenheit nutzen und mich bei Assoz. Prof. Dipl.-Ing. Dr. Barbara Wessner für ihre Unterstützung und die Betreuung meiner Magisterarbeit bedanken.

Ein großes Dankeschön gebührt auch Karin Mesicek, MTA, die mir bei diversen Fragen hilfsbereit zur Seite gestanden ist.

Herzlicher Dank geht an meine Eltern und an meine Schwestern, die mich während des Studiums unterstützt haben und mir immer mit Rat beigestanden sind.

Ein besonderer Dank gilt meinem amerikanischen Gastbruder Drake Hyman für das Korrekturlesen meiner Magisterarbeit!

Abschließend möchte ich mich auch bei meiner Freundin Vici für ihre Geduld während meines Studiums und für ihre (mentale) Unterstützung bei diversen Abgaben bedanken.

Zusammenfassung

Einleitung: Neben Talent, Hingabe und Ehrgeiz scheint es als hätten genetische Variationen einen großen Einfluss auf die sportliche Leistungsfähigkeit, da sie unter anderem mit Muskelfunktion, kardiorespiratorische Funktion, Schnellkraft, Maximalkraft und Ausdauerleistungsfähigkeit in Verbindung gebracht werden. Trotz zahlreicher Studien, die den Zusammenhang zwischen einzelnen genetischen Variationen und sportlicher Leistungsfähigkeit untersucht haben, ist eine klare Aussage nach wie vor schwierig.

Fragestellung: In dieser Magisterarbeit wurde der Zusammenhang zwischen sportlicher Leistungsfähigkeit und genetischen Variationen (Polymorphismen) untersucht. Dabei wurden Unterschiede zwischen trainierten und untrainierten Personen sowie zwischen Handballern und Fußballern betrachtet.

Methodik: 234 männliche Personen (38 Handballer, 81 Fußballer und 115 Kontrollpersonen) wurden auf fünf mit Kraft bzw. Schnellkraft assoziierten Polymorphismen (ACTN3 (rs181539), MSTN (rs1805086), IGF-1 (rs35757), IGF-2 (rs3213221), IGF-2 (rs7924316) und auf sechs mit Ausdauer assoziierten Polymorphismen (ADRB1 (r1801252), ADRB2 (rs1042713), ADRB2 (rs1042714), NRF2 (rs7181866), PPARG1A (rs8192678), HIF1A (rs11549465) untersucht. Chi-Quadrat Tests wurde gerechnet, um Zusammenhänge hinsichtlich genetischer Varianten zwischen Profisportlern (n=119) und einer Kontrollgruppe (n=115) zu erkennen. Im Anschluss daran wurden die beiden Subgruppen Handballer und Fußballer auf die gleichen Zusammenhänge untersucht.

Resultate: Es konnten keine signifikanten Zusammenhänge zwischen der sportlichen Leistungsfähigkeit und genetischen Varianten gefunden werden. Die Resultate des Chi-Quadrat Test waren im Bereich von p=0,051 und p=0,888. Auch in Bezug auf genetische Variationen und Sportarten konnten signifikanten Ergebnisse gefunden werden. Die Ergebnisse des Chi-Quadrat Tests waren im Bereich von p=0,999 und p=0,087.

Conclusio: Im Gegensatz zu bereits publizierten Ergebnissen konnte bei keiner einzigen genetischen Variante ein signifikantes Ergebnis hinsichtlich der Bedeutung für die sportliche Leistungsfähigkeit/Sportart gefunden werden. Zusätzliche Untersuchungen mit größeren Kohorten müssen durchgeführt werden um Zusammenhänge erkennen zu können. Es scheint notwendig, dass bei weiteren Studien die unterschiedlichen Spielpositionen in den Sportteams berücksichtigt werden.

Schlagwörter: ACTN3, MSTN, IGF-1, IGF-2, ADRB1, ADRB2, NRF2, PPARG1A, HIF1A, Fußball, Handball, Sportliche Leistungsfähigkeit, Polymorphismen

Abstract

Introduction: Beside talent, dedication, and ambition it is very likely that genetic variations have a huge impact on one's athletic performance as these are associated with muscular function, cardiorespiratory function, speed-strength, power, and endurance. Although many studies investigating the relationship between genetic variations and athletic performance have been published, there is no consensus on their meaning.

Research question: The aim of this master thesis was to investigate the relationship between athletic performance and genetic variations (polymorphisms). In this respect differences between trained and untrained persons as well as between soccer and handball players were investigated.

Methods: 234 male subjects (38 Handball player, 81 Soccer player and 115 controls) were tested for five with strength and power associated polymorphisms (ACTN3 (rs181539), MSTN (rs1805086), IGF-1 (rs35757), IGF-2 (rs3213221), IGF-2 (rs7924316) and for six with endurance associated polymorphisms ((ADRB1 (r1801252), ADRB2 (rs1042713), ADRB2 (rs1042714), NRF2 (rs7181866), PPARG1A (rs8192678), HIF1A (rs11549465). Chi square tests were calculated to find relationships between genetic variations and professional athletes (n=119) compared to a control group. Additionally the subgroups of soccer players and handball players were investigated for the same relationships.

Results: No relationship between sports performance and genetic variations could be found. Chi square test showed a range from p=0.051 to p=0.888. Also no significant relationship between type of sports (handball or soccer) and genetic variations could be found. Chi square test showed a range from p=0.087 to p=0.999.

Conclusion: Contrary to published studies no relationship between any genetic variation and athletic status/type of sports could be found. Further studies with bigger groups have to be done to find coherencies. Additionally it seems to be necessary to distinguish the different playing positions within the soccer team and the handball team.

Keywords:

ACTN3, MSTN, IGF-1, IGF-2, ADRB1, ADRB2, NRF2, PPARG1A, HIF1A, Soccer players, Handball players, athletic status, polymorphisms

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1. Introduction and aims of the study

In 1990 a worldwide project to sequence the human genome was started. This research was organized and coordinated by the Human Genome Organization (HUGO). The goal of HUGO was to find different gene sequences and gene functions. The puzzle of gene sequences could be solved in 2001 two years faster than expected. Now scientists try to find out how different genes work and which influence they have on physical activity and sports performance (Henn & Meese, 2007, pp. 8-12).

According to Roth (2007) men share more than 99% of its DNA-sequence. That means that men differ less than 1% from each other, no matter where they come from. The differences within this 1% disparity can cause huge effects and make a deep impact on one's athletic performance (Roth, 2007, p. 40).

Researchers and scientists estimate that athletic performance has a big relationship to genetic variations. In other words they think that about 27% to up to more than 40% of athletic performance can be related to different genetic variations (MacArthur & North, 2005, p. 332).

In the 2015 published study "Current Progress in Sports Genomics" the authors point out that at least 120 genetic markers are associated with elite athlete status. Nevertheless they have to admit that more studies with bigger cohorts have to be done so that results can be linked to the practice of sport and performance predicting (Ahmetov & Fedotovskaya, 2015, p. 247).

The purpose of this master thesis is of the same tenor. Its aim is to investigate if certain genetic variations influence athletic performance positively. Therefore 234 male subjects (81 Austrian Soccer players, 38 Handball players and 115 controls) were tested for polymorphisms associated with athletic performance. Up to now the relationship between specific genotypes and athletic status is not clear.

Hence the goal of this master thesis is to close this gap and to answer the following questions:

.) Are there differences related to specific genetic variations between professional athletes (handball and soccer) and a control group?

.) Are there any differences between Austrian elite athletes (handball and soccer) and specific performance related polymorphisms?

.) Is it possible to determine a polygenetic profile for handball and/or soccer players based on the results of this study?

The first part of this master thesis (chapter 2 and chapter 3) deals with genetic basics, the investigated polymorphisms and related literature based on a qualitative literature analysis.

Chapter 4 provides information about the procedure of recruitment and the steps of the polymerase chain reaction.

In chapter 5 all statistical analyses and results are presented. Different figures and tables help the reader to understand the statistical procedures and its results.

In chapter 6 the results of this thesis will be discussed and compared to other studies.

The final chapter of this master thesis will conclude all findings and give an outlook on future studies.

2. Genetic basics

The aim of this chapter is to give an overview about the most important genetic basics. Hence different aspects of human genetics like the structure of the DNA, the function of proteins, and the different types genetic variations will be explained.

2.1. Nucleic acids and their structure

The two most important nucleic acids are the deoxyribonucleic acid (DNA) and the ribonucleic acid (RNA). DNA and RNA are both build of nucleotides chains. DNA is compound of two strands and RNA of one strand. Every nucleotide consists of a nucleoside and phosphor molecule (see Figure 1) (Poeggel & Meitinger, 2003, p. 3; Aubele, 2014, p. 7).

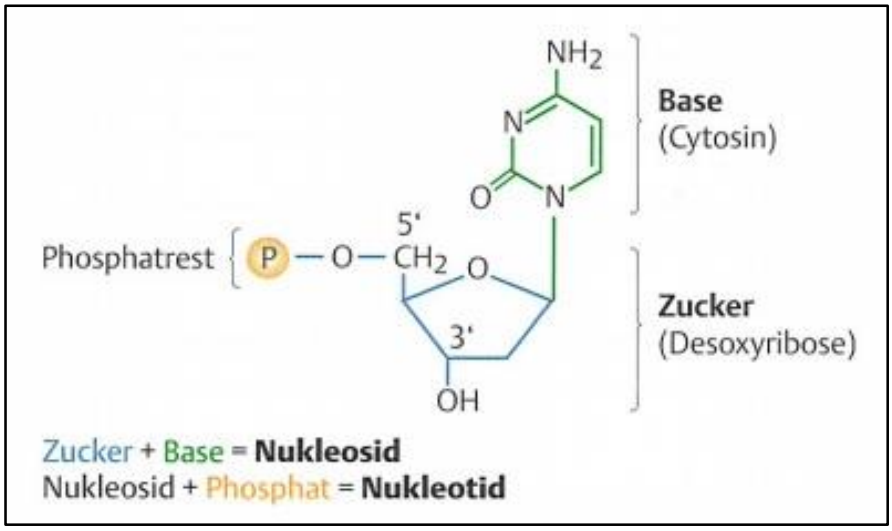

Figure 1: Basics structure of a nucleotide (Poeggel & Meitinger, 2003, p. 3)

For that reason nucleotides can also be referred as nucleoside monophosphates. Each nucleoside is composed of a sugar molecule and a base (Poeggel & Meitinger, 2003, p. 3; Weaver, 2012, p. 5). For the DNA this base belongs either to the purines or to the pyrimidines. Purine is the parent base of adenine (A) and guanine (G) and pyrimidine is the parent base of cytosine (C) and thymine (T). The relationship between the bases can be easily seen in Figure 2. RNA compounds of the same bases like the DNA except that thymine is substituted by uracil (U) (Weaver, 2012, p. 15; Poeggel & Meitinger, 2003, p. 4).

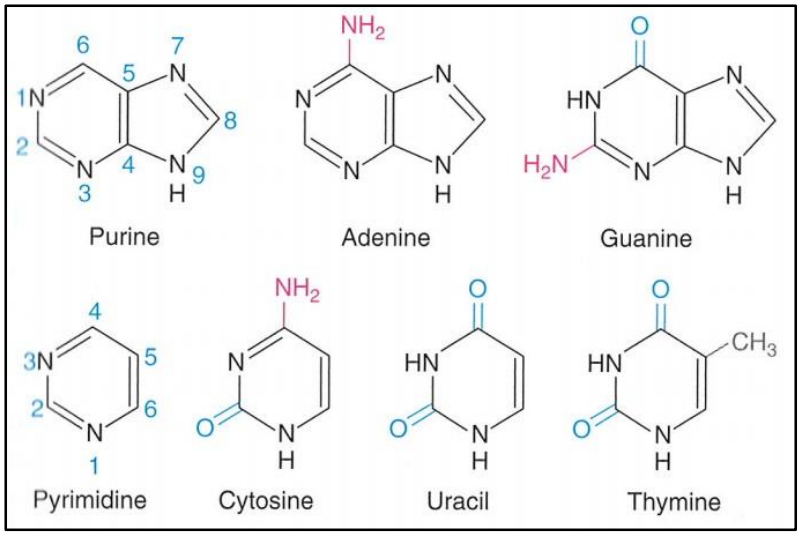

Figure 2: The bases of DNA and RNA (Weaver, 2012, p. 17)

Figure 3 shows an overview of the bases of RNA and DNA.

Base	*Nucleoside (RNA)*	*Deoxynucleoside (DNA)*
Adenine	Adenosine	Deoxyadenosine
Guanine	Guanosine	Deoxyguanosine
Cytosine	Cytidine	Deoxycytidine
Uracil	Uridine	Not usually found
Thymine	Not usually found	(Deoxy)thymidine

Figure 3: Overview of RNA and DNA bases (Weaver, 2012, p. 16)

The DNA compounds of a double-stranded chain which is rotated into a double helix. The connection between the nucleotides is called phosphodiester bond which results in two different ends. One end of this group of three nucleotides is called 5'-end and the other end of the group is called 3'-end (Weaver, 2012, p. 17; Alberts et al., 2010, pp. 173-177). Every base links to the complementary base. Thymine and adenine are complementary and guanine and cytosine are complementary. The different building blocks and the structure of the DNA as well was the complementary bases are presented in Figure 4 (Alberts, 2011, pp 177-178). The binding between the strands is always antiparallel therefore the sequence 5'-CATT-3' on one strand is the complementary sequence to 3'-AATG-5' on another strand (Poeggel & Meitinger, 2011, p. 5).

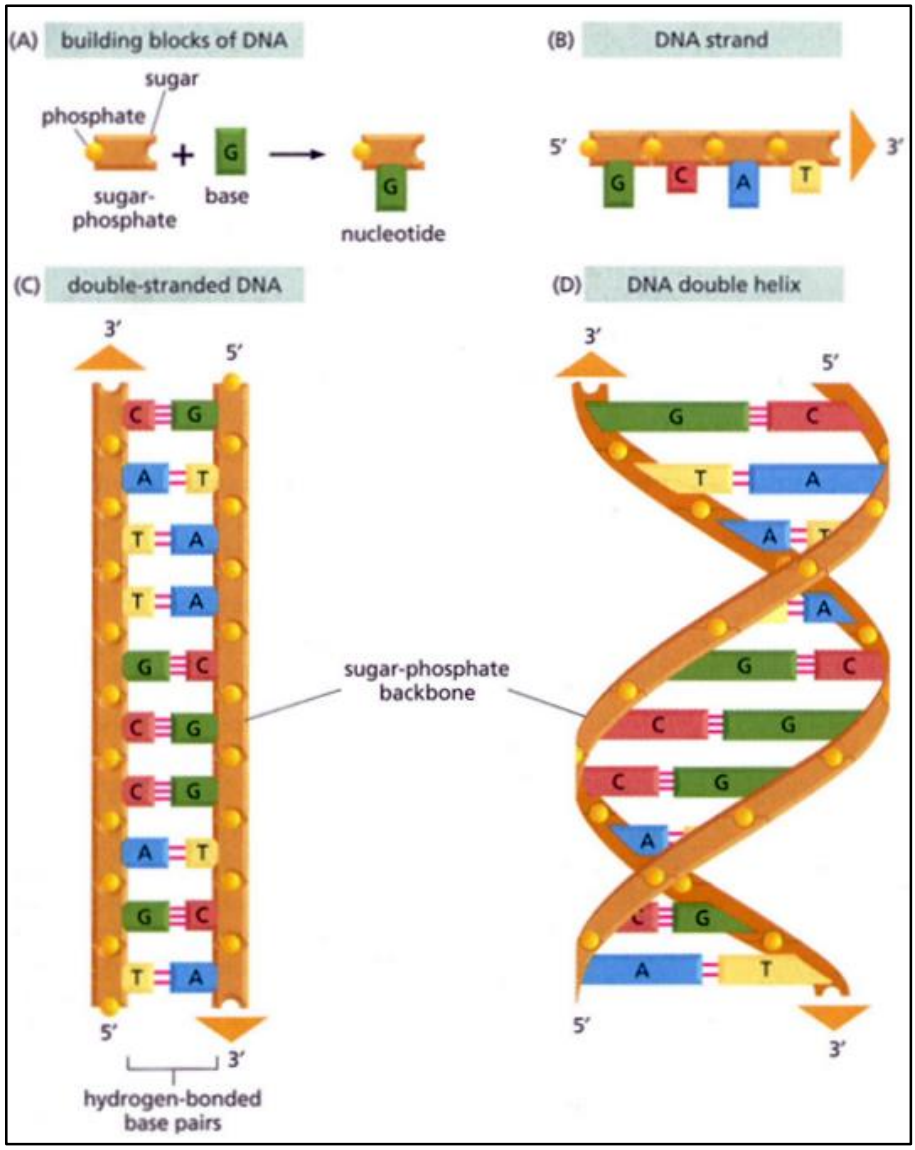

Figure 4: DNA is made of four nucleotide building blocks (Alberts, Bray, Hopkin, Johnson, Lewis, Raff, Roberts & Walter, 2010, p. 173)

2.2. Proteins and amino acids

Proteins have an important role in genetics. They have different tasks such as giving shape to cells or as carrier of signals from one cell to another (Weaver, 2012, p. 35). According to Alberts et al. (2010, p. 120) 9 general categories of protein functions can be set up. Figure 5 shows the classification of Alberts et al. (2010, p. 120).

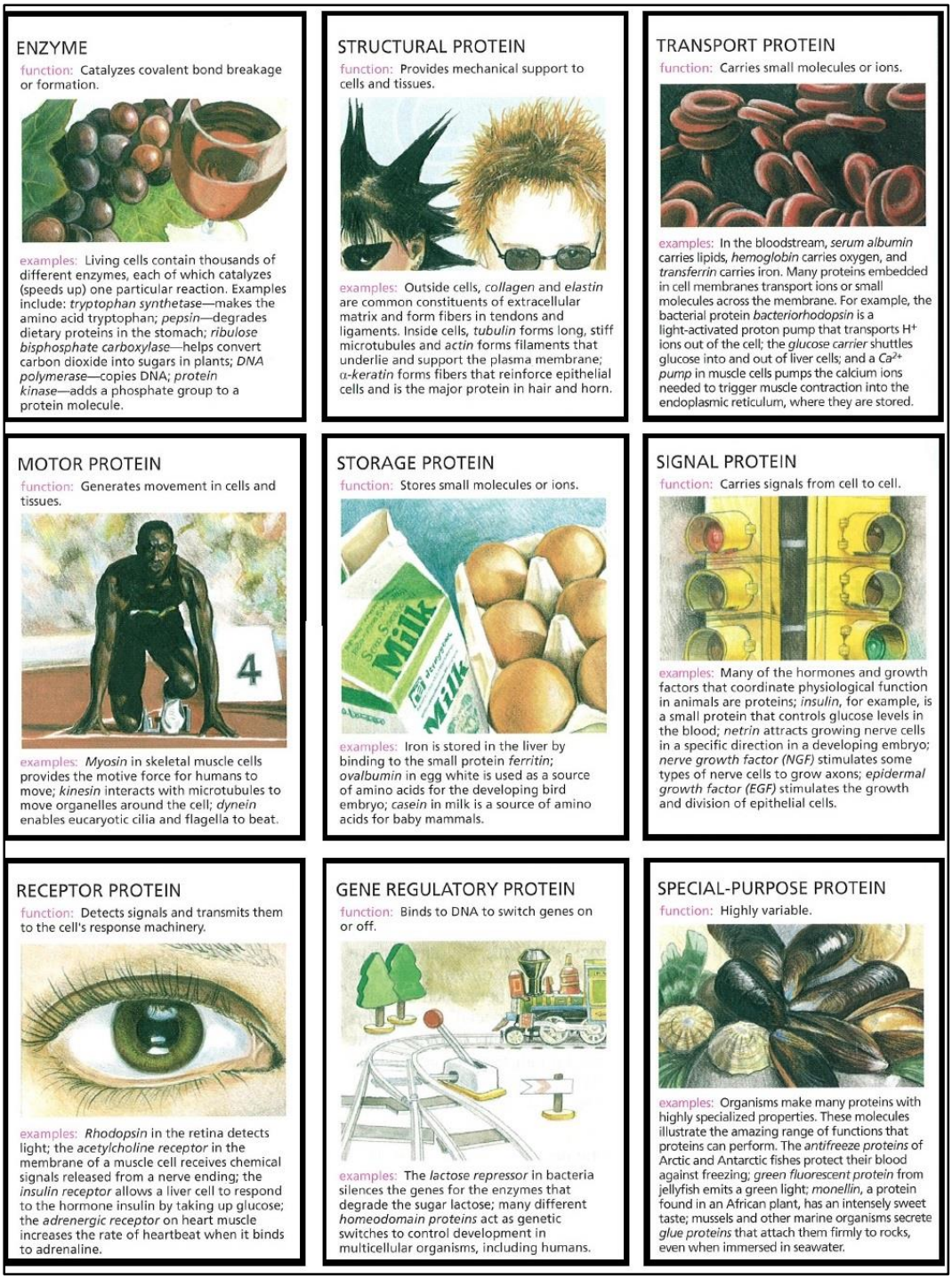

Figure 5: Examples of general protein functions (Alberts et al., 2010, p. 120)

There are 20 different amino acids which built one protein. Proteins are long chains of those different amino acids (Alberts et al., 2010, p. 121). Most proteins have a length between 50 and 2000 amino acids. Nevertheless some proteins are quite short with a length of about 30 amino acids. On the other hand there are amino acids with a length of more than 10.000 amino acids (Albert et al., 2010, p. 125) Each amino acid has

different characteristics and therefore different chemical functions (Alberts et al., 2010, p. 121). Amino acids can be distinguished into two main groups. Group one are the polar amino acids which hydrophilic, group two are the nonpolar amino acids which are hydrophobic (Alberts et al., 2010, p. 122). Beside that each amino acid has either basic side chains, acidic side chains, uncharged polar side chains or nonpolar side chains (Alberts et al., 2010, pp. 72-73).

The chemical structure of the amino acids are presented in Figure 6, Figure 7, Figure 8, and Figure 9.

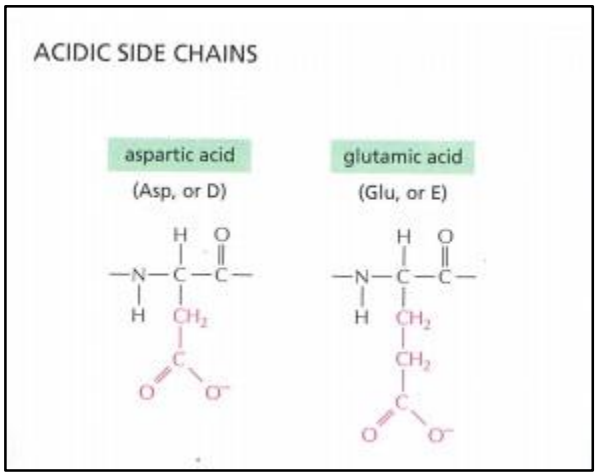

Figure 6: Acidic side chains (Alberts et al., 2010, p. 72)

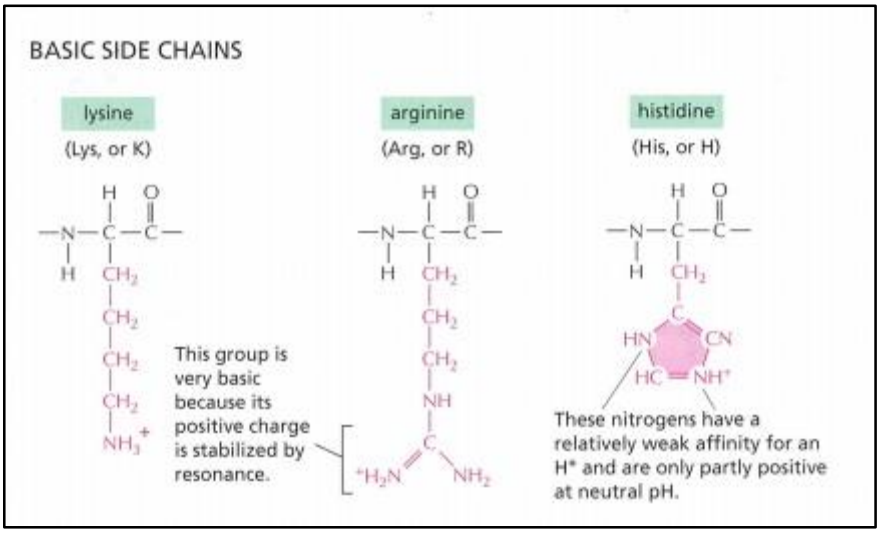

Figure 7: Basic side chains (Alberts et al., 2010, p. 73)

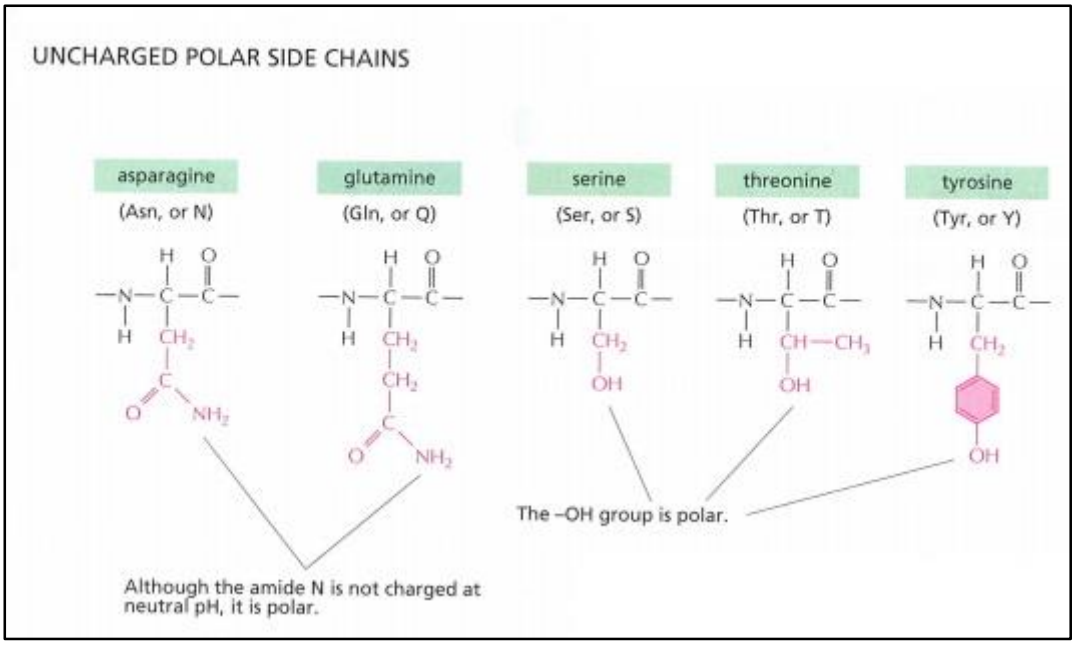

Figure 8: Uncharged polar side chains (Alberts et al., 2010, p. 73)

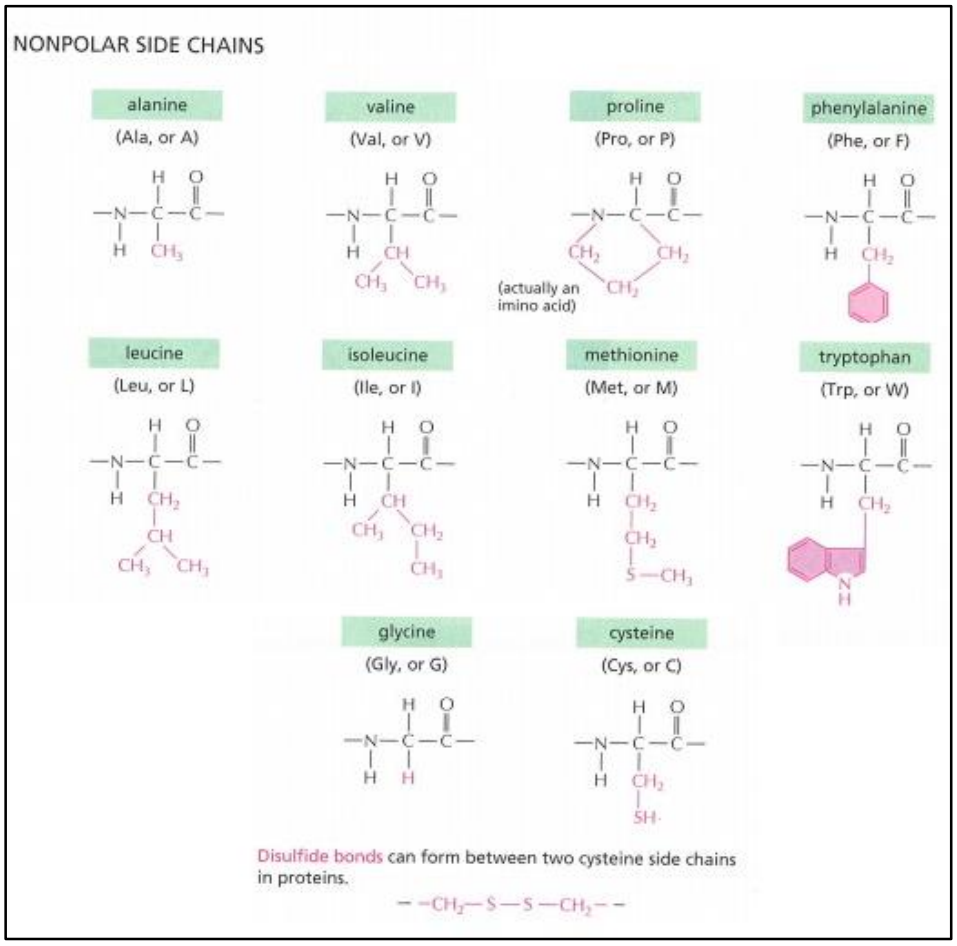

Figure 9: Nonpolar side chains (Alberts et al., 2010, p. 73)

Each amino acid has a three-letter-abbreviation. All amino acids and their abbreviations presented in Table 1.

Table 1: Amino acids and their abbreviations (modified from Alberts et al., 2010, p. 122)

Amino Acid	Abbreviation
Aspartic acid	ASP
Glutamic acid	Glu
Arginine	Arg
Lysine	Lys
Histidine	His
Asparagine	Asn
Glutamine	Gln
Serine	Ser
Threonine	Thr
Tyrosine	Tyr
Alanine	Ala
Glycine	Gly
Valine	Val
Leucine	Leu
Isoleucine	Ile
Proline	Pro
Phenylalanine	Phe
Methionine	Met
Tryptophan	Trp
Cysteine	Cys

Between the nucleic acids (DNA and RNA) and the proteins there are quite some differences. Therefore a translation form one language to another is necessary in order to make the protein synthesis work. Each group of three nucleotides in RNA is called codon. Because RNA and DNA are made of four different nucleotides there are 64 possible combinations of codons. On the other hand only 20 amino acids build the proteins. Therefore different codons can code for the same amino acid (Alberts et al., 2010, pp. 246-247). An overview is presented below (Table 2).

Table 2: Translations of amino acids to RNA (modified from Alberts et al., 2010, p. 247)

GCA GCC GCG GCU	AGA AGG CGA CGC CGG CGU	GAC GAU	AAC AAU	UGC GUG	GAA GAG	CAA CAG	GGA GGC GGG GGU	CAC CAU	AUA AUC AUU
Ala	Arg	Asp	Asn	Cys	Glu	Gln	Gly	His	Ile
A	R	D	N	E	E	Q	G	H	I

UUA UUG CUA CUC CUG CUU	AAA AAG	AUG	UUC UUU	CCA CCC CCG CCU	AGC AGU UCA UCC UCG UCU	ACA ACC ACG ACU	UGG	UAC UAU	GUA GUC GUG GUU
Leu	Lys	Met	Phe	Pro	Ser	Thr	Trp	Tyr	Val
Leu	K	M	F	P	S	T	W	Y	V

As one can see only 61 codons are presented in Table 2. The three missing codons UAA, UAG, and UGA are so called stop codons. These codons signal the end of the sequence (Alberts et al., 2010, p. 247).

2.3. Types of genetic variations

As mentioned in chapter 1 men's DNA-sequence differs by less than 1%. That means although we share about 99% of our DNA-sequence 1% makes a huge difference am makes every men unique (Roth, 2007, p.40).

Within this 1% range polymorphisms and mutations are responsible for the difference of men. Scientists talk about polymorphisms when this genetic variations are common in more than 1% of the human population (Müller-Myhosk, 2003, p. 98, in Ganten & Ruckpaul, 2003). When a genetic variations occurs less than 1% in the human population it is called mutation (Görtzen & Rüdiger, 2007, S. 1170; Müller-Myhosk, 2003, p. 98, in Ganten & Ruckpaul, 2003).

In 1994 Passarge defined polymorphisms in the following way:

> "Als Polymorphismus wird das Vorkommen von 2 oder mehr durch verschiedene Allele determinierte unterschiedliche Phänotypen bezeichnet, wobei die seltenste Form nicht durch wiederholte Mutation aufrechterhalten werden kann. Ein Genlocus wird als polymorph bezeichnet, wenn das oder die seltenen Allele eine Häufigkeit von mindestens 1% haben." (Passarge, 1994 quoted from Müller-Myhosk, 2003, p. 98, in: Ganten & Ruckpaul, 2003).

Polymorphisms can affect genes in different ways. Its influence depends on its position within the gene. One very common effect of polymorphisms is an amino acid exchange within the coded protein. These changes lead to a function change (Jacobi et al., 2005, p. 277f).

Genetic polymorphism can occur on different levels. Chromosomal polymorphisms for instance are changes of the structure or the form of a chromosome on a specific locus. Within protein polymorphisms two different types must be distinguished. One possible polymorphism are the marker polymorphisms, the other option are functional polymorphism which can lead to a change of protein function. The last group of polymorphisms are based on the DNA level (Müller-Myhsok, 2003, p.98-99, in: Ganten & Ruckpaul, 2003).

Müller-Myhsok (2003, p.99, in Ganten & Ruckpaul, 2003) points out that the three different types of genetic variations within the DNA sequence are single nucleotide polymorphisms (SNPs), variable number of tandem repeats (VNTRs), and short tandem repeats (STRs).

Evans & Manson (2008, p. 138-139) mention two different types of DNA sequence polymorphisms which are single nucleotide polymorphisms (SNP) and variable number tandem repeats (VNTR). The difference to Müller-Myhsok is that Evans & Manson distinguish within the VNTR between microsatellite DNA and minisatellite DNA. Therefore one can say that there are three different types of genetic variations, no matter if you count STRs as part of VNTRs or not. In the following passage all three types will be described.

SNPs can also be called restriction fragment length polymorphisms (RFLPs). This type of DNA sequence variations occurs when a single nucleotide is changed. These changes occur when base pairs (thymine and guanine, and adenine and cytosine) are exchanged. Those changes can be G→T, T→G, C→A, and A→G. It is estimated that those changes occur approximately once in every 200-500 nucleotides. Up to now more than 100.000 SNPs are known. Many of them are located in non-coding regions of the genome and therefore they are not associated with any important changes. Others are associated with important function changes and therefore they are subject to many different studies. (Evans & Masons, 2008, pp. 138-139; Müller-Myhsok, 2003, p. 99, in: Ganten & Ruckpaul, 2003)

VNTRs usually have a length of about 100 base pairs. According to the number of repetitions different RFPLs can occur. Most VNTRs have more than two alleles. (Evans & Masons, 2008, p. 138; Müller-Myhsok, 2003, p. 99, in: Ganten & Ruckpaul, 2003)

STRs are similar to VNTRs. The main difference is that STRs repeat between one and six base pairs characteristically (Evans & Mason, 2008, p. 138). According to Müller-Myhsok (2003) the repetition length of STRs is either every two, three or four nucleotides. Therefore they can be called dinucleotide marker, trinucleotide marker, and tetranucleotide marker (Müller-Myhsok, 2003, p. 99, in: Ganten & Ruckpaul, 2003).

3. Investigated Polymorphisms and Literature Review

This master thesis deals with possible genetic differences between professional athletes and a control group. The aim of this thesis is to find out why some people are able to compete at a professional level compared to others who cannot participate in such events. Before the investigated polymorphisms will be described, an overview over current researches will be provided.

It has been suggested that genetic variation can make the difference between people who become professional sportsmen and those who do not. Therefore relevant literature was needed in order to support the results. Unfortunately not many researches discussing genetic variations and either soccer or handball have been published. Most studies investigated soccer and different polymorphisms. Only one study dealing with handball and polymorphisms was found. Therefore a short overview about studies which examined soccer will be given.

Subsection 3.3. and 3.4. provide an overview on the investigated polymorphisms (see Table 3: Overview on investigated gene polymorphisms). Also a short introduction into recent literature will be provided. Therefore a literature search on PubMed using the rs-numbers was done.

For a better understanding the polymorphisms (see Table 1) have been divided into two categories. In chapter 3.3 strength performance related polymorphisms are explained and in chapter 3.4. endurance related polymorphisms are illustrated. Neither handball nor soccer is only strength-orientated or endurance-orientated. Therefore it is necessary to look closely at both (all) types of genetic variations.

Table 3: Overview on investigated gene polymorphisms

Gene (full name)	Gene (abbreviation)	Database SNP (rs#)
Alpha-actinin-3	ACTN3	rs1815739
Myostatin	MSTN	rs1805086
Insulin-like growth factor 1	IGF-1	rs35767
Insulin-like growth factor 2	IGF-2	rs3213221

Insulin-like growth factor 2	IGF-2	rs7924316
Adrenergic receptor beta-1	ADRB1	rs1801252
Adrenergic receptor beta-2	ADRB2	rs1042713
Adrenergic receptor beta-2	ADRB2	rs1042714
Nuclear respiratory factor 2	NRF2	rs7181866
Peroxisome proliferator-activated receptor gamma, coactivator 1 alpha	PPARGC1A	rs8192678
Hypoxia inducible factor 1 alpha	HIF1A	rs11549465

3.1. Handball and related genetic variations

Like many other team sports there are many different playing positon such as goalkeepers, backs, pivot players and wings (Chaouachi, Brughelli, Levin, Boudhina, Cronin & Chamari, 2009, p. 153). Body height and fat free body mass are larger in professional handball players compared to recreational handball players. Professional players are able to produce higher mean power (in relation to their body weight) and jump higher than the recreational group (Nikolaidis & Ingebrigtsen, 2013, p. 119). Sprint tests, horizontal jumping tests, endurance test, throwing velocity, and test for muscle strength seem to be appropriated tests to find different performance levels in handball playing positions. However no significant relationship between performance and playing position could be found. Nevertheless difference in height and weight were found (Chaouachi et al., 2009, pp. 152-154). Although some studies investigating performance related topics in handball have been published up to now (September 10th, 2015) only one study dealing with the relationship between genetic variations and handball players was published. Together with 25 cyclists, 20 runners, and 400 healthy controls 15 handball players were genotyped for the angiotensin-I-converting enzyme (ACE)-insertion/deletion polymorphism (ACE). The handball players were all members of the Spanish national team who competed in the 1997 (3rd place) and 1999 (4th place) world

championships. The main finding of this study was that the ACE-I allele had significant higher frequency in athletes than in controls. Therefore the authors assume a relationship between the ACE I allele and elite athlete status. (Alvarez et al., 2000, p. 117-119).

3.2. Soccer and related genetic variations

Soccer has just like Australian Rules football, handball, rugby, and American Football different playing positions (Chaouachi et al., 2009, p. 151). Typical playing positions in soccer are goalkeeper, defender, midfielder, and forward. However Di Salvo, Baron, Tschan, Calderon Montero, Bachl & Pigozzi (2007, p. 223) distinguished defenders between central defenders and external defenders, and midfielder between central midfield players and external midfield players. According to them genetic variations play a role in picking playing positons because of one's physiological characteristics (Di Salvo et al., 2007, p. 224). They conclude that training may not outweigh missing genetic potential (Di Salvo et al., 2007, p. 227). Searching the term "polymorphism AND soccer" on PubMed showed that 31 articles dealing with this topic have been published (as on September 10th, 2015). All findings are presented in Table 2.

Table 4: Studies dealing with soccer and polymorphisms

Study	**Authors (year)**	**Investigated Polymorphisms (rs-number)**
Sport physiology, dopamine and nitric oxide - Some speculations and hypothesis generation.	Landers & Esch (2015)	NOS3-786T/C
Distribution of Angiotensin-1 Converting Enzyme Insertion/Deletion and α-Actinin-3 Codon 577 Polymorphisms in Turkish Male Soccer Players.	Ulucan et al. (2015)	ACTN3, ACE
The Association between MCT1 A1470T Polymorphism and Fat-	Massidda et al. (2015)	MCT1 A1470T

Free Mass in Well-Trained Young Soccer Players.		
Effect of creatine supplementation on physical performance are related to the AMPD1 and PPARG genes polymorphisms in football players.	Lifanov et al. (2014)	C34T AMPD1, Pro12Ala PPARG
The alpha-actinin-3 R577X polymorphism and physical performance in soccer players.	Coelho et al. (2015)	ACTN3
Influence of the COL5A1 rs12722 on musculoskeletal injuries in professional soccer players.	Massidda et al. (2015)	COL5A1 (rs12722)
PPARα gene variants as predicted performance-enhancing polymorphisms in professional Italian soccer players.	Proia et al. (2014)	PPARα intron 7 G/C (rs4253778)
Genomics DNA profiling in elite professional soccer players: a pilot study.	Kambouris et al. (2014)	n.A.
The polygenic profile of Russian football players.	Egorova et al. (2014)	ACE D, ACTN3 Arg577, PPARA (rs4253778) C, UCP2 55Val
Variation in the ACE, PPARGC1A and PPARA genes in Lithuanian football players.	Gineviciene et al. (2014)	ACE (I/D), PPARGC1A (G/A), PPARA (G/C)

New genetic model for predicting phenotype traits in sports.	Massidda et al. (2014)	ACE, ACTN-3, BDKRB2, VDR-ApaI, VDR-BsmI, VDR-FokI
Single nucleotide polymorphisms associated with non-contact soft tissue injuries in elite professional soccer players: influence on degree of injury and recovery time.	Pruna et al. (2013)	ELN, TNC, TTN, SOX15, IGF2, CCL2, COL1A1, COL5A1,
Effect of ACTN3 gene on strength and endurance in soccerplayers.	Pimenta et al. (2013)	ACTN3
American Medical Society for Sports Medicine position statement: concussion in sport.	Harmon et al. (2013)	APOE G-219T
Effect of astaxanthin supplementation on paraoxonase 1 activities and oxidative stress status in young soccer players.	Baralic et al. (2013)	n.A.
Gene variants within the COL1A1 gene are associated with reduced anterior cruciate ligament injury in professional soccerplayers.	Ficek et al. (2013)	COL1A1 -1997G/T, COL1A1 +1245G/T
Genetic markers and explosive leg-muscle strength in elite Italiansoccer players.	Massidda et al. (2012)	ACTN3, ACE, BDKRB2

The C allele in NOS3 -786 T/C polymorphism is associated with elite soccer player's status.	Eynon et al. (2012)	NOS3-786 T/C (rs2070744)
The ACTN3 genotype in soccer players in response to acute eccentric training.	Pimenta et al. (2012)	ACTN3
Angiotensin-converting enzyme/vitamin D receptor gene polymorphisms and bioelectrical impedance analysis in predicting athletic performances of Italian young soccer players.	Micheli et al. (2012)	ACE, VDR
Apolipoprotein E genotype and concussion in college athletes.	Tierney et al. (2010)	APOE2 Arg158Cys, APOE4 Cys112Arg
Apolipoprotein E genotyping and concussion: time to fish or cut bait.	Gordon (2010)	APOE
Distribution of the Trp64Arg polymorphism in the ß 3-adrenergic receptor gene in athletes and its influence on cardiovascular function.	Kim et al. (2010)	ADRB3 Trp64Arg
Vitamin D receptor gene FokI polymorphisms influence bone mass in adolescent football (soccer) players.	Diogenes et al. (2010)	VDR
Influence of angiotensinogen and angiotensin-converting enzyme polymorphisms on cardiac hypertrophy and improvement on maximal aerobic capacity caused by exercise training.	Alves et al. (2009)	angiotensinogen M235T, ACE I/D

Steroid profiles of professional soccer players: an international comparative study.	Strahmet al. (2009)	UGT2B17
Genotype distributions in top-level soccer players: a role for ACE?	Juffer et al. (2009)	ACE I/D, GDF-8K153R, GDF-8E164K, GDF-8P198A, GDF-8I225T, AMPD1 C34T
APOE, APOE promoter, and Tau genotypes and risk for concussion in college athletes.	Terrell et al. (2008)	APOE promoter G-219T
The distribution of I/D polymorphism in the ACE gene among Korean male elite athletes.	Oh (2007)	ACE I/D
ACE I/D polymorphism and cardiac adaptations in adolescent athletes.	Rizzo et al. (2003)	ACE I/D
RAS genes influence exercise-induced left ventricular hypertrophy: an elite athletes study.	Fatini et al. (2000)	ACE I/D, AT1R A1166C, AT1R CA

The findings of some of the studies mentioned in Table 2 will be presented exemplarily in the following passages.

In 2009 a study was published by Juffer et al. investigating the relationship between ACE and elite sport status. In this study male soccer players, endurance athletes and a control group were compered. The main findings were that soccer players, compared to endurance athletes, had a higher frequency of the ACE ID polymorphism. The frequency of the ACE II polymorphism was in soccer players lower than in endurance athletes. The

authors conclude that for soccer a phenotype associated with strength and speed-strength is beneficial (Juffer et al., 2009).

Similar results were published by Micheli et al. in 2011. They found a high correlation between the frequency of the ACE ID polymorphism and jumping performances (squat jump, counter movement jump) in young soccer players. Players who were tested heterozygous scored better results in jumping tests than those who were not tested positive for the ACE ID polymorphism. All results were compared to a control group. This comparison leads to the conclusion that the ACE ID polymorphism effects strength and speed-strength positively (Micheli et al., 2011, p. 2084). Micheli et al. drew a final conclusion: “Our results suggest that determination of ACE and VDR genotypes might help select those young athletes harboring the most favorable genetic potential to succeed in soccer.” (Micheli et al., 2011, p. 2084)

Comparable results were found by Massidda et al. (2012). They found a strong relationship between the frequency of certain ACE and ACTN3 polymorphisms and the performance in jumping tests. Nevertheless they conclude that genetic variations do not have a big impact on soccer performance. Furthermore they suggest that the RR genotype of the ACTN3 can be associated with elite gymnast status. No differences in frequency of ACE and ACTN3 polymorphisms between Soccer players and a control group could be found (Massidda, et al., 2012, p. 51).

Another recently published study suggests that there is no relationship between the ACTN3 genotype and athletic performance in Brazilian soccer players (Coelho et al., 2015).

3.3. Polymorphisms associated with strength performance

The following four genes have been associated with strength performance and anaerobic capacity. Table 2 shows an overview of the relevant polymorphisms and its functions. For the IGF-2 two different locations (rs3213221 and rs7924316) were investigated.

Table 5: Investigated polymorphisms associated with strength and its influence

Gene (abbreviation)	Database SNP (rs#)	Function
ACTN3	rs1815739	Muscle performance and strength

MSTN	rs1805086	Development of muscles, response to training and peak power
IGF-1	rs35767	Growth factor activity
IGF-2	rs3213221	Cleavage
IGF-2	rs7924316	Cleavage

3.3.1. Alpha-actinin-3 (rs1815739)

The 577 polymorphism of the ACTN3 can occur in three different versions: R577R, R577X and X577. The polymorphism R577X is a transition substitution from CGA to TGA. TGA is a stop codon, therefore this change results in a nonsense function of the ACTN3 (AppliedBiosystems, 2015b; NCBI, 2015f).

The ACTN3 R577X polymorphism (rs1815739) has been associated with strength and power performance and has been well investigated. Nevertheless there are big differences between studies according to sample size, study design, ethnic background, and sport status (Eynon et al., 2012, p. e43132).

3.3.2. Myostatin (rs1805086)

The K153R polymorphism of the MSTN gene is an amino acid exchange of lysine to arginine. This allele change from AAG to AGG is a transition substitution and results in a missense function (AppliedBiosystems, 2015a; NCBI, 2015e).

According to Santiago et al. (2011, p. e16323) the MSTN K153R polymorphism is a candidate genetic variations to influence skeletal muscle phenotypes. Garatachea et al. (2013, p. 2445) found a possible relationship between the K153R polymorphism and longevity due to its role for muscular phenotypes and sarcopenia.

3.3.3. Insulin-like growth factor 1 (rs35767)

The selected polymorphism -1246C/T (rs 35767) in the gene coding for IGF-1 is a transition substitution of C by T. Up to now no clinical association could be found for this nearGene-5 polymorphism (AppliedBiosystems, 2015c; NCBI, 2015a).

Ben-Zaken et al. (2014, p. 470) explain that the IGF-1 -1246C/T polymorphism is associated with top performances in endurance and strength related sports. An influence of the TT genotype on an increased osteoporosis risk in Chinese women was found by Zhang et al. (2015, p. 7655).

3.3.4. Insulin-like growth factor 2 (rs3213221)

The C13790G polymorphism of the IGF-2 (rs3213221) gene is a transversion substitution of C by G. This polymorphism is located in the intron region of the gene and has an unknown clinical association (AppliedBiosystems, 2015j; NCBI, 2015g).

The IGF-2 C13790G polymorphism is assumed to influence muscular damage such as muscle soreness, sarcopenia, higher CK levels (Devaney et al., 2007, p. 1815).

3.3.5. Insulin-like growth factor 2 (rs7924316)

The G11711T polymorphism (rs7924316) coding for the IGF-2 gene is a transversion substitution of G by T in the intron region of this gene with an unknown clinical significance (AppliedBiosystems, 2015i; NCBI, 2015h).

Like the IGF-2 C13790G polymorphism the IGF-2 G11711T polymorphism is associated with muscular damage (Devaney et al., 2007, p. 1815).

3.4. Polymorphisms associated with endurance performance

The following 5 genes have been associated with endurance performance. Table 3 shows an overview of the relevant polymorphisms and its functions. The ADRB2 was investigated on two different locations (rs1042713 and rs1042714).

Table 6: Investigated polymorphisms associated with endurance and its influence

Gene (abbreviation)	Database SNP (rs#)	Function
ADRB1	rs1801252	Cardiac control
ADRB2	rs1042713	Metabolism, obesity, asthma and relaxation of nonstriated muscles
ADRB2	rs1042714	Metabolism, obesity and asthma and relaxation of nonstriated muscles
NRF2	rs7181866	Transcription factor
PPARGC1A	rs8192678	Mitochondrial biogenesis
HIF1A	rs11549465	Transcription factor (oxygen sensor)

3.4.1. Adrenergic receptor beta-1 (rs1801252)

The Ser49Gly polymorphism of the MSTN gene is an amino acid exchange of serine to glycine. This allele change from AGC to GGC is a transition substitution and results in a missense function (AppliedBiosystems, 2015g; NCBI, 2015b).

Adrenergic receptor beta-1 (ADRB1) is an important genotype for cardiac control. This factor leads to ADRB1 important role when it comes to athletic performance. Dependent on the different variations of the Ser49Gly polymorphism it influences human heart rate. The polymorphism is responsible for a high or a low heart rate. These changes are independent from age, sex and BMI. Therefore ADRB1 is associated with endurance athlete status in different publications (Nieminen et al., 2006; Ranade et al., 2002)

3.4.2. Adrenergic receptor beta-2 (rs1042713)

The selected polymorphism rs1042713 in the gene coding for ADRB2 is a transition substitution of arginine to glycine. The allele change from AGA to GGA results in a missense function (AppliedBiosystems, 2015e; NCBI, 2015b).

According to Wolfarth et al. (2000) the R16G (Arg16Gly) polymorphism of the ADRB2 gene can be associated with endurance performance. They note that this polymorphism

affects the cardiovascular system as well as the regulation of lipolysis (Wolfarth et al., 2000, p. 1711). In the cardiovascular system the ADRB2 are associated with arterial vasodilation and myocardial contractility (Wolfarth et al., 2007, p. 1651). Additionally Garenc et al. (2003, p. 615) found a connection between the Arg16Gly polymorphism and obesity in white women. Obese white women carrying the Gly16Gly (G16G) expression of this polymorphism tend to have less fat accumulation than those with other genotypes (Garenc et al., 2003, 613).

3.4.3. Adrenergic receptor beta-2 (rs1042714)

The Q27E (Gln27Glu) polymorphism of the ADRB2 gene is an amino acid exchange of glutamine (Gln) to glutamic acid (Glu). This transversion substitution from CAA to GAA leads to a missense function (AppliedBiosystems, 2015f; NCBI, 2015c).

Garenc et al. (2003, p. 612) found a relationship between white men and their amount of body fat. This assumption counts for men only because they could not find any connection between this polymorphism, obesity and women (Garenc et al., 2003, p. 613). The authors added the information that those results only count for white men and not for blacks. Therefore they conclude that these findings are race-specific (Garenc et al., 2003, p. 613).

3.4.4. Nuclear respiratory factor 2 (rs7181866)

The polymorphism rs7181866 in the coding for NRF2 is a transition substitution of A by G. This polymorphism is located in the intron region of the gene and has an unknown clinical association (AppliedBiosystems, 2015k; NCBI, 2015i).

Associations between NRF2 and the induction of mitochondrial biogenesis as well as in nucleo-mitochondrial interactions (Enyon et al., 2009; Kanaki et al., 2004; Kelly, D.P. & Scarpulla, R.C., 2004). Enyon et al. (2009, p. 79) found a correlation between Israeli elite endurance athletes and the G allele of the NRF2 AG polymorphism. Nevertheless they note that further studies have to be done to get clear and better results.

3.4.5. Peroxisome proliferator-activated receptor gamma, coactivator 1 alpha (rs8192678)

The selected polymorphism G482S coding for the gene PPARGC1A is transition substitution of G by A. The allele change from GGT to AGT results in an amino acid exchange of glycine to serine. These changes result in a missense function. (AppliedBiosystems, 2015d; NCBI, 2015j).

PPARGC1A carrying allele A can lead to higher triacylglycerols and therefore it can be suggested that it increases the possibility of cardiovascular diseases as well as type 2 diabetes in Brazilian children (Queiroz et al., 2015, p. 595). Additionally it is assumed that, because of its influence on mitochondrial biogenesis, it effects the respond to cardiovascular training (Liang & Ward, 2006, p. 145).

3.4.6. Hypoxia inducible factor 1 alpha (rs11549465)

The P582S polymorphism in the coding for the HIF1A gene is an amino acid exchange of proline to serine. The allele change from CCA to TCA is transition substitution and leads to a missense function (AppliedBiosystems, 2015h; NCBI, 2015k).

The HIF1A Pro582Ser polymorphism has been associated with an improvement of glucose metabolism and cell proliferation and differentiation (Gabbasov et al., 2013, p. 2055). Bahadori et al. (2010) explain an influence of the P582S polymorphism on inflammation and therefore on peripheral artery diseases.

4. Methods

This chapter intends to provide the reader information about subject recruitment, DNA-analysis, and statistical analysis.

4.1. Subjects

Subjects were recruited by the Institute of Sport Science (University of Vienna). Prior to the collection of samples the permission of the ethical commission (Medical University of Vienna) was necessary. Persons who wanted to participate in this study had to complete a questionnaire (questions concerning sports and lifestyle) and had to sign a consent form. All relevant documents are attached to this master thesis. Altogether DNA samples of 470 men and women were collected. Of those 470 people, 235 persons competed on a professional level in different types of sports. 235 people were tested as part of the control group.

Every person who participated in this research and who provided saliva samples was a randomly picked number assigned. This procedure was used to guarantee the anonymity of all subjects. Saliva samples as well as the questionnaire are only marked with the number. This ensures that afterwards only sex, gender, and the answers of the questionnaire (Klepic, 2011, p. 47.)

According to Brandstetter (2009, p. 26) several exclusion criteria for athletes were set. A modified version can be seen in Table 4.

Table 7: Exclusion criteria for athletes (modified from Brandstetter, 2009, p. 26.)

- Proven intake of performance-enhancing substances (WADA)
- Existence of coronary heart diseases
- Existence of chronic diseases
- Existence of a manifest diabetes mellitus type I (insulin-dependent) or type II. A manifest type II diabetes mellitus is given if fasting blood glucose levels are higher than 126 mg/dl and there appear typical symptoms as polyuria and thirst at two different days or fasting blood glucose levels are higher than 126 mg/dl and not-fasting blood glucose exceeds 200 mg/dl at two different days.
- Must compete in either soccer or handball professionally

Brandstetter (2009, p. 26) has also described several important inclusion criteria for the control group. The modified inclusion criteria is presented in Table 5.

Table 8: Inclusion criteria for the control group (modified from Brandstetter, 2009, p. 26)

- Good health status
- No existence of a manifest diabetes mellitus type I (insulin-dependent) or type II.
- No known coronary heart diseases or other chronical disorders
- no professional sport engagement
- At least 18 years old
- Must not have competed in any sports on a professional level

After dismissing non-relevant samples a total of 234 samples remained to be relevant for this master thesis. 119 played either soccer (n=81) or handball (n=38) on a professional level. The control group consisted of 115 men who play sports on a non-professional level. Percentage distributions show that 49.1% of all subjects belong to

the control group and that 50.9% are part of the group of professional athletes. Within in the professional athletes group 68.1% of the subjects play soccer on a professional level whereas only 31.9% play handball on a professional level.

4.2. Methodology

Saliva samples from all 234 subjects were collected at the Institute of Sport Science (University of Vienna). After collecting saliva samples via mouthwash the DNA was extracted. The following steps had to be completed to get good results.

1. Collect saliva in 10ml Listerine mouthwash solution (spill for 30s) (subject must not consume any food or drinks 30min prior to sample collection)
2. Add 10ml HBSS -/-
3. Centrifuge at 2000xg for 10’ at RT, carefully decant supernatant
4. Wash with 5ml HBSS -/-
5. Centrifuge at 1800xg for 5’ at RT, decant supernatant (rest with pipette)
6. Fill up to 400µ with HBSS -/-, mix with pipette
7. Transfer 2 x 200µl to 2 sample tubes CB
8. Transfer to QIA Cube
   Run the protocol on QIA Cube (takes about 30 minutes): DNA QIAamp Mini Kit
   Blood or body fluid
   Standard
9. After finishing take out the rotor adapters, close collection tubes, place spin columns onto clean collection tubes
10. Pipet 200µl buffer AE onto the membrane
11. Incubate for 1’ at RT
12. Centrifuge at 6000xg for 1’
13. Vortex and spin down shortly
14. Measure with NanoDrop
15. Aliquot in 0,6ml tubes (50µl), store at -80°C

At step 14 the DNA concentration was measured. This was necessary in order to get comparable results. For each polymorphism 4 different DNA concentrations were tested. Levels for these tests were set at 1ng, 5ng, 10ng and 20ng DNA for each reaction.

The results for the best DNA concentration are presented in the table below (see Table 7).

Table 9: DNA-concentration for all relevant polymorphisms

Polymorphism	DNA-concentration in ng
Adrenoceptor β-1 (rs1801252)	5
Adrenoceptor β-2 (rs1042713)	1
Adrenoceptor β-2 (rs1042714)	5
Nuclear respiratory factor 2 (rs7181866)	10
PPARG coactivator 1α (rs8192678)	5
HIF1A (rs11549465)	5
ACTN3 (rs1815739)	1
Myostatin (rs1805086)	5
IGF-1 (rs35767)	10
IGF-2 (rs3213221)	1
IGF-2 (rs7924316)	5

4.3. Polymerase Chain Reaction

In the last decade real-time polymerase chain reaction (PCR) has been established as the state of the art technology of microbiology research (Bustin, Zaccara & Nolan, 2012, p. 3). The goal of PCR is to explore the characteristics of a single nucleic acid. Nowadays PCRs are fluorescence-based which means that one fluorescent dye detector matches perfectly to allele 1 and one fluorescent dye detector matches perfectly to allele 2. Allele 1 is called “wild type” and allele 2 is called “mutation” (AppliedBiosystems, 2010, p. 3).

There are three possible results of PCR. Option 1 and Option 2 are that the results are homozygous. In this case samples have either only the wild type or the mutation. Option 3 is that the results are heterozygous. In this case samples have the wild type as well as the mutation (AppliedBiosystems, 2010, p. 3).

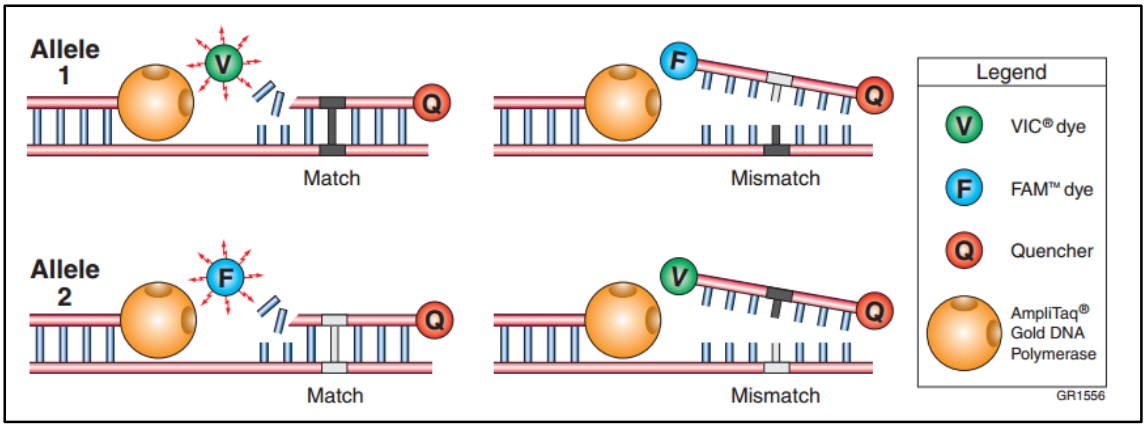

Figure 10: Matches and mismatches between target and probe sequences in TaqMan® SNP Genotyping Assays (AppliedBiosystems, 2010, p. 3)

Figure 1 shows the principle of PCR. If the fluorescent dye matches perfectly to either the wild type allele or the mutation allele the DNA single strands can be replicated. In case of a match the Quencher is used to delete the fluorescent dye. After this procedure the fluorescent dye starts to fluorescent and therefore it can be measured. Nevertheless it is necessary to repeat this process several times to get a strong signal of the fluorescent dye detector. If there are higher levels of the VIC® dye the samples are homozygous for allele 1. If the level of FAM™ dye is higher the probes are homozygous for allele 2. In case that both signals are at the same level it is an indication for a heterozygosity (AppliedBiosystems, 2010, p. 3).

According to Bustin et al. (2012, p. 21) several different factor can influence the PCR positively and negatively. Factors such as analysis method, sample quality and reporting details play an important role in the PCR process.

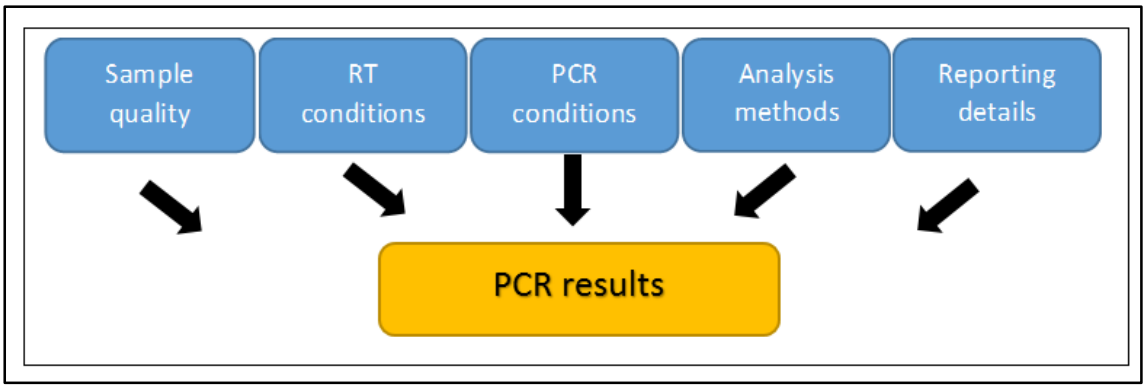

Figure 11: Factors for good PCR results (modified from Bustin et al., 2012, p. 21)

4.4. Statistical procedures

All statistical analysis were calculated using SPSS 23 for Windows XP/Vista/7/8/8.1. The level of significance (P-value) was set at 0.05. This means every value larger than 0.05 is significant. If the calculated value is larger than 0.05 the null hypothesis (H0) has to be dismissed and the alternative hypothesis (Ha) has to be accepted. If the calculated is below the significance level of 0.05 the H0 has to be accepted. (Moore & McCabe, 2006, p. 406-407). According to Moore and McCabe the statistical significance can be defined in the following way: “If the P-value is as small or smaller than α, we say that the data are statistically significant at level α.” (Moore & McCabe, 2006, p. 407)

For all questions Chi-square tests were performed. This test is used to determine, if there is any relationship between different datasets (Moore & McCabe, 2006, p. 595-596).

Table 10: Chi-square statistic (Moore and McCabe, 2006, p. 596)

“The chi-square statistic is a measure of how much the observed cell counts in a two-way table diverge from the expected cell counts. The formula for this statistic is $$X^2 = \sum \frac{(observed\ count - expected\ count)^2}{expected\ count}$$ where “observed” represents an observed sample count, “expected” represents the expected count for the same cell, and the sum is over r x cells in the table.”

The H0 is that there is no relationship between the row and column variables. The H1 is that there is a relationship between the row and column variables (Moore & McCabe, 2006, p. 597).

As mentioned in chapter 1 histograms will be used to illustrate the percentage distribution of the different genetic variations. This method is used to help the reader understand the statistics more easily (Schendera, 2015, p. 232)

5. Results

First the data and calculation for the control and the professional athletes group will be presented. Afterwards the statistical analysis for the type of sports (soccer and handball) and its relationship to the genetic variations will be shown.

5.1. Adrenoceptor β-1 (rs1801252)

5.1.1. ADRB1 (rs1801252) in athletes and controls

In table 9 the distribution of the ADRB1 Ser49Gly (rs1801252) is presented. Six subjects were not tested for this genetic variation. Usually there are three different polymorphisms (AA, AG and GG) of this gene. There are just small differences between the expected and the measured frequencies. Nevertheless it is notable that none of the subjects was tested homozygous for the GG allele of the ADRB1 Ser49Gly (rs1801252).

Table 11: ADRB1 Ser49Gly (rs1801252) distribution in athletes and controls

			ADRB1 Ser49Gly (rs1801252)		
			AA	AG	Total
Group	Untrained	Count	77	32	109
		Expected Count	79,8	29,2	109,0
	Professional Athletes	Count	90	29	119
		Expected Count	87,2	31,8	119,0
Total		Count	167	61	228
		Expected Count	167,0	61,0	228,0

The percentage distribution of the ADRB1 (rs1801252) shows only small variances between the professional athletes and the control group. Professional athletes have a higher frequency of the AA genotype (75.63%) than controls (70.64%). The control group

has a higher frequency of the AG genotype (29.36%) than professional athletes (24.37%). For the histogram of this distribution see Figure 1.

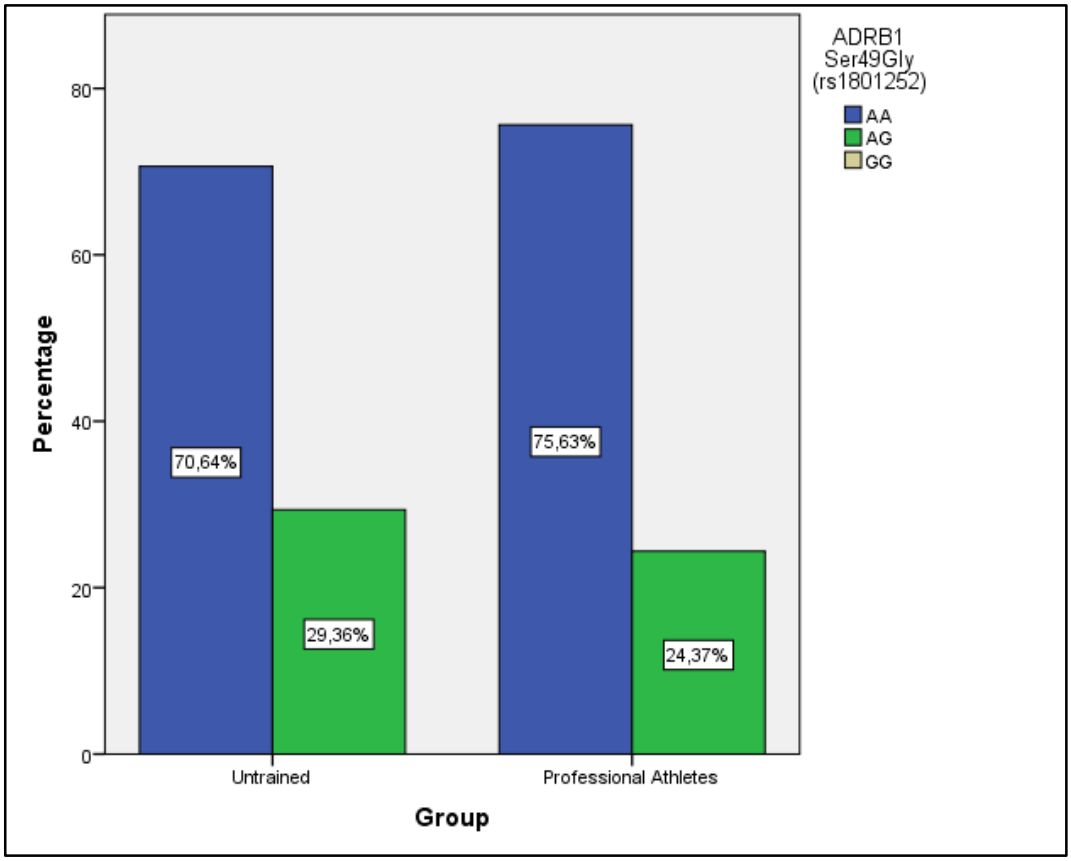

Figure 12: Percentage distribution of ADRB1 Ser49Gly (rs1801252) in athletes and controls

For the Chi square test the following null hypothesis was set up.

H0: There is no significant relationship between the level of sports and the genetic variation of the ADRB1 Ser49Gly (rs1801252).

The result of the Chi square test shows a significance level of 0.395. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the level sports and the genetic variation of the ADRB1 Ser49Gly (rs1801252).

5.1.2. ADRB1 (rs1801252) in soccer and handball players

All professional athletes were tested for this genetic variation. Table 12 provides information about the distribution of the ADRB1 Ser49Gly (rs1801252). AA frequency is in soccer players bigger than expected and AG frequency is lower than expected. Bigger frequencies of AG and lower frequencies of AA than expected were tested in handball players. No player was tested positive for the GG polymorphism of the ADRB1.

Table 12: ADRB1 Ser49Gly (rs1801252) distribution in soccer and handball players

			ADRB1 Ser49Gly (rs1801252)		
			AA	AG	Total
Sport	Soccer	Count	65	16	81
		Expected Count	61,3	19,7	81,0
	Handball	Count	25	13	38
		Expected Count	28,7	9,3	38,0
Total		Count	90,0	29	119
		Expected Count	90,0	29,0	119,0

The percentage distribution of the ADRB 1 (rs1801252) show that handball players (34.21%) have higher frequencies of the AG polymorphism than soccer players (19.75%). Soccer players (80.25%) have higher frequencies of the AA polymorphism than handball players (65.79%). Neither one of the soccer players nor one of the handball players was tested positive for the GG polymorphism. Detailed information can be seen in Figure 2.

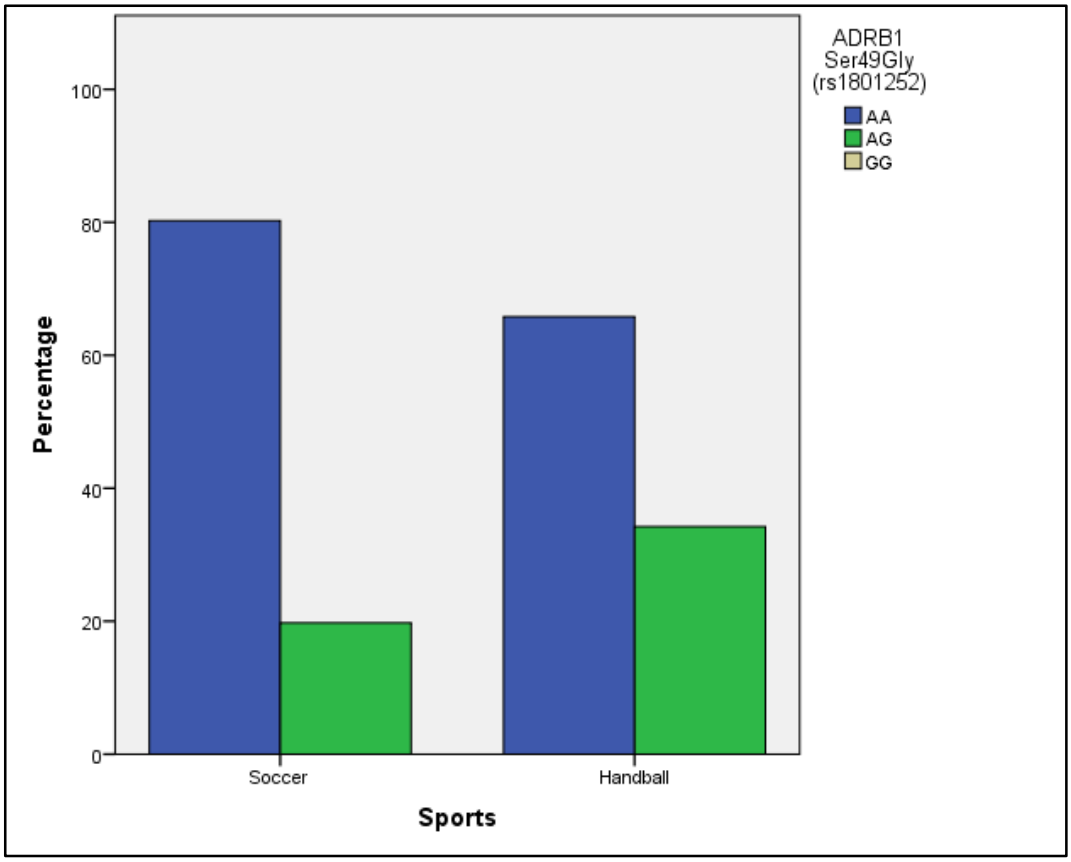

Figure 13: Percentage distribution of ADRB1 Ser49Gly (rs1801252) in soccer and handball players

To find out if there is any relationship between the type of sports and the genetic variation, the following null hypothesis was set up.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the ADRB1 Ser49Gly (rs1801252).

The result of the Chi square test shows a significance level of 0.087. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between type of sports and the genetic variation of the ADRB1 Ser49Gly (rs1801252) in professional athletes.

5.2. Adrenoceptor β-2 (rs1042713)

5.2.1. ADRB2 (rs1042713) in athletes and controls

The ADRB2 Arg16Gly (rs1042713) was tested in 228 subjects. 6 subjects of the control group were not tested for the polymorphisms of this gene. The frequency of the ADRB2 Arg16Gly (rs1042713) polymorphisms is presented in table 11. The three possible genetic variations of this gene are AA (homozygous), AG (heterozygous) and GG (homozygous). There are just small differences between the expected and the measured frequencies. It is interesting to see that more controls are homozygous to GG and less controls heterozygous to AG than expected. It seems that athletes are more likely to be heterozygous for AG and less likely to be homozygous for GG.

Table 13: ADRB2 Arg16Gly (rs1042713) distribution in athletes and controls

			ADRB2 Arg16Gly (rs1042713)			
			AA	AG	GG	Total
Group	Untrained	Count	14	52	43	109
		Expected Count	13,9	55,5	39,7	109,0
	Professional Athletes	Count	15	64	40	119
		Expected Count	15,1	60,5	43,3	119,0
Total		Count	29	116	83	228
		Expected Count	29,0	116,0	83,0	228,0

According to the percentage distribution only little differences in the frequency of AA were measured between professional athletes (12.61%) and controls (12.84%). Variances were measured in the frequencies of the AG and AA polymorphisms. As one can see in Figure 3 53.78% of athletes were tested positive for the AG polymorphism compared to 47.71% of the controls. The GG genotype seems to occur more likely in controls (39.45%) than in professional athletes (33.61%).

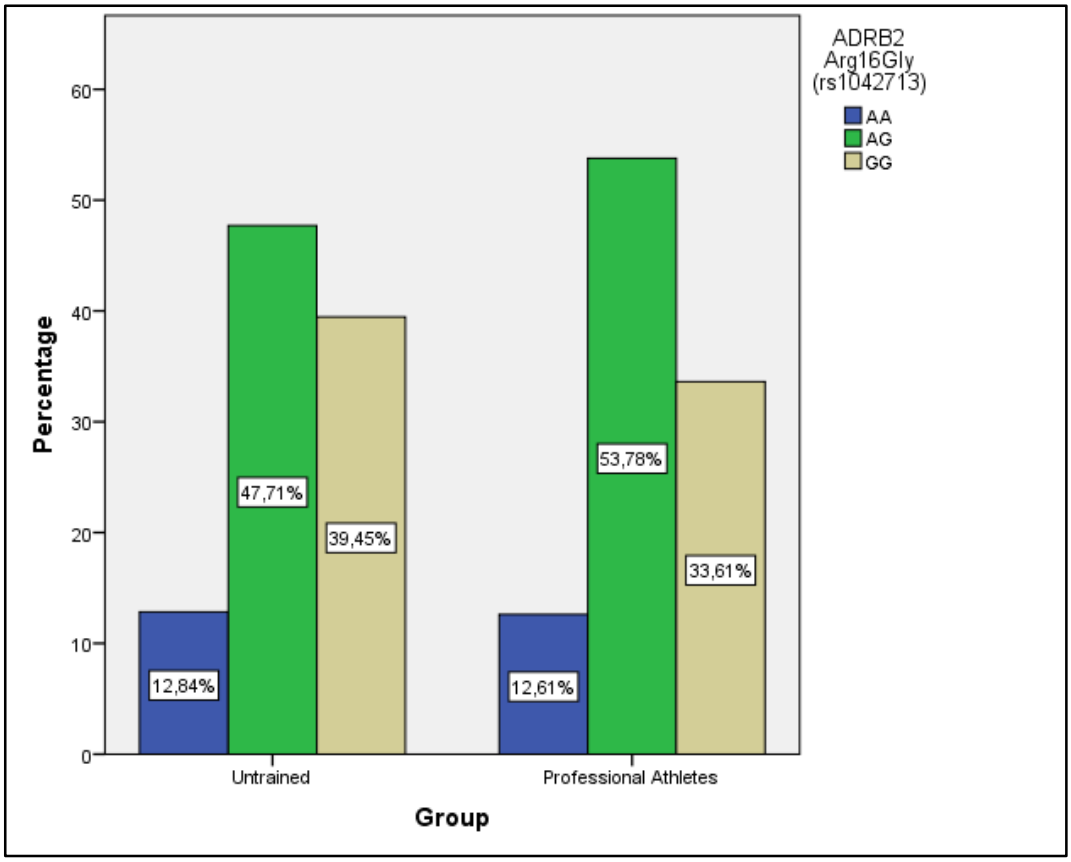

Figure 14: Percentage distribution of ADRB2 Arg16Gly (rs1042713) in athletes and controls

To calculate the Chi square test the following null hypothesis was built.

H0: There is no significant relationship between the level of sports and the genetic variation of the ADRB2 Arg16Gly (rs1042713).

The result of the Chi square test shows a significance level of 0.623. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the level of sports and the genetic variation of the ADRB2.

5.2.2. ADRB2 (rs1042713) in soccer and handball players

The ADRB2 Arg16Gly (rs1042713) was tested in all professional athletes (n=119). The frequency of the ADRB2 Arg16Gly (rs1042713) polymorphisms is presented in Table 14. Only little differences between expected count and measured count can be seen. Soccer

players have higher frequencies of the AA than expected. AG and GG polymorphisms were counted more than expected in handball players.

Table 14: ADRB2 Arg16Gly (rs1042713) distribution in soccer and handball players

			ADRB2 Arg16Gly (rs1042713)			
			AA	AG	GG	Total
Sport	Soccer	Count	9	44	28	81
		Expected Count	10,2	43,6	27,2	81,0
	Handball	Count	6	20	12	38
		Expected Count	4,8	20,4	12,8	38,0
Total		Count	15	64	40	119
		Expected Count	15,0	64,0	40,0	119,0

The percentage distribution of ADRB2 (rs1042713) show higher frequencies of GG and AG in soccer players (54.32% and 34.57%) than in handball players (52.63% and 31.58%). Higher frequencies of the AA polymorphism were measured in handball players (15.79%) than in soccer players (11.11%). For further details see Figure 4.

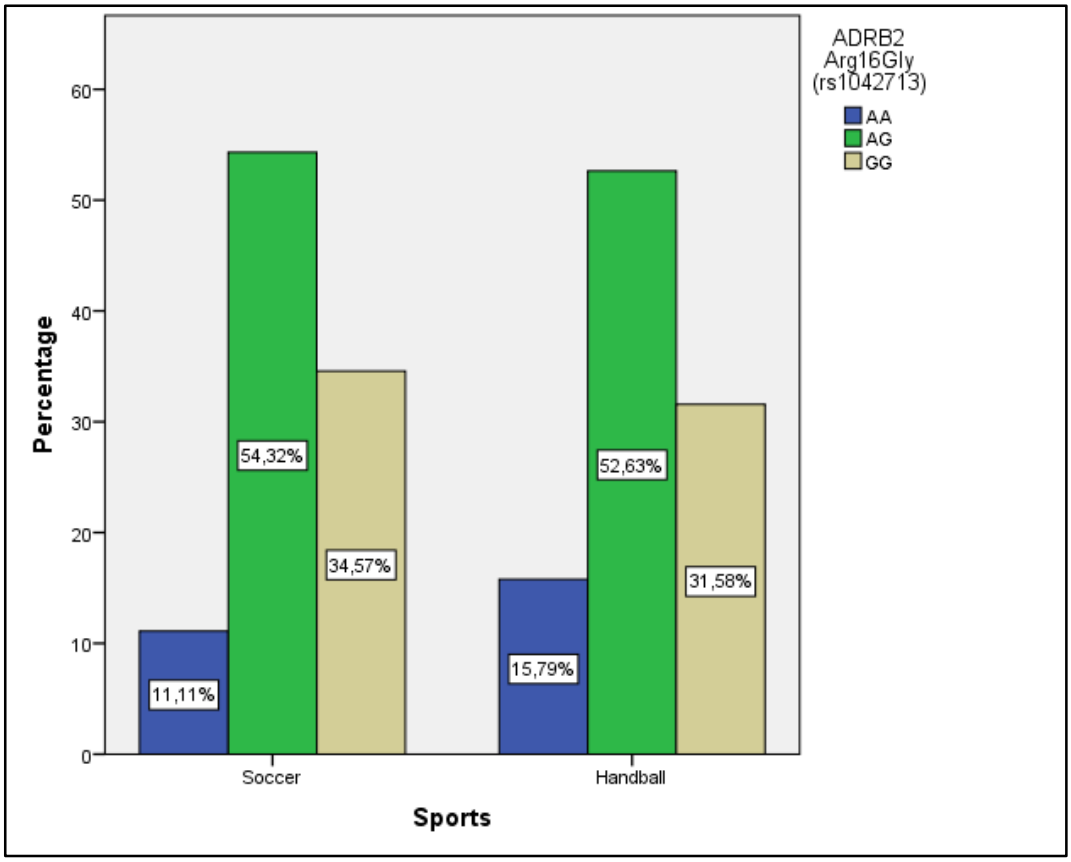

Figure 15: Percentage distribution of ADRB2 Arg16Gly (rs1042713) in soccer and handball players

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the ADRB2 Arg16Gly (rs1042713).

The result of the Chi square test shows a significance level of 0.767. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between type of sports and the genetic variation of the ADRB2 Arg16Gly (rs1042713) in professional athletes.

5.3. Adrenoceptor β-2 (rs1042714)

5.3.1. ADRB2 (rs1042714) in athletes and controls

Six men of the control group were not tested for this gene expression. The frequency of the ADRB2 (rs1042714) are presented in Table 15. The Polymorphism Gln27Glu can appear in three different variations. Homozygous for allele C (CC), homozygous for allele G (GG) and heterozygous (CG). More professional athletes were tested homozygous for C than expected. More controls than expected were tested positive for the CG and GG polymorphisms.

Table 15: ADRB2 Gln27Glu (rs1042714) distribution in athletes and controls

			ADRB2 Gln27Glu (rs1042714)			
			CC	CG	GG	Gesamt
Group	Untrained	Count	49	42	18	109
		Expected Count	43,0	46,9	19,1	109,0
	Professional Athletes	Count	41	56	22	119
		Expected Count	47,0	51,1	20,9	119,0
Total		Count	90	98	40	228
		Expected Count	90,0	98,0	40,0	228,0

The frequency distribution in Figure 16 shows that more professional athletes (47.06%) are heterozygous (CG) than controls (38.53%). More controls (44.95%) are homozygous for allele C than athletes (34.45%). Little differences occur in the frequency of men tested homozygous for the G allele. 18.49% of athletes and 16.51% of controls were tested positive for the GG expression of this gene.

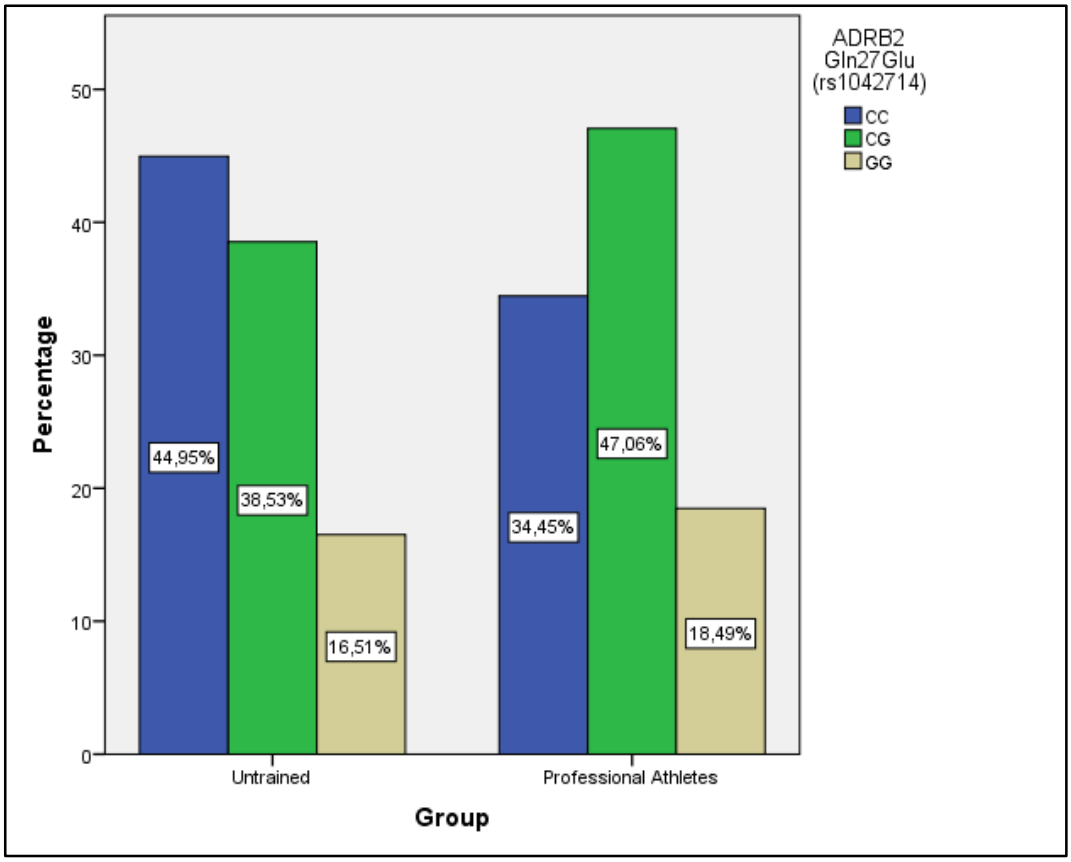

Figure 16: Percentage distribution of ADRB2 Gln27Glu (rs1042714) in athletes and controls

H0: There is no significant relationship between the level of sports and the genetic variation of the ADRB2 Gln27Glu (rs1042714).

The result of the Chi square test shows a significance level of 0.262. This value is not significant, therefore the H0 cannot be dismissed.

Result: No significant relationship between the level of sports and the ADRB2 variation could be found.

5.3.2. ADRB2 (rs1042714) in soccer and handball players

All soccer and handball players were tested for the ADRB2 Gln27Glu (rs1042714). No differences between expected count and measured count can be recognized (see Table 16).

Table 16: ADRB2 Gln27Glu (rs1042714) distribution in Soccer and Handball players

			ADRB2 Gln27Glu (rs1042714)			
			CC	CG	GG	Total
Sport	Soccer	Count	28	38	15	81
		Expected Count	27,9	38,1	15,0	81,0
	Handball	Count	13	18	7	38
		Expected Count	13,1	17,9	7,0	38,0
Total		Count	41	56	22	119
		Expected Count	41,0	56,0	22,0	119,0

The percentage distribution (see Figure 17) shows only little differences (max. 0.46%).

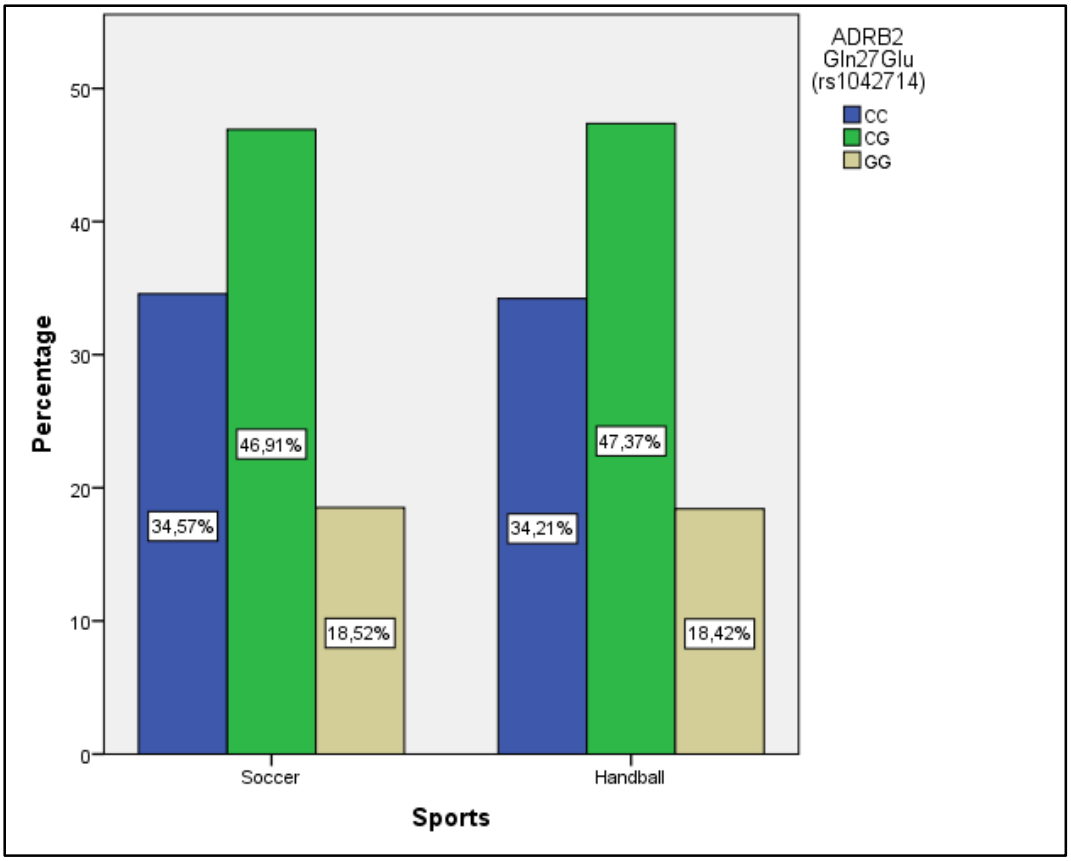

Figure 17: Percentage distribution of ADRB2 Gln27Glu (rs1042714) in soccer and handball players

For the Chi square test the following null hypothesis was set up.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the ADRB2 Gln27Glu (rs1042714).

The result of the Chi square test shows a significance level of 0.999. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the type of sports and the genetic variation.

5.4. Nuclear respiratory factor 2 (rs7181866)

5.4.1. NRF2 (rs7181866) in athletes and controls

A total number of 51 men were not tested for this genetic variation. Therefore only data of 183 subjects is available. Frequencies (expected counts and measured counts) can be seen in Table 17. Of the three possible genetic expressions (AA, AG, and GG) only two occur. No men, neither professional athletes nor control, was tested homozygous for the G allele.

Only small differences between expected and measured counts can be observed.

Table 17: NRF2 intron 3 A/G (rs7181866) distribution in athletes and controls

			NRF2 intron 3 A/G (rs7181866)		
			AA	AG	Gesamt
Group	Untrained	Count	107	2	109
		Expected Count	106,6	2,4	109,0
	Professional Athletes	Count	72	2	74
		Expected Count	72,4	1,6	74,0
Total		Count	179	4	183
		Expected Count	179,0	4,0	183,0

The histogram (see Figure 18) shows that the AA polymorphism dominates both groups. The control group has slightly more men homozygous for allele A (98.17%) than the professional athletes (97.30%). Athletes (2.70%) have a marginal higher percentage of men tested heterozygous (AG) than controls (1.83%).

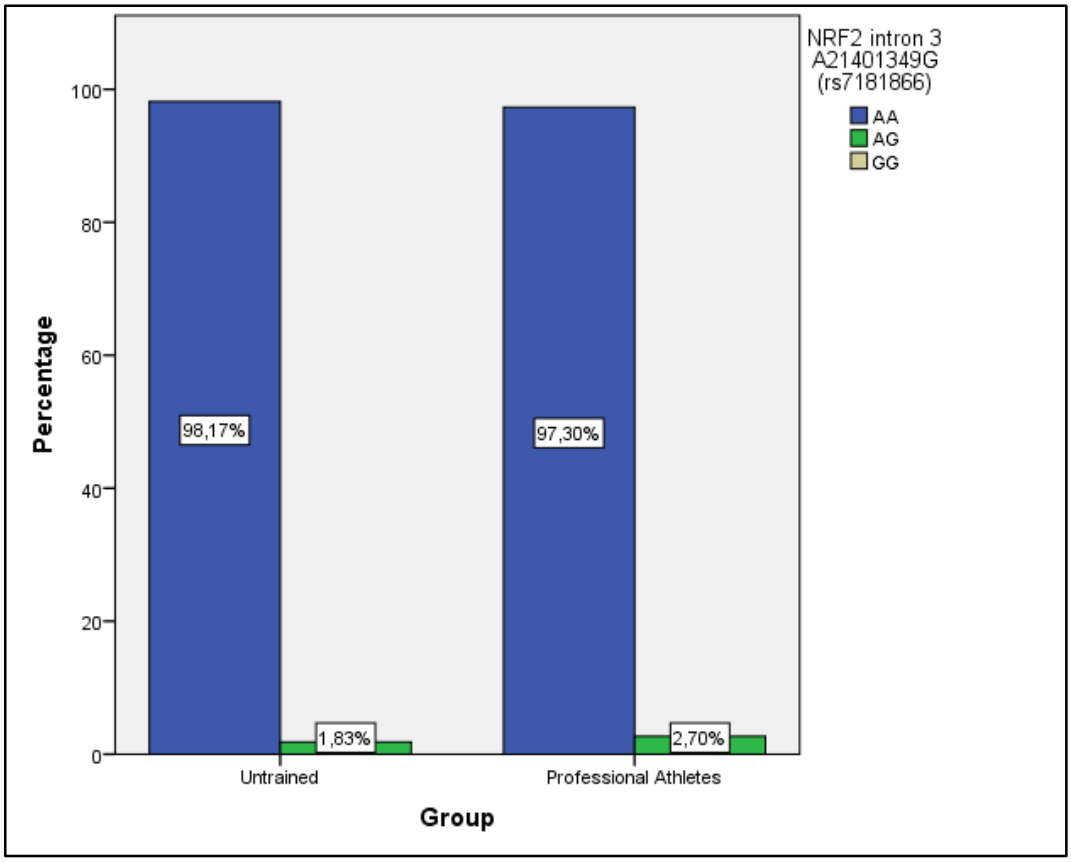

Figure 18: Percentage distribution of NRF2 intron 3 A/G (rs7181866) in athletes and controls

Prior to the Chi square test the following null hypothesis was set up.

H0: There is no significant relationship between the level of sports and the genetic variation of the NRF2 intron 3 A/G (rs1781866).

The result of the Chi square test shows a significance level of 0.694. This value is not significant, therefore the H0 cannot be dismissed.

Result: No significant relationship between the level of sports and the investigated genetic variation could be found.

5.4.2. NRF2 (rs7181866) in soccer and handball players

45 men were not tested for this genetic variation. Therefore only data of 74 subjects is available. Frequencies (expected counts and measured counts) as well as further information can be seen in Table 17. Of the three possible genetic expressions (AA, AG,

and GG) only two occur. Furthermore no soccer player was tested heterozygous. More handball players than expected were tested for this expression of the polymorphism.

Table 18: NRF2 intron 3 A/G (rs7181866) distribution in soccer and handball players

			NRF2 intron 3 A21401349G (rs7181866)		
			AA	AG	Total
Sport	Soccer	Count	36	0	36
		Expected Count	35,0	1,0	36,0
	Handball	Count	36	2	38
		Expected Count	37,0	1,0	38,0
Total		Count	72	2	74
		Expected Count	72,0	2,0	74,0

The percentage distribution (see Figure 19) shows that both groups are dominated by the AA expression of the polymorphism.

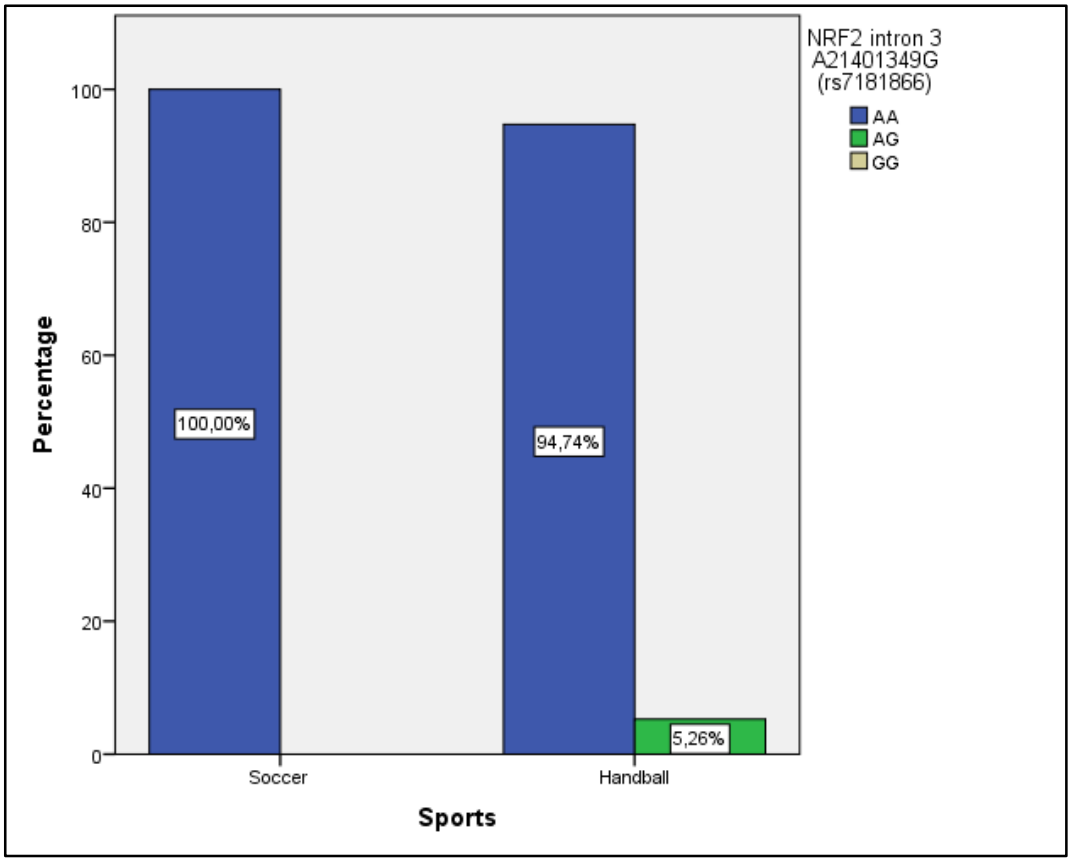

Figure 19: Percentage distribution of NRF2 intron 3 A/G (rs7181866) in soccer and handball players

Before the Chi square test was performed the null hypothesis had to be set up.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and genetic variation of the NRF2 intron 3 A/G (rs1781866).

The result of the Chi square test shows a significance level 0.163. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the type of sports and the genetic variation of the NRF2 (rs1781866).

5.5. PPARG coactivator 1α (rs8192678)

5.5.1. PPARGC1A (rs8192678) in athletes and controls

Six men of the control group were not tested for the polymorphism of the PPARGC1A (rs8192678) gene. Consequently N is for this gene 228. The control group is more often

homozygous to either the T allele or the C allele than expected. The TC variation occurs in the control group less than expected. Professional athletes show higher measured count than expected for TC expression of this polymorphism. Both other variations (TT and CC) occur less than expected.

Table 19: PPARGC1A Gly482Ser (rs8192678) distribution in athletes and controls

			PPARGC1A Gly482Ser (rs8192678)			
			TT	TC	CC	Total
Group	Untrained	Count	14	42	53	109
		Expected Count	12,9	44,5	51,6	109,0
	Professional Athletes	Count	13	51	55	119
		Expected Count	14,1	48,5	56,4	119,0
Total		Count	27	93	108	228
		Expected Count	27,0	93,0	108,0	228,0

The histogram (see Figure 20) shows similar distribution in both groups. The control group has more men homozygous for allele T (12.84%) than the professional athletes (10.92%). Athletes (42.86%) have a higher percentage of men tested heterozygous (TC) than controls (38.53%). More men of the control group (48.62%) than of the athlete group (46.22%) were tested homozygous for the C allele of this gene.

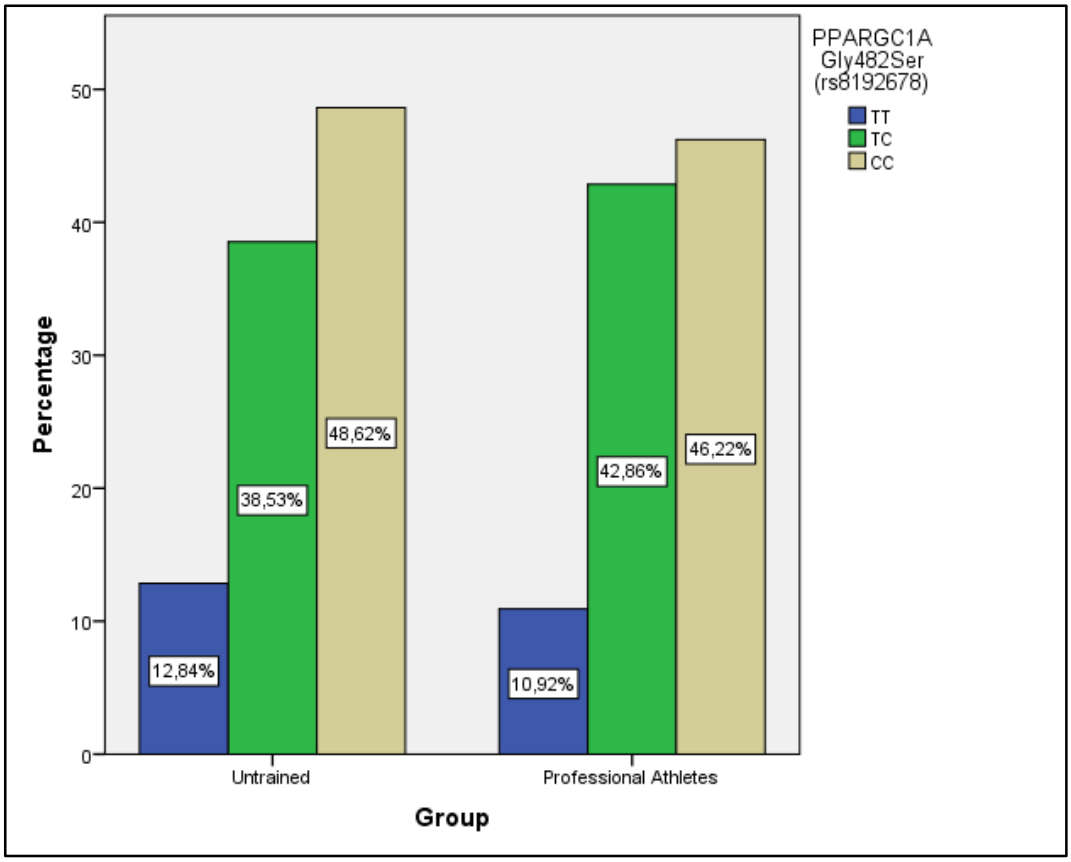

Figure 20: Percentage distribution of PPARGC1A Gly482Ser (rs8192678) in athletes and controls

Prior to the Chi square test the null hypothesis was built.

H0: There is no significant relationship between the level of sports and the genetic variation of the PPARGC1A Gly482Ser (rs8192678).

The result of the Chi square test shows a significance level of 0.776. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the level of sports and the investigated genetic variation.

5.5.2. PPARGC1A (rs8192678) in soccer and handball players

81 Soccer and Handball players were tested for the polymorphism of the PPARGC1A (rs8192678) gene (see Table 20). Soccer players have lower frequencies of the TT and the TC expression of this polymorphism but higher frequency of the CC expression of

this polymorphism than expected. Different results can be seen for handball players. Handball players have higher frequencies than expected of the TT and the TC variation but lower frequency of C homozygous.

Table 20: PPARGC1A Gly482Ser (rs8192678) distribution in soccer and handball players

			PPARGC1A Gly482Ser (rs8192678)			
			TT	TC	CC	Total
Sport	Soccer	Count	7	32	42	81
		Expected Count	8,8	34,7	37,4	81,0
	Handball	Count	6	19	13	38
		Expected Count	4,2	16,3	17,6	38,0
Total		Count	13	51	55	119
		Expected Count	13,0	51,0	55,0	119,0

Figure 21 shows variances in the distribution between soccer and handball players. More handball players (15.79%) than soccer players (8.64%) were tested positive for the TT expression of this polymorphism. 50.00% of handball players are heterozygous to the investigated polymorphism compared to 39.51% of soccer players. (42.86%) have a higher percentage of men tested heterozygous (TC) than controls (38.53%). More men of the soccer group (51.85%) than of the handball group (34.21%) were tested homozygous for the C allele of this gene.

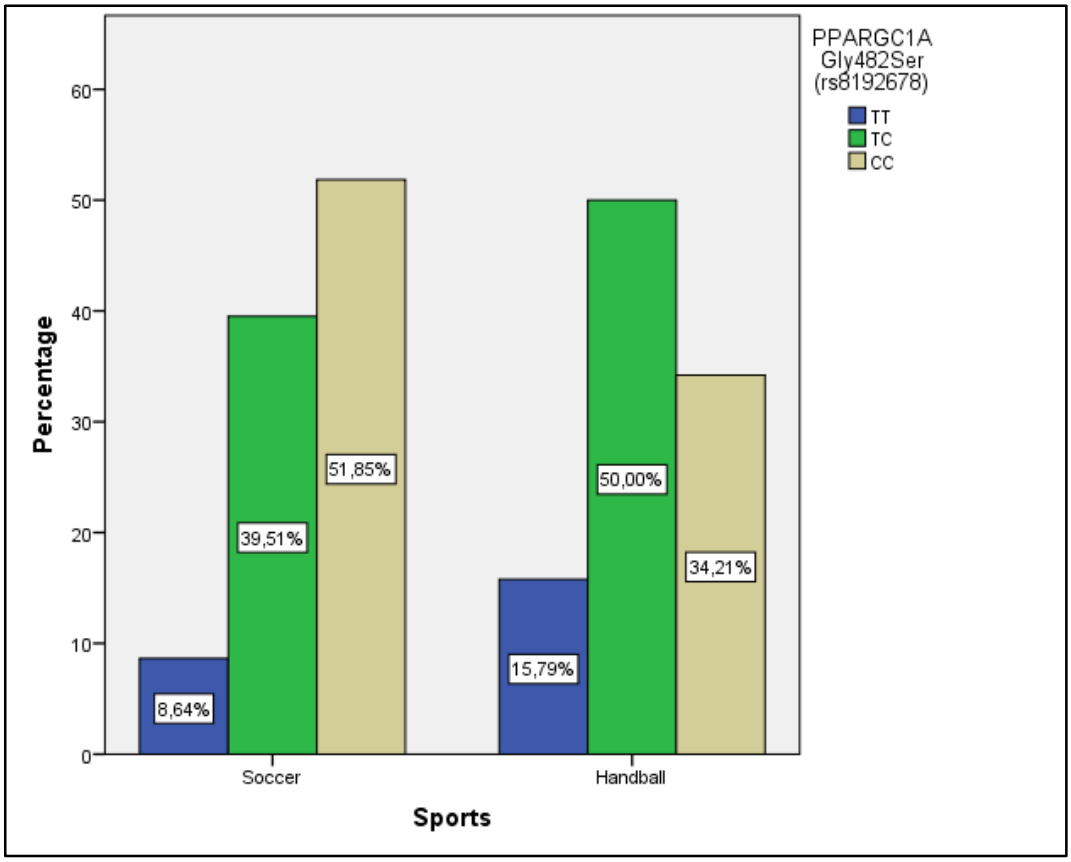

Figure 21: Percentage distribution of PPARGC1A Gly482Ser (rs8192678) in soccer and handball players

Before calculations were performed the following null hypothesis was set up.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the PPARGC1A Gly482Ser (rs8192678).

The result of the Chi square test shows a significance level of 0.164. This value is not significant, therefore the H0 cannot be dismissed.

Result: No significant relationship between the type of sports and the PPARGC1A (rs8192678) was found.

5.6. HIF1A (rs11549465)

5.6.1. HIF1A (rs11549465) in athletes and controls

Six men of the control group were not tested for the Pro582Ser polymorphism of the HIF1A (rs11549465) gene. No member of the control group but more professional athletes than expected were tested homozygous for the T allele. Athletes have higher frequency of the CC expression of this polymorphism and lower frequency of the CT variation than expected. The control group has less frequency of the CC but higher frequency of the CT expression than expected. Detailed results are presented in Table 21.

Table 21: HIF1A Pro582Ser (rs11549465) distribution in athletes and controls

			HIF1A Pro582Ser (rs11549465)			
			CC	CT	TT	Total
Group	Untrained	Count	92	17	0	109
		Expected Count	88,0	19,6	1,4	109,0
	Professional Athletes	Count	92	24	3	119
		Expected Count	96,0	21,4	1,6	119,0
Total		Count	184	41	3	228
		Expected Count	184,0	41,0	3,0	228,0

The histogram (see Figure 22) shows that 84.40% of the control group are homozygous to C. 15.60% of the same group are heterozygous but 0.00% are homozygous to T. 77.31% of the professional athletes are homozygous to C and 2.52% are homozygous to T. 20.17% of the athletes are heterozygous.

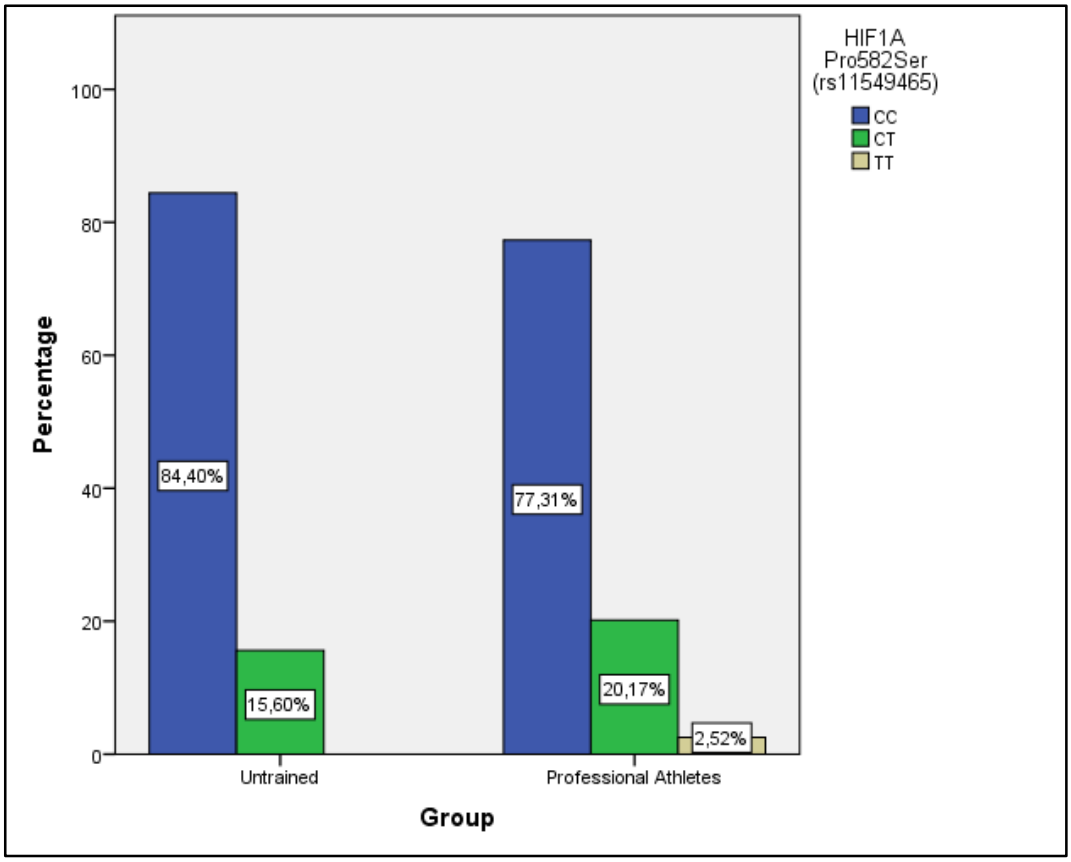

Figure 22: Percentage distribution of HIF1A Pro582Ser (rs11549465) in athletes and controls

Prior to the statistical analysis the null hypothesis for the Chi square test had to be set up.

H0: There is no significant relationship between the level of sports and the genetic variation of the HIF1A Pro582Ser (rs11549465).

The result of the Chi square test shows a significance level of 0.152. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the level of sports and the investigated polymorphism.

5.6.2. HIF1A (rs11549465) in Soccer and Handball players

In the crosstable (see Table 22) little differences between expected count and measured count can be seen. In soccer players higher frequency of CT could be measured than

expected. CC occurs less often in soccer players than expected. In handball players the CC expression of the gene was measured more often than expected. Lower frequency of the CT expression was measured than estimated.

Table 22: HIF1A Pro582Ser (rs11549465) distribution in soccer and handball players

			HIF1A Pro582Ser (rs11549465)			
			CC	CT	TT	Total
Sport	Soccer	Count	61	18	2	81
		Expected Count	62,6	16,3	2,0	81,0
	Handball	Count	31	6	1	38
		Expected Count	29,4	7,7	1,0	38,0
Total		Count	92	24	3	119
		Expected Count	92,0	24,0	3,0	119,0

The histogram (see Figure 23) shows similar distribution in soccer and handball players. 75.31% of handball players and 81.58% of soccer players were tested homozygous for allele C. 22.22% of soccer players and 15.79% of handball players were tested positive for the CT expression of this polymorphism. The minority of soccer players (2.47%) and handball players (2.63%) was tested homozygous for the T allele.

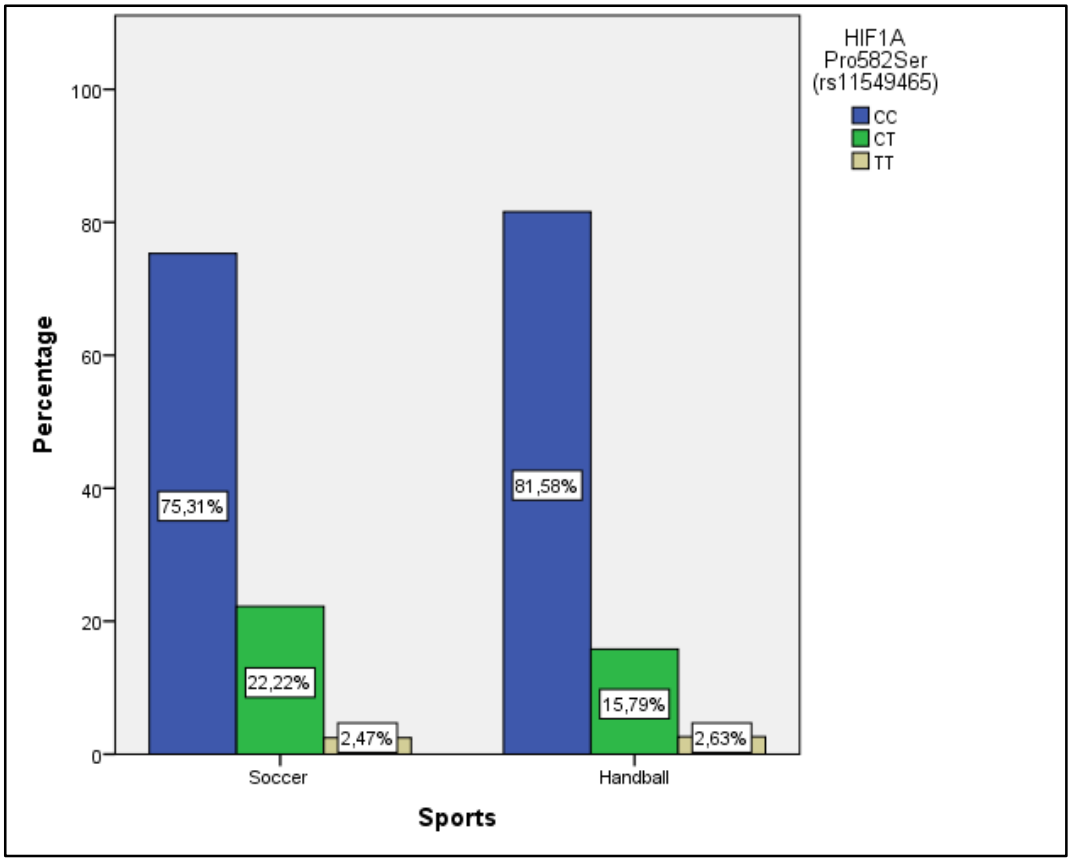

Figure 23: Percentage distribution of HIF1A Pro582Ser (rs11549465) in soccer and handball players

Before Chi square test was performed the null hypothesis was built.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the HIF1A Pro582Ser (rs11549465).

The result of the Chi square test shows a significance level of 0.717. This value is not significant, therefore the H0 cannot be dismissed.

Result: No significant relationship between the type of sports and the genetic variations could be found.

5.7. ACTN3 (rs1815739)

5.7.1. ACTN3 (rs1815739) in athletes and controls

Table 23 shows the frequency of the R577X polymorphism in the ACTN3 (rs1815739) gene. Controls show higher frequencies of the XX expression than expected. Little differences between expected count and measured count for RX and RR in controls can be seen. In professional athletes there are higher frequencies of the RR and RX expression of the polymorphism than expected. In athletes the XX polymorphism occurs less often than expected.

Table 23: ACTN3 R577X (rs1815739) distribution in athletes and controls

			ACTN3 R577X (rs1815739)			
			RR	RX	XX	Total
Group	Untrained	Count	37	51	27	115
		Expected Count	40,3	52,1	22,6	115,0
	Professional Athletes	Count	45	55	19	119
		Expected Count	41,7	53,9	23,4	119,0
Total		Count	82	106	46	234
		Expected Count	82,0	106,0	46,0	234,0

The percentage distribution (see Figure 24) shows differences between professional athletes and controls. More athletes (37.82%) were tested homozygous for the R allele than controls (32.17%). Similar distribution for the RX expression of the ACTN3 gene could be found in athletes (46.22%) and controls (44.35%).

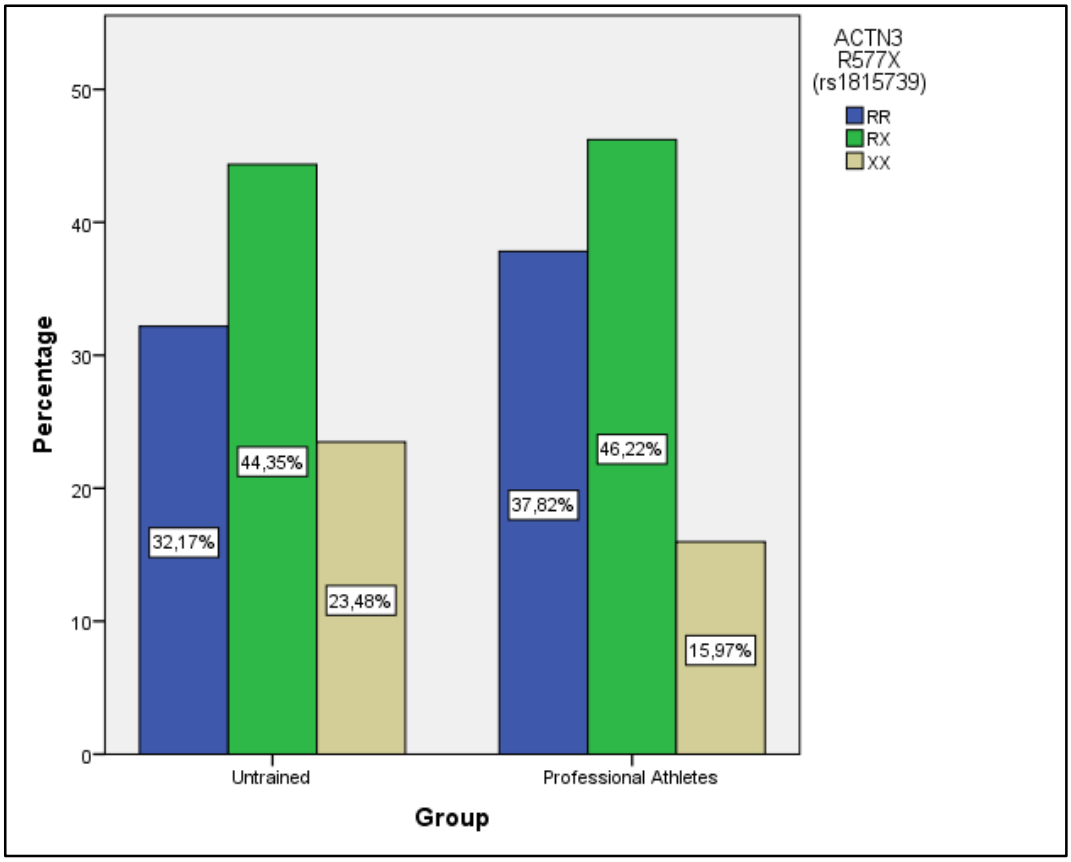

Figure 24: Percentage distribution of ACTN3 R577X (rs1815739) in athletes and controls

H0: There is no significant relationship between the level of sports and the genetic variation of the ACTN3 R577X (rs1815739).

The result of the Chi square test shows a significance level of 0.324. This value is not significant, therefore the H0 cannot be dismissed.

5.7.2. ACTN3 (rs1815739) in soccer and handball players

119 people were tested for the R577X polymorphism of the ACTN3 gene. Little differences can be seen in both investigated groups. In soccer players the frequency of RR is higher than expected and the frequency of XX is lower than expected. Little differences in handball players can be seen (see Table 24)

Table 24: ACTN3 R577X (rs1815739) distribution in soccer and handball players

			ACTN3 R577X (rs1815739)			
			RR	RX	XX	Total
Sport	Soccer	Count	32	37	12	81
		Expected Count	30,6	37,4	12,9	81,0
	Handball	Count	13	18	7	38
		Expected Count	14,4	17,6	6,1	38,0
Total		Count	45	55	19	119
		Expected Count	45,0	55,0	19,0	119,0

45.68% of soccer players and 47.37% of handball players were tested heterozygous for the investigated polymorphism. More handball players (18.42%) were tested homozygous for the X allele than soccer players (14.81%). Percentage distribution shows that more soccer players (39.51%) are homozygous to allele R than handball players (34.21%). All information is graphically illustrated in Figure 25.

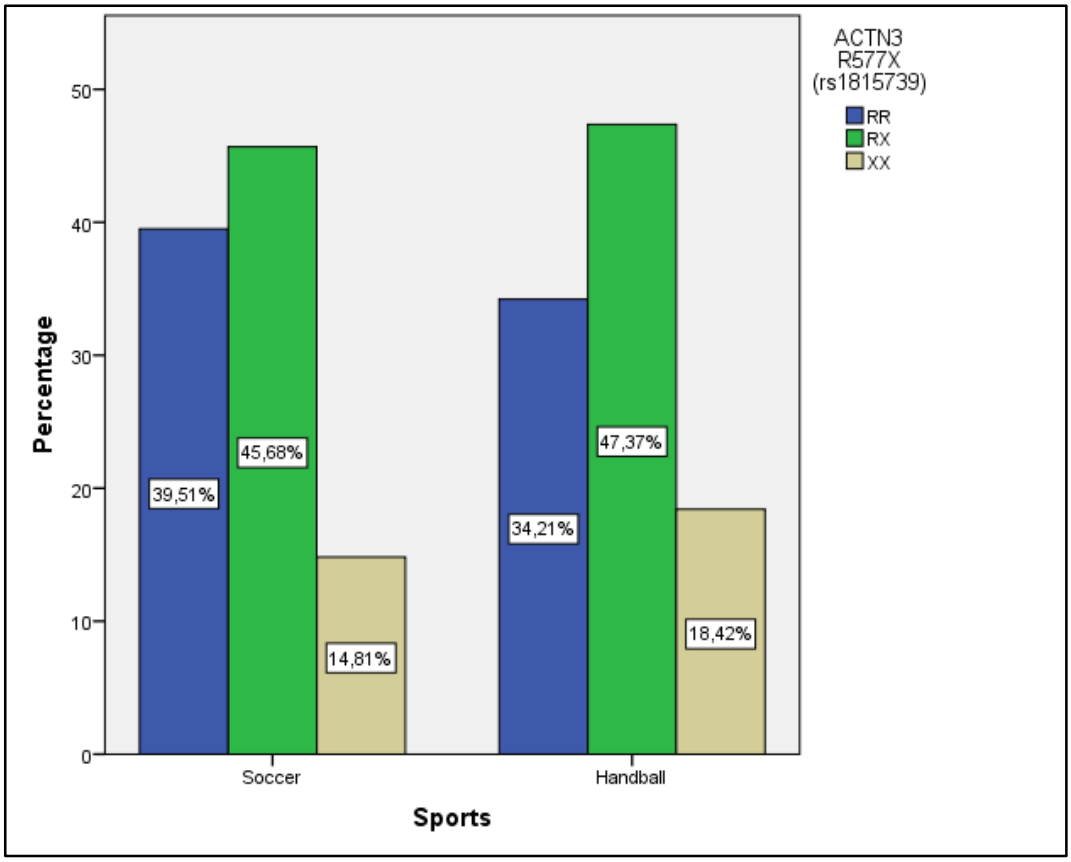

Figure 25: Percentage distribution of ACTN3 R577X (rs1815739) in soccer and handball players

The null hypothesis for the relationship between ACTN3 (Rs1815739) was set prior to the statistical analysis.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the ACTN3 R577X (rs1815739).

The result of the Chi square test shows a significance level of 0.811. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the type of sports and the investigated genetic variation.

5.8. Myostatin (rs1805086)

5.8.1. Myostatin (rs1805086) in athletes and controls

234 subjects were tested for the K153R polymorphism of the MSTN (rs1805086) gene. No differences between expected count and measured count can be observed in the control group. Also in professional athletes only little differences between expected count and measured count can be seen. Nevertheless it is notable that no professional athlete was tested positive for the RR expression of the K153R polymorphism.

Table 25: MSTN K153R (rs1805086) distribution in athletes and controls

			MSTN K153R (rs1805086)			
			KK	KR	RR	Total
Group	Untrained	Count	112	2	1	115
		Expected Count	112,1	2,5	,5	115,0
	Professional Athletes	Count	116	3	0	119
		Expected Count	115,9	2,5	,5	119,0
Total		Count	228	5	1	234
		Expected Count	228,0	5,0	1,0	234,0

Percentage distribution (see Figure 26) shows a strong tendency towards the KK expression of the MSTN K153R gene in professional athletes (97.48%) and in the control group (97.35%). 1.74% of the control group and 2.52% of the professional athletes are heterozygous. 0.87% of controls are homozygous to allele R.

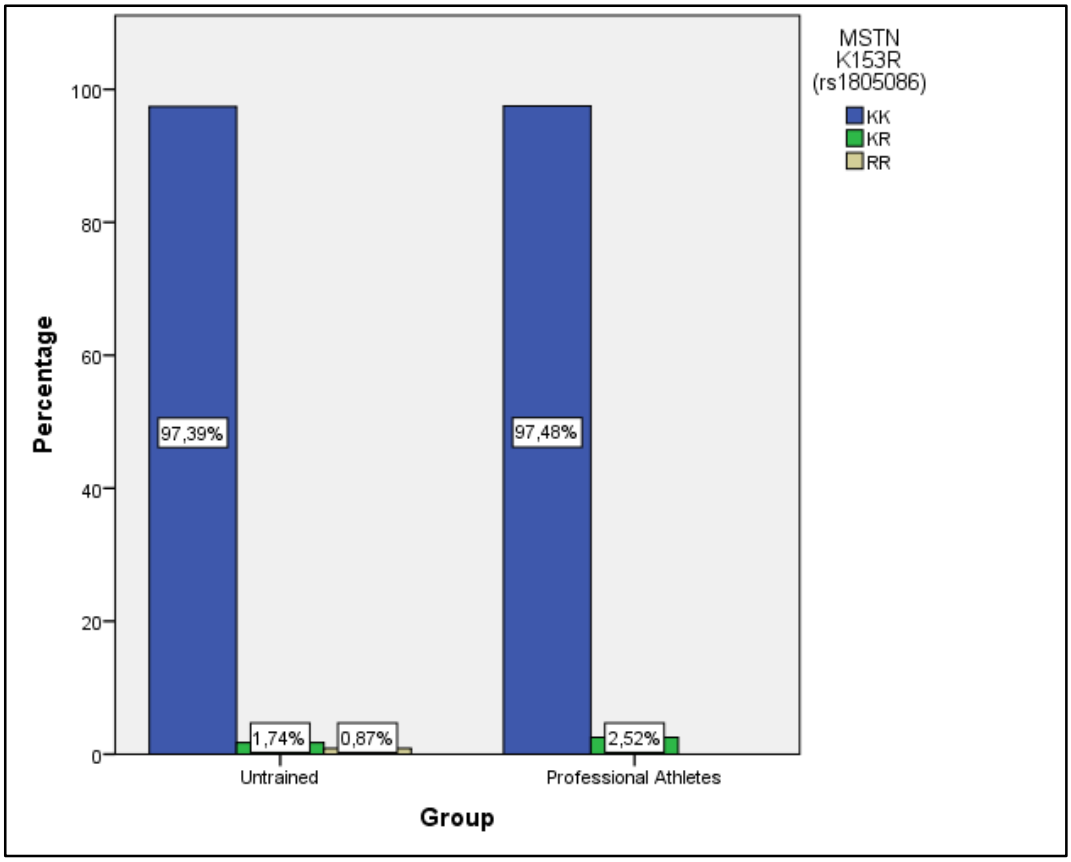

Figure 26: Percentage distribution of MSTN K153R (rs1805086) in athletes and controls

H0: There is no significant relationship between the level of sports and the genetic variation of the MSTN K153R (rs1805086).

The result of the Chi square test shows a significance level of 0.548. This value is not significant, therefore the H0 cannot be dismissed.

5.8.2. Myostatin (rs1805086) in soccer and handball players

81 soccer players and 38 handball players were tested for the K153R polymorphism of the MSTN (rs1805086) gene. Little differences between expected count and measured count can be observed in both groups.

Table 26: MSTN K153R (rs1805086) distribution in soccer and handball players

			MSTN K153R (rs1805086)		
			KK	KR	Total
Sport	Soccer	Count	78	3	81
		Expected Count	79,0	2,0	81,0
	Handball	Count	38	0	38
		Expected Count	37,0	1,0	38,0
Total		Count	116	3	119
		Expected Count	116,0	3,0	119,0

The histogram (see Figure 27) shows that 100% of all handball players are homozygous to allele K. 96.30% of soccer players are homozygous to allele K and 3.70% are heterozygous.

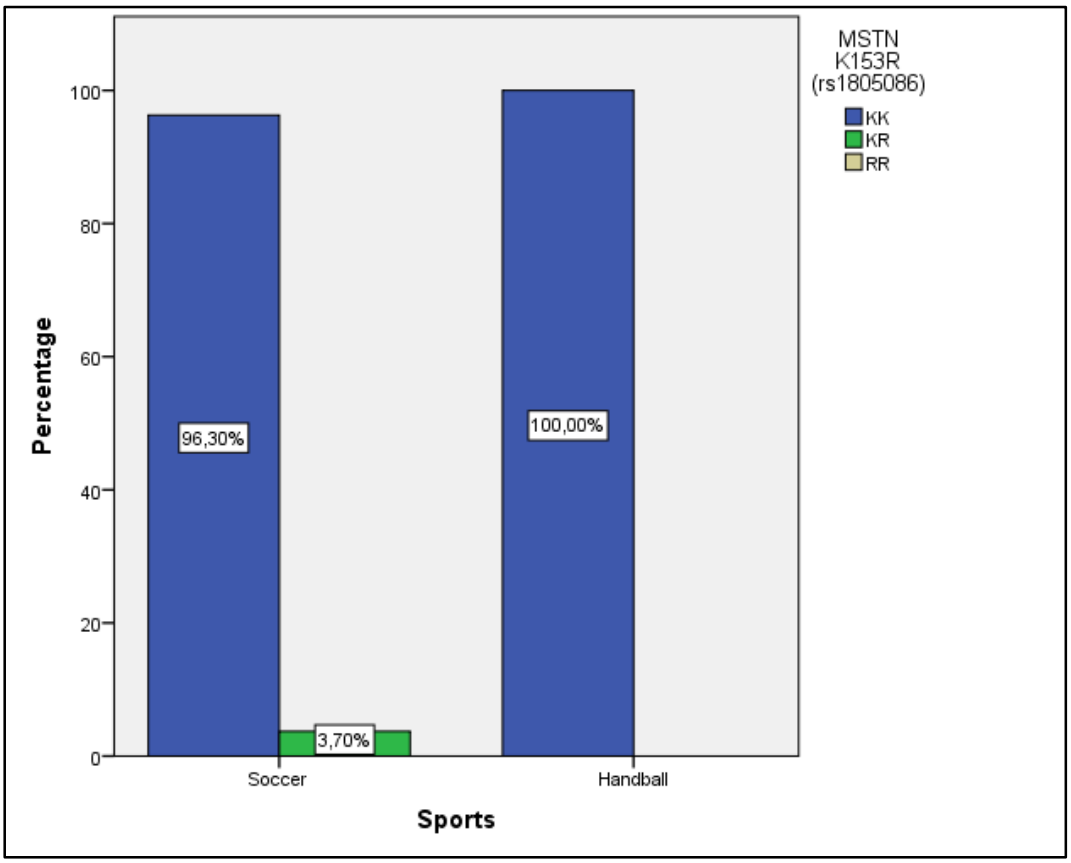

Figure 27: Percentage distribution of MSTN K153R (rs1805086) in soccer and handball players

Prior to statistical analysis the following null hypothesis was set up.

H0: There is no significant relationship between the level of sports and the genetic variation of the MSTN K153R (rs1805086).

The result of the Chi square test shows a significance level of 0.230. This value is not significant, therefore the H0 cannot be dismissed.

Result: No significant relationship between MSTN K153R (rs1805086) could be found.

5.9. IGF-1 (rs35767)

5.9.1. IGF-1 (rs35767) in athletes and controls

All subjects were tested for the -1246C/T polymorphism of the IGF1 (rs35767) gene. The control group shows higher frequencies in the AG as well as in the GG variation of this polymorphism than expected. None of the controls was tested homozygous for the A allele. This value is less than expected. More professional athletes than expected were tested homozygous for the A allele. The AG and the GG variation in athletes were measured less than expected. For further information see Table 27.

Table 27: IGF-1 -1246C/T (rs35767) distribution in athletes and controls

			IGF1 -1246C/T (rs35767)			
			AA	AG	GG	Gesamt
Group	Untrained	Count	0	27	88	115
		Expected Count	2,9	26,0	86,0	115,0
	Professional Athletes	Count	6	26	87	119
		Expected Count	3,1	27,0	89,0	119,0
Total		Count	6	53	175	234
		Expected Count	6,0	53,0	175,0	234,0

The percentage distribution of the IGF1 (rs35767) shows variances between the control group and the group of professional athletes (see Figure 28). 5.04% of athletes were tested homozygous for the A allele compared to 0.00% of the control group. Controls (23.48% and 76.52%) show higher frequencies in the AG and the GG variation of the polymorphism compared to athletes (21.85% and 73.11%).

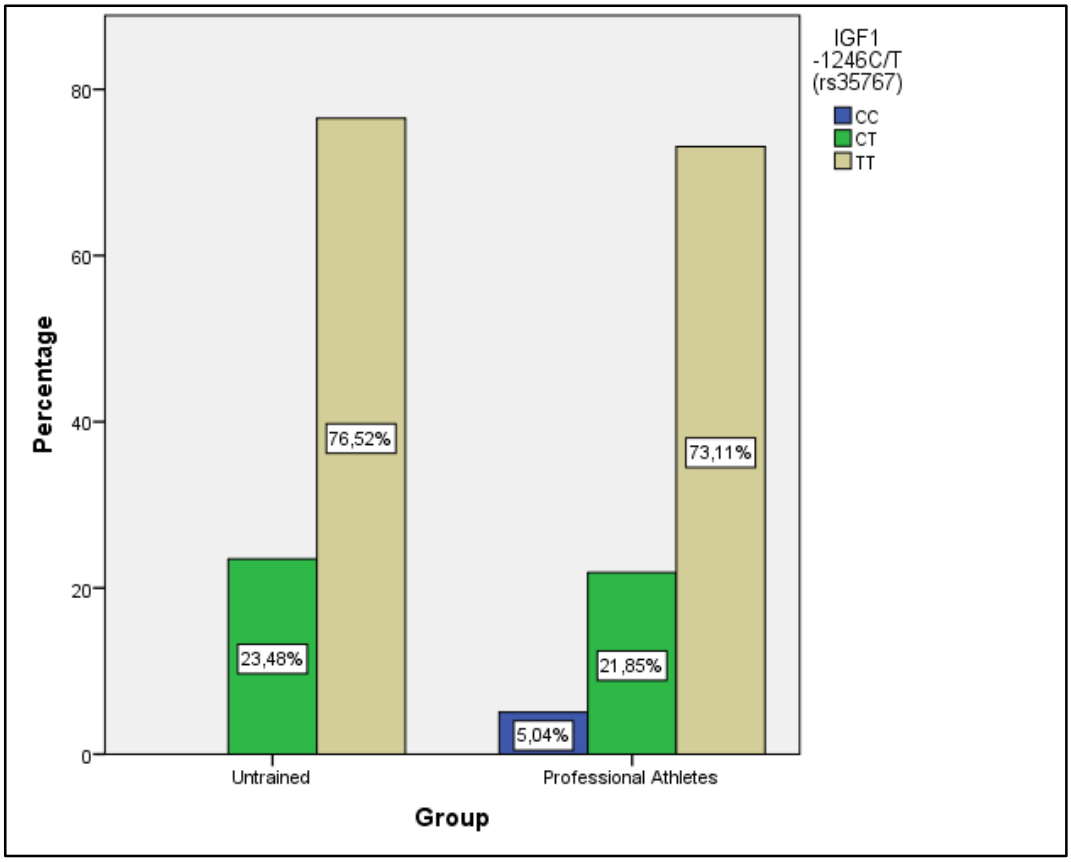

Figure 28: Percentage Distribution of IGF1 -1246C/T (rs35767) in athletes and controls

Prior to the Chi square test the following null hypothesis was set up.

H0: There is no significant relationship between the level of sports and the genetic variation of the IGF1 -1246C/T (rs35767).

The result of the Chi square test shows a significance level of 0.051. This value is not significant, therefore the H0 cannot be dismissed.

Result: No significant relationship between the level of sports and the genetic variation of the IGF1 (rs35767) could be found.

5.9.2. IGF1 (rs 35767) in soccer and handball players

All subjects were tested for the -1246C/T polymorphism of the IGF1 (rs35767) gene. The group of soccer players shows higher frequencies in the AA as well as in the AG variation of this polymorphism than expected. More handball players than expected were tested

homozygous for the G allele. The AG and the GG variation in handball players were measured less than expected. For further information see Table 28.

Table 28: IGF1 -1246C/T (rs35767) distribution in soccer and handball players

			IGF1 -1246C/T (rs35767)			
			AA	AG	GG	Total
Sport	Soccer	Count	5	20	56	81
		Expected Count	4,1	17,7	59,2	81,0
	Handball	Count	1	6	31	38
		Expected Count	1,9	8,3	27,8	38,0
Total		Count	6	26	87	119
		Expected Count	6,0	26,0	87,0	119,0

The percentage distribution of the IGF1 (rs35767) shows differences between soccer and handball players (see Figure 29). 81.58% of handball players compared to 69.14% of soccer players were homozygous for the G allele. The frequency of the AA expression and of the AG expression was in soccer players (6.17% and 24.69%) higher than in handball players (2.63% and 15.79%).

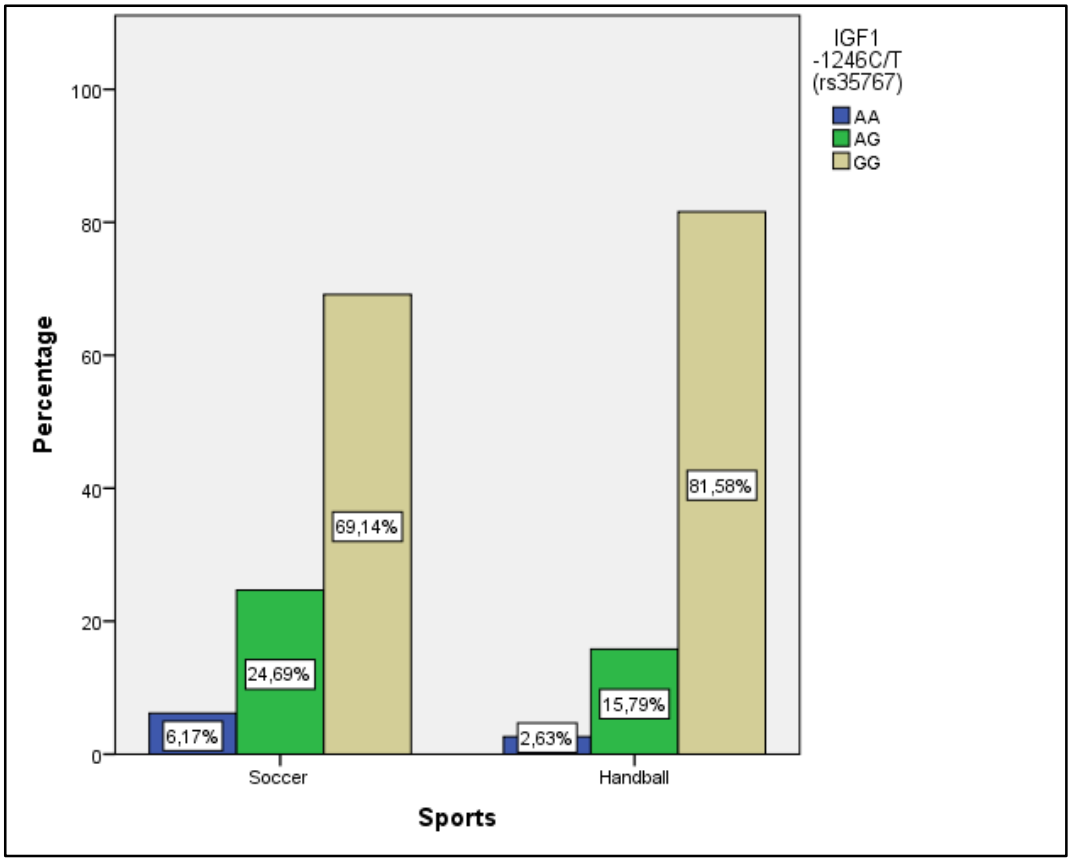

Figure 29: Percentage Distribution of IGF1 -1246C/T (rs35767) in soccer and handball players

For the Chi square test the following null hypothesis was set up.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the IGF1 -1246C/T (rs35767)).

The result of the Chi square test shows a significance level of 0.345. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the type of sports and the investigated genetic variation.

5.10. IGF-2 (rs3213221)

5.10.1. IGF-2 (rs3213221) in athletes and controls

One professional athlete was not tested for the C13790G polymorphism in the IGF2 (rs3213221) gene. Therefore n=233. The control group has a lower frequency of CG and higher frequencies of CC and GG than expected. Professional athletes have a higher frequency of CG and lower frequencies of CC and GG than expected.

Table 29: IGF2 C13790G (rs3213221) distribution in athletes and controls

			IGF2 C13790G (rs3213221)			
			CC	CG	GG	Total
Group	Untrained	Count	16	51	48	115
		Expected Count	15,8	52,8	46,4	115,0
	Professional Athletes	Count	16	56	46	118
		Expected Count	16,2	54,2	47,6	118,0
Total		Count	32	107	94	233
		Expected Count	32,0	107,0	94,0	233,0

The percentage distribution of IGF2 C13790G (rs3213221) shows variances between the control group and the professional athletes (see Figure 30). Professional athletes (47.46%) have higher frequency of the CG genotype than controls (44.35%). Controls (13.91% and 41.74%) show higher percentage of the CC and the GG variation than professional athletes (13.56% and 13.91%).

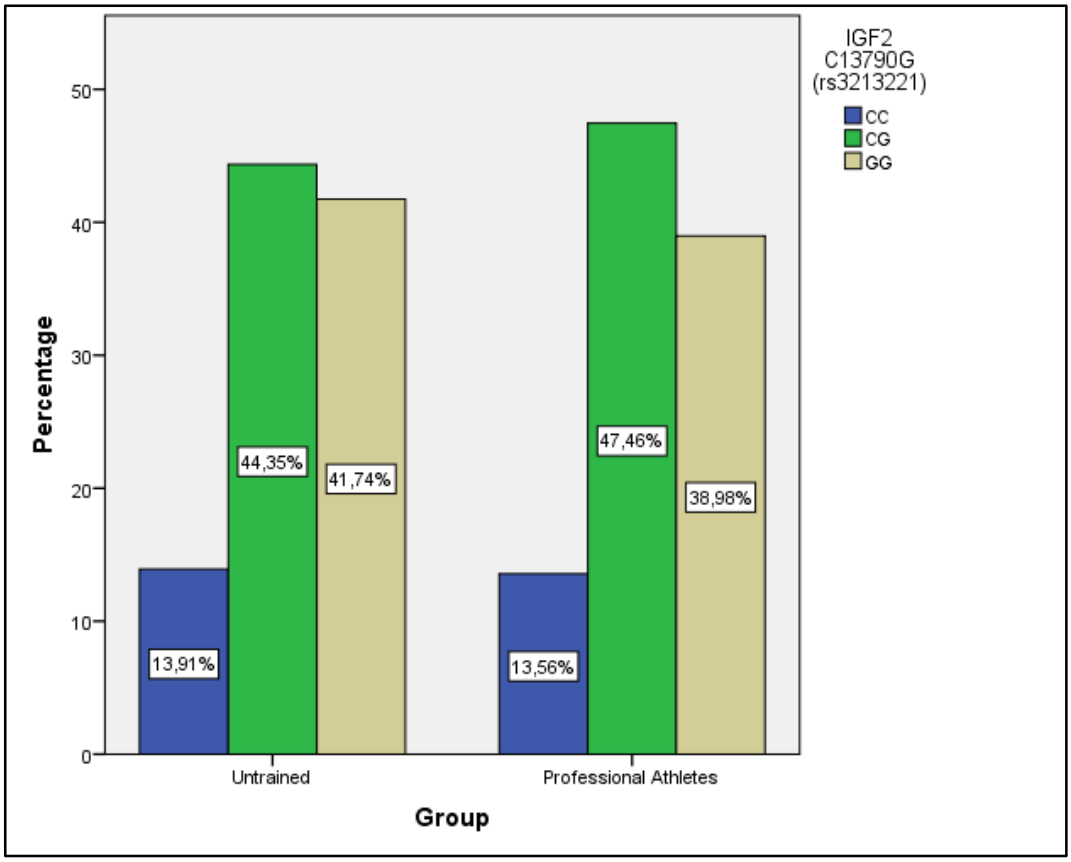

Figure 30: Percentage Distribution of IGF2 C13790G (rs3213221) in athletes and controls

For the Chi square test the following null hypothesis was set up.

H0: There is no significant relationship between the level of sports and the genetic variation of the IGF2 C13790G (rs3213221).

The result of the Chi square test shows a significance level of 0,888. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the level of sports and the investigated genetic variation.

5.10.2. IGF-2 (rs3213221) in soccer and handball players

One soccer player was not tested for the C13790G polymorphism in the IGF2 (rs3213221) gene. Soccer players have lower frequencies of CC and CG and a higher frequency of GG than expected. Handball players have a lower frequency of GG and

higher frequencies of CC and CG than expected. Detailed results are presented in Table 30.

Table 30: IGF2 C13790G (rs3213221) distribution in soccer and handball players

			IGF2 C13790G (rs3213221)			
			CC	CG	GG	Total
Sport	Soccer	Count	9	36	35	80
		Expected Count	10,8	38,0	31,2	80,0
	Handball	Count	7	20	11	38
		Expected Count	5,2	18,0	14,8	38,0
Total		Count	16	56	46	118
		Expected Count	16,0	56,0	46,0	118,0

The percentage distribution for IGF2 (rs3213221) show variances in the frequency of the expression of the polymorphism (see Figure 31). 52.63% of handball players were tested heterozygous compared to 45.00% of the soccer players. Handball players (18.42%) show also higher frequencies of the CC variation than soccer players (11.25%). 43.75% of soccer players were homozygous for the G allele compared to 28.95% of the handball players.

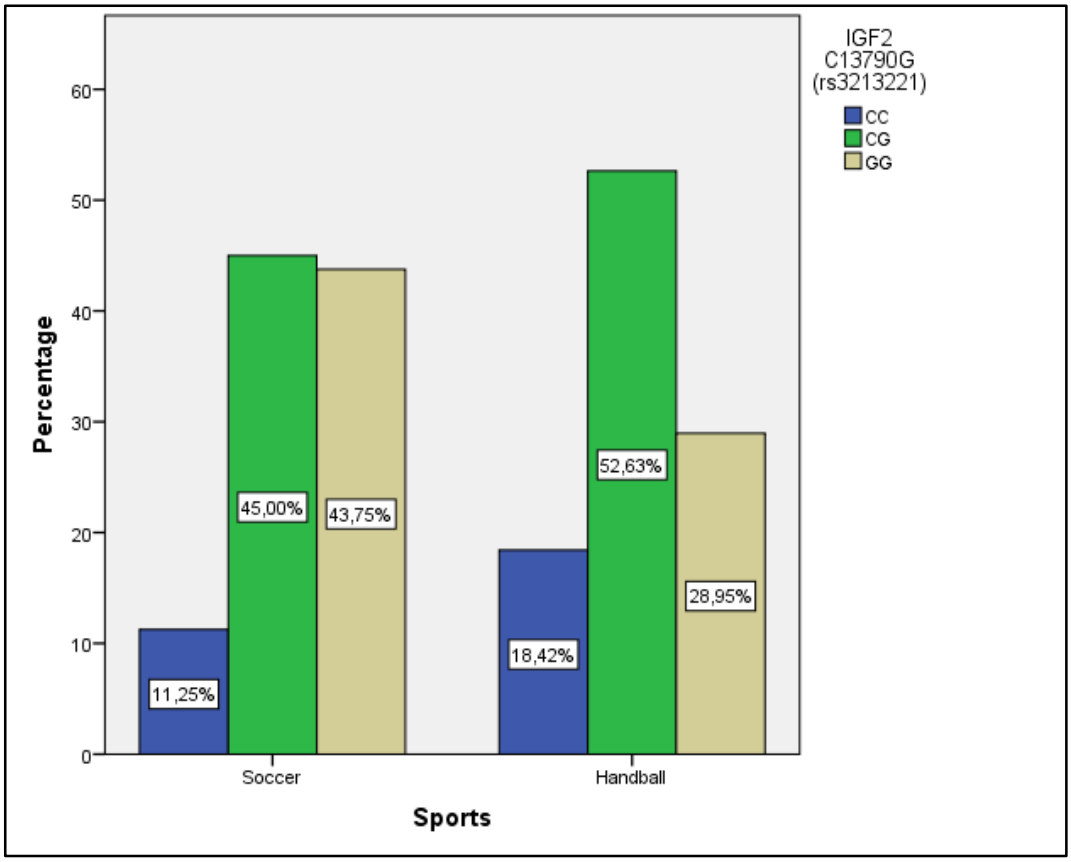

Figure 31: Percentage distribution of IGF2 C13790G (rs3213221) in soccer and handball players

Chi square test was performed. Therefore the following null hypothesis was set up.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the IGF2 C13790G (rs3213221).

The result of the Chi square test shows a significance level of 0.254. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant relationship between the type of sports and the genetic variation of the IGF2 (rs3213221).

5.11. IGF-2 (rs7924316)

5.11.1. IGF-2 (rs7924316) in athletes and controls

The G11711T polymorphism of the IGF2 (rs7924316) gene was tested in all subjects (n=234). Little differences between expected count and measured count can be seen in Table 31. The controls have higher frequencies of GG and TT and lower frequencies of GT than expected. Professional athletes have lower frequencies of GG and TT and higher frequencies of GT than expected.

Table 31: IGF2 G11711T (rs7924316) distribution in athletes and controls

			IGF2 G11711T (rs7924316)			
			GG	GT	TT	Total
Group	Untrained	Count	35	56	24	115
		Expected Count	35,9	54,1	25,1	115,0
	Professional Athletes	Count	38	54	27	119
		Expected Count	37,1	55,9	25,9	119,0
Total		Count	73	110	51	234
		Expected Count	73,0	110,0	51,0	234,0

The percentage distribution of IGF2 G11711T shows variances between the control group and the professional athletes (see Figure 33). Professional athletes have higher frequency of the GG genotype and the TT genotype than controls. 31.93% of the athletes were tested homozygous for G allele compared to 30.43% of the controls. 22.69% of the athletes were tested homozygous for the T allele compared to 20.87% of the controls. Controls (48.70%) show higher percentage of the GT polymorphism than professional athletes (45.38%).

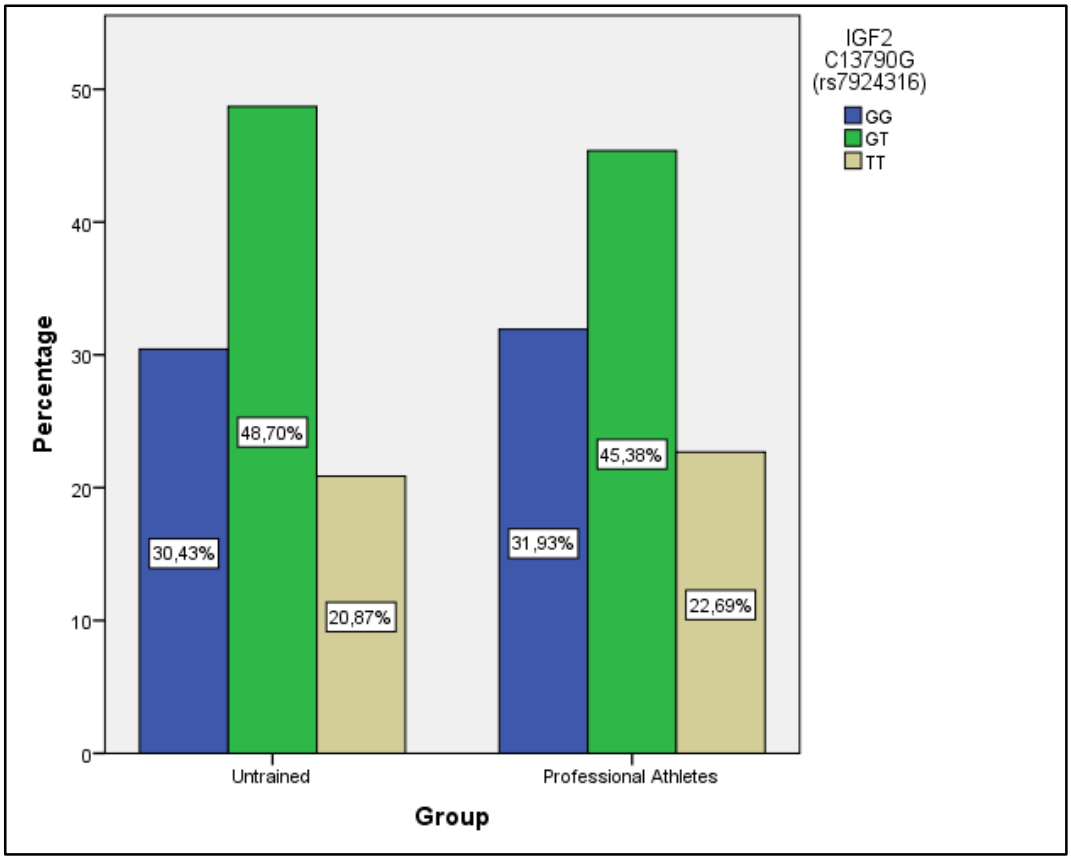

Figure 32: Percentage distribution of IGF2 G11711T (rs7924316) in athletes and controls

Prior to the execution of the Chi square test a null hypothesis had to be set up.

H0: There is no significant relationship between the level of sports and the genetic variation of the IGF2 G11711T (rs7924316).

The result of the Chi square test shows a significance level of 0.875. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is not significant relationship between the level of sports and the genetic variation of the IGF2 (rs7924316).

5.11.2. IGF-2 (rs7924316) in soccer and handball players

The G11711T polymorphism of the IGF2 (rs7924316) gene was tested in all subjects (n=119). Differences between expected count and measured count can be seen in Table 32. Soccer players have lower frequencies of GT and TT and higher frequencies of GG

than expected. Handball players have lower frequencies of GG and higher frequencies of GT and TT than expected.

Table 32: IGF2 G11711T (rs7924316) distribution in soccer and handball players

			IGF2 G11711T (rs7924316)			
			GG	GT	TT	Total
Sport	Soccer	Count	31	33	17	81
		Expected Count	25,9	36,8	18,4	81,0
	Handball	Count	7	21	10	38
		Expected Count	12,1	17,2	8,6	38,0
Total		Count	38	54	27	119
		Expected Count	38,0	54,0	27,0	119,0

The percentage distribution for IGF2 (rs7924316) show variances in the frequency of the polymorphism. 55.26% of handball players were tested heterozygous compared to 40.74% of the soccer players. Handball players (26.32%) show also higher frequencies of the TT variation than soccer players (20.99%). 38.27% of soccer players were homozygous for the G allele compared to 18.42% of the handball players.

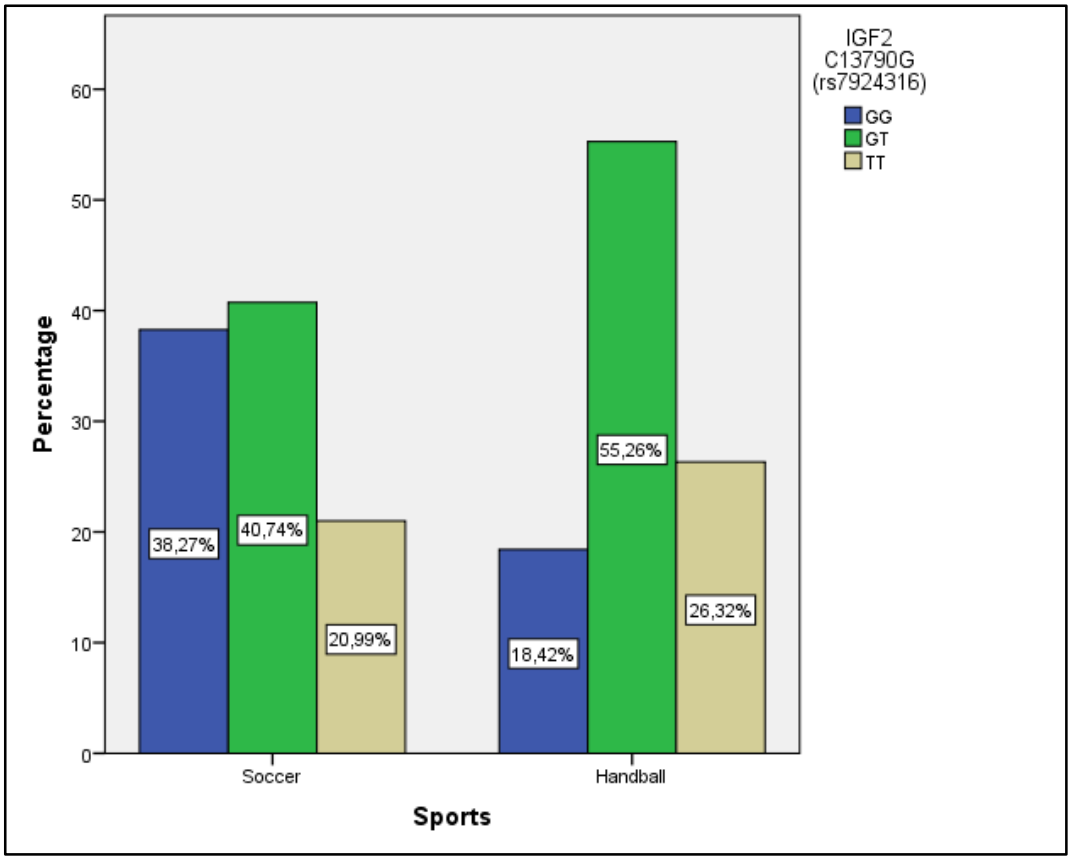

Figure 33: Percentage distribution of IGF2 C13790G (rs7924316) in soccer and handball players

To find out if there is any relationship between the type of sports and the genetic variation the following null hypothesis was set up before the Chi-square test was calculated.

H0: There is no significant relationship between the type of sports (soccer or handball) in professional athletes and the genetic variation of the IGF2 G11711T (rs7924316).

The result of the Chi square test shows a significance level of 0.095. This value is not significant, therefore the H0 cannot be dismissed.

Result: There is no significant difference between the type of sport and the genetic variation of the IGF2 (rs7924316) in professional athletes.

6. Discussion

In the last part of this master thesis all in chapter 5 described and presented results will be interpreted and discussed. For the discussion all findings are compared to the outcome of several other studies dealing with the same topic. The first part of this chapter will sum up the results of statistical analyses and the last part will verify if those results are supported by the conclusion of other authors.

The goal of this thesis was to investigate if there are any relationships between genetic variations of professional athletes and a control group as well as between soccer players and handball players. DNA samples were collected and for 11 different polymorphisms tested. In the next step Chi-square-tests were calculated. Neither for endurance related polymorphisms nor for strength related polymorphisms any relationship between the level of sports and the genetic variations could be found. All tests showed non-significant results. Also the Chi-square test for genetic variations in relationship to the type of sport did not show any significant results

Based on those results it can be assumed that genetic variations neither have an influence on the level of sports nor an influence on the type of sports in Austrian athletes. Hence it seems to be impossible to develop a polygenetic profile for Austrian athletes with the current data.

On the following pages every investigated polymorphism and the findings will be compared to relevant literature. Regrettably up to now (October 21st, 2015) no studies investigating any of the polymorphisms described in chapter 2 and their relationship to soccer or handball have been published. Therefore the results will be compared to studies dealing with one relevant rs-number and “sport” (PubMed search term: “rs# AND sport”).

The first investigated polymorphism was the ADRB1 Ser49Gly (rs1801252). As already mentioned it did not show any significant results neither in groups (p=0.395) nor in type of sports (p=0.087). Up to now only one study investigating the relationship between rs1801252 and sports has been published. Druk et al. (2015, p. 75) conclude that the ADRB1 Ser49Gly polymorphism, together with other genetic variations, increases the risk of adverse cardiovascular manifestations. Nevertheless, it is doubtable if those findings are controversy to the results shown in chapter 5 because their study included Russian children and it is questionable how those results can be compared to an Austrian cohort in adults.

Neither soccer players nor handball players and neither athletes nor controls showed any preference for the ADRB2 Arg16Gly (rs1042713) polymorphism. Pearson square showed no significant results in groups (p=0.623) and type of sports (p=0.767). Those findings are supported by Santiago et al. (2011, p. 147) who did not find any relationship between this polymorphism and endurance athletes, elite power athletes, and non-athletic controls. Tsianos et al. (2010, p. 567) conclude that the ADRB2 rs1042713 has some influence on endurance performance among habitual runners when statistics were limited to men whose favorite sports is running. Without this criteria no significant results could be found. Nevertheless they admit that further studies have to be done.

Also for the ADRB2 Gln27Glu (rs1042714) polymorphism no significant results could be found. Chi-square for athletes and controls showed a power of p=0.262 and a power of 0.999 for soccer players and handball players. As mentioned in the paragraph above those results are supported by Santiago et al. (2011, p. 147) who did not find any relationship between the ADRB2 Gln27Glu polymorphism and endurance athletes, elite power athletes, and non-athletic controls.

The NRF2 intron 3 A/G (rs7181866) has been associated with endurance performance (Maciejewska-Karłowska et al., 2012; Eynon et al., 2013; Eynon et al., 2009; He et al., 2007). However those findings were not found in groups (p=0.694) and type of sports (p=0.163). However it has to be mentioned that neither soccer nor handball can be classified as endurance specific. These results are controversial to the findings of 2009 Enyon et al. (2009, p. 76) who found a significant relationship between the AG genotype and endurance athlete status. This outcome is supported by Maciejewska-Karłowska et al. (2012, p. 115) who conclude that the NRF2 intron 3 A/G can be taken into consideration as a genetic variation which influences endurance performance. Especially the A/G genotype (p=0.012) and the G allele (p=0.014) can be associated with endurance performance. None the less they ask for further studies to support their findings. Those results are not supported by Enyon et al. (2013, p. 137) who did not find any significant relationship between the NRF2 intron 3 A/G polymorphism and sports performance in groups of endurance athletes, power-orientated athletes, and non-athletic controls. Additionally He et al. (2007, p. 717) conclude that some variances of the NRF2 gene potentially influence endurance capacity.

Pearson's Chi-square test for the PPARG1A Gly482Ser rs8192678 polymorphism showed no relationship between sports level and genetic expression (p=0.776). Also the statistical tests for type of sports did not bring any significant results (p=0.164). The outcome of this is the assumption that the investigated polymorphism is not relevant for

sports performance in the investigated groups. The results of Eynon et al. (2009, p. 1147) and Eynon et al. (2010, p. e145) are contrary to these findings. The interaction between the PPARD T294CC and the PPARGC1A Gly482Ser genotypes showed significant results and therefor it seems to be very likely that those polymorphisms are beneficial on endurance performance (Eynon et al., 2009, p. 1150). However it has to be admitted that separate calculations for the PPARD polymorphism did not result in any significant findings (Eynon et al., 2009, p. 1149). Therefore they conclude that the PPARGC1A Gly482Ser polymorphism has some important influence on the level of endurance performance because it controls other genetic variations like PPARD T294C (Eynon et al., 2009, p. 1151). Statistical tests in 155 Israeli athletes (sprinters and endurance athletes) displayed significant differences of the PPARGC1A Ser482Gly polymorphism frequencies between sprinters and endurance athletes ($p=0.005$), concluding that a lower frequency of the A allele and a higher occurrence of the GG genotype can be associated with increased endurance performance (Eynon et al., 2010, p. e149).

The last endurance related polymorphism which was investigated is the rs11539465 genotype coding for the HIF1A gene. Neither tested for group differences ($p=0.152$) nor for sports types ($p=0.717$) any significant results could be found. This findings are supported by Eynon et al. (2010, p. 861) who also could not find any differences in the frequency of the HIF1A Pro582Ser polymorphisms between endurance athletes, sprinters and a control group. Hence they conclude that the HIF1A Pro582Ser polymorphism is not an important factor in determining sprint or endurance performance (Eynon et al., 2010, p. 861). In exchange Döring et al. (2010, p. 1497) found confirmation for a relationship between the HIF1A Pro582Ser polymorphism and elite endurance athlete status in Caucasian men. Different results were found by Gabbasov et al. (2013, p. 2055). They found a relationship between the appearance of the HIF1A Pro582Ser polymorphism and elite strength athlete (weightlifters, wrestlers) status and conclude that their findings open possible ways for sports selection although they note that further studies have to be done (Gabassov et al., 2013, p. 2057f).

Neither Pearson's Chi-square test for sports type and its relationship to the K153R polymorphism of the Myostatin gene nor the statistical test for the sports groups and their relationship to the K153R polymorphism of the Myostatin gene showed any significant results ($p=0.548$ and $p=0.230$). The rs1805086 locus and its relationship to sports has only been investigated once. Santiago et al. (2011, p. e16323) explain that the MSTN K153R polymorphism can be associated with peak power during muscle contraction and hence it is a candidate genetic variation to explain specific muscular characteristics.

For the IGF-1 -1246C/T polymorphism two comparable studies were found. The findings of this Magisterarbeit showed that there is a not yet significant relationship between the investigated genetic variation and athlete status (p=0.051) as well as between the studied genetic variation and type of sports (p=0.345) are not supported by Ben-Zaken et al. (2013, p. 175). They found contrary results nevertheless their finding that athletes were TT carrier (4.8%) and no subject of the control group carried this genotype are superimposable to the findings presented in chapter 4.9.1. (5.04%; 0.0%). Statistic tests in their cohort showed a significant relationship between the IGF-1 TT genotype and athlete status. They suggest an association between the uncommon IGF-1 TT genotype and endurance athlete status as well as power athlete status in Israeli athletes (Ben-Zaken et al., (2013, p. 175). The data of this master thesis is somehow different because although there are differences in frequencies of the investigated polymorphisms no significant results could be observed. Similar results were found by Ben-Zaken et al. (2014, p. 470). The frequency of the IGF-1 TT genotype (5.3%) in endurance swimmers was comparable to the findings mentioned earlier. They conclude that obviously findings in endurance athletes and power athletes cannot not be adducted to make statements about preferable genetic variations for swimmers (Ben-Zaken et al., 2014, p. 470).

Unfortunately no studies to discuss the results found for the IGF-2 rs3213221 and IGF-2 rs7924316 polymorphisms have been published yet. As a result no clear suggestions can be made and further studies investigating those two polymorphisms have to be done in order to compare the findings.

No significant relationship between athlete status (p=0.324) and type of sports (p=0.811) and the ACTN3 R577X polymorphism were found. Although the data of this master thesis showed a difference in frequencies of the XX and the RR genotype no significant results were found. The results are supported by Eynon et al. (2014, p. 102) who found no link between the ACTN3 R577X polymorphism and elite team sports. Also no relationship between the ACTN3 R577X polymorphism and explosive leg muscle power (in elite Volleyball players) could be found (Ruiz et al., 2011, p. e34) Those findings are contrary to the results of several studies investigating the relationship between the ACTN3 R577X polymorphism and power athlete status (Kikuchi et al., 2015; Eynon et al., 2012; Enyon et al., 2009c). An important role of the ACTN3 R allele for top-level sprint performance (and therefore for power and strength) was assumed by Enyon et al. (2009c, p. 695). The suggestion of Enyon et al. (2009c, p. 695) that the RX and the XX genotypes may influence endurance performance was disproved by Döring et al. (2010a, p. 1355) for Caucasian men. Kikuchi et al. (2015, p. 1) found a significant relationship between the

ACTN3 RR and the ACTN RX genotype and elite sprint/power athlete status as well as long-distance athletic status in Japanese athletes.

7. Conclusion

The general observation of this study is that no significant relationships between investigated polymorphisms and sports level were found. Also within the professional athletes no significant relationship between the Soccer players and the Handball players with regard to genetic variations could be found. As mentioned in the introduction of this master thesis it aimed to find significant relations between sports, sports level (sports type), and genetic variation. Based on those findings a polygenetic profile for Austrian Soccer players and Handball players should have been derived.

Unfortunately and trough the lack of not enough data it was not possible to determine a polygenetic profile for Austrian athletes. Difficulties occurred because too many different genetic variations are associated with performance capacity. This variety of candidate genes makes it challenging to choose the right polymorphisms for the development of a polygenetic profile because little changes can have big effects.

Another problem was that no data was collected concerning the different playing positions of the subjects. It can be assumed that (e.g.) forwards have different phenotypes than goalkeepers. The absence of this information leads to a heterogeneity of the phenotype and its frequencies. Study investigating different playing positions and their relationship to specific genetic variations must be realized to find better results.

Additionally it is necessary to clarify elite athlete status. Without a clear definition similar problems as mentioned in the paragraph before are very likely to happen. It is necessary to develop objective criteria (e.g. time for a marathon, time for 100m; participation in Olympic Games, world championships, European championships, etc.) for the group of professional athletes.

Finally it can be concluded that more researches have to be done. The present master thesis could not find any relationship between athletic status and genetic variations. Additionally no relationship between type of sports and genotype distribution could be found. To close this gap more studies with more detailed information (e.g. playing position) have to be published. This is crucial in order to develop a polygenetic profile for Austrian athletes.

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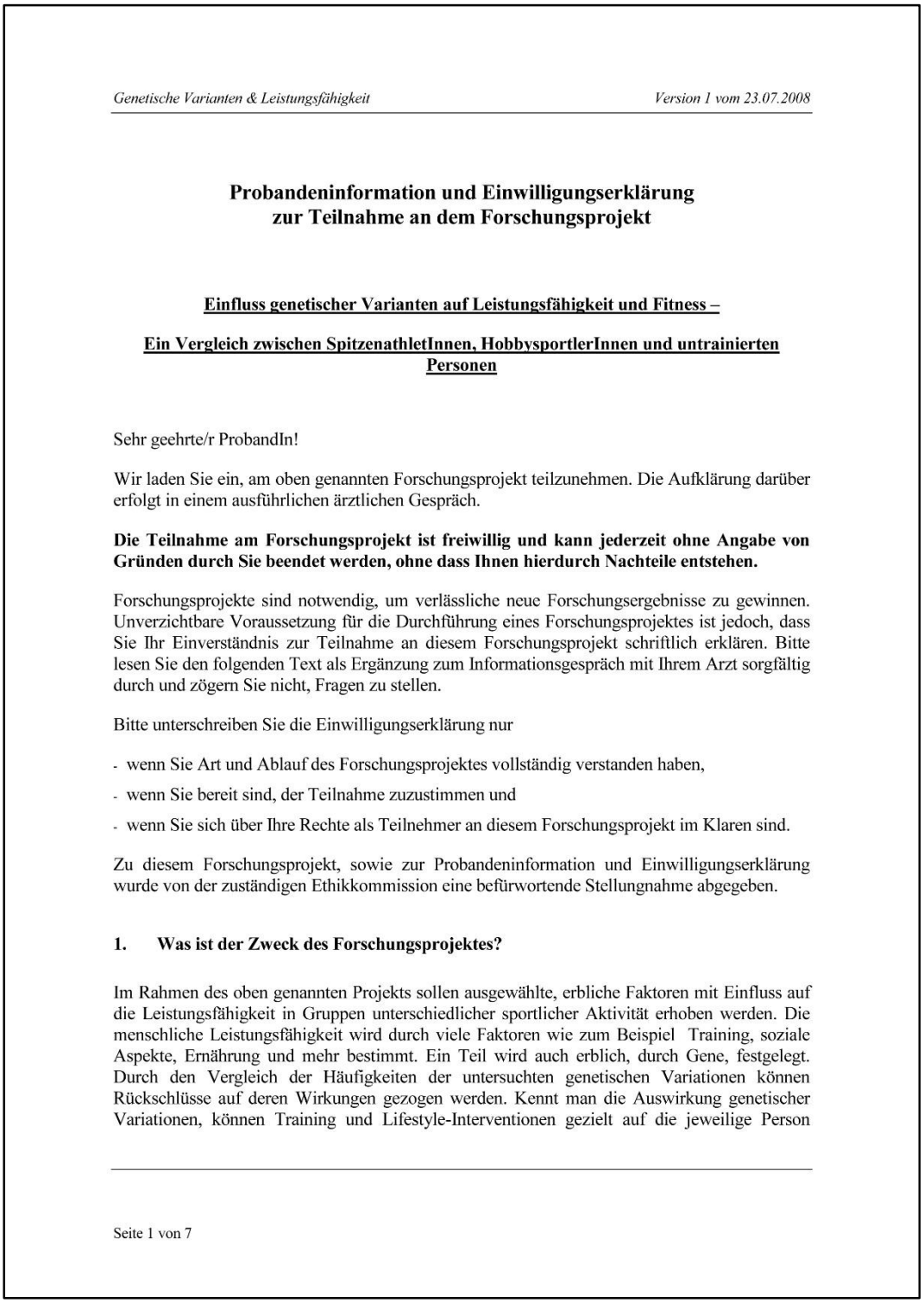
9. Anhang

9.1. Probandeninformation und Einwilligungserklärung



Probandeninformation und Einwilligungserklärung zur Teilnahme an dem Forschungsprojekt

Einfluss genetischer Varianten auf Leistungsfähigkeit und Fitness –

Ein Vergleich zwischen SpitzenathletInnen, HobbysportlerInnen und untrainierten Personen

Sehr geehrte/r ProbandIn!

Wir laden Sie ein, am oben genannten Forschungsprojekt teilzunehmen. Die Aufklärung darüber erfolgt in einem ausführlichen ärztlichen Gespräch.

Die Teilnahme am Forschungsprojekt ist freiwillig und kann jederzeit ohne Angabe von Gründen durch Sie beendet werden, ohne dass Ihnen hierdurch Nachteile entstehen.

Forschungsprojekte sind notwendig, um verlässliche neue Forschungsergebnisse zu gewinnen. Unverzichtbare Voraussetzung für die Durchführung eines Forschungsprojektes ist jedoch, dass Sie Ihr Einverständnis zur Teilnahme an diesem Forschungsprojekt schriftlich erklären. Bitte lesen Sie den folgenden Text als Ergänzung zum Informationsgespräch mit Ihrem Arzt sorgfältig durch und zögern Sie nicht, Fragen zu stellen.

Bitte unterschreiben Sie die Einwilligungserklärung nur

- wenn Sie Art und Ablauf des Forschungsprojektes vollständig verstanden haben,
- wenn Sie bereit sind, der Teilnahme zuzustimmen und
- wenn Sie sich über Ihre Rechte als Teilnehmer an diesem Forschungsprojekt im Klaren sind.

Zu diesem Forschungsprojekt, sowie zur Probandeninformation und Einwilligungserklärung wurde von der zuständigen Ethikkommission eine befürwortende Stellungnahme abgegeben.

1. Was ist der Zweck des Forschungsprojektes?

Im Rahmen des oben genannten Projekts sollen ausgewählte, erbliche Faktoren mit Einfluss auf die Leistungsfähigkeit in Gruppen unterschiedlicher sportlicher Aktivität erhoben werden. Die menschliche Leistungsfähigkeit wird durch viele Faktoren wie zum Beispiel Training, soziale Aspekte, Ernährung und mehr bestimmt. Ein Teil wird auch erblich, durch Gene, festgelegt. Durch den Vergleich der Häufigkeiten der untersuchten genetischen Variationen können Rückschlüsse auf deren Wirkungen gezogen werden. Kennt man die Auswirkung genetischer Variationen, können Training und Lifestyle-Interventionen gezielt auf die jeweilige Person

Figure 34: Probandeninformation Seite 1 (Brandstetter, 2009, p. II)

abgestimmt werden. Dies ermöglicht eine effektive Planung von sowohl gesundheitsorientiertem als auch leistungsorientiertem Training.

2. Wie läuft das Forschungsprojekt ab?

Dieses Forschungsprojekt wird am Zentrum für Sportwissenschaft und Universitätssport durchgeführt. Untersucht werden sowohl jeweils 250 österreichische SpitzensportlerInnen auf internationalem Niveau als auch 250 untrainierte Personen bzw. GesundheitssportlerInnen.

Voraussetzungen für die Teilnahme an der Studie:

- gute Gesundheit
- kein manifester Diabetes mellitus Typ I (insulinpflichtig) oder Typ II
- kein Vorliegen von koronaren Herzkrankheiten
- kein Vorliegen von chronischen Erkrankungen
- Vollendetes 18. Lebensjahr

In die Gruppe der GesundheitssportlerInnen/untrainierte Personen fallen all jene, die

- maximal 3 Stunden/Woche sportliche Aktivität oder anstrengende Freizeitbeschäftigung ausüben und früher nicht professionell sportlich aktiv waren.

Für die LeistungssportlerInnen aus den verschiedenen Disziplinen gelten folgende Einschlusskriterien:

- Ski alpin: Zugehörigkeit zum Österreichischen Skiverband National-, A-, B- oder C-Kader
- Snowboard: Zugehörigkeit zum Österreichischen Skiverband National-, A-, B- oder C-Kader
- Skilanglauf: Zugehörigkeit zum Österreichischen Skiverband National-, A-, B- oder C-Kader
- Biathlon: Zugehörigkeit zum Österreichischen Skiverband National-, A-, B- oder C-Kader
- Nordische Kombination: Zugehörigkeit zum Österreichischen Skiverband National-, A-, B- oder C-Kader
- Sprunglauf: Zugehörigkeit zum Österreichischen Skiverband National-, A-, B- oder C-Kader
- Leichtathletik: Mittel- und LangstreckenläuferInnen (800 Meter bis Marathon, inklusive Hindernis) sowie TeilnehmerInnen an den Wurfdisziplinen (Speer, Kugel, Diskus, Hammer) mit einer aktuellen oder früheren Leistung, die 800 oder mehr Punkten laut IAAF Punktetabelle entspricht.

Figure 35: Probandeninformation Seite 2 (Brandstetter, 2009, p. III)

- Straßenradfahren: Teilnahme an Weltcup-Rennen der Internationalen Radsportunion in der aktuellen oder einer früheren Saison und Zugehörigkeit zu einem professionellem Radteam
- Gewichtheben: aktuelle oder frühere Leistung von mind. 2kg/kg Körpergewicht (Frauen) bzw. mind. 2,5kg/kg Körpergewicht (Männer)
- Kraftdreikampf: Aktuelle oder ehemalige Punkteleistung für die Angehörigkeit am internationalen A- oder B-Kraftdreikampfkader (Männer: >409 Punkte; Frauen: >369 Punkte)

Ihre Teilnahme an dieser Studie erfolgt einmalig durch die Abnahme einer Speichelprobe und Ausfüllen eines Fragebogens zur Erhebung des Trainingsumfanges bzw. der körperlichen Aktivität.

Speichelproben werden mittels speziellen Bürstchen zur Speichelsammlung von der Mundschleimhaut entnommen.

Bei dieser Form der Probenziehung bestehen für die ProbandInnen keine Risken und nur ein Minimum an Aufwand. Die Probenabnahme erfolgt zweimal direkt nacheinander (einmal an der rechten Wange, einmal an der linken Wange). Die Dauer der Probenabnahme beträgt nur wenige Minuten, davor soll für eine Dauer von 30 Minuten nicht gegessen oder getrunken werden. Die Probenabnahme erfolgt an einem von Ihnen frei wählbaren, angenehmen Zeitpunkt.

Die in den Zellen der Speichelprobe enthaltene Erbinformation (DNA) wird anschließend gereinigt und isoliert.

Mittels „real-time polymerase chain reaction", einer molekularbiologischen Methode, wird die DNA vervielfältigt und die Abfolge der darin enthaltenen Basen bestimmt. Dabei können individuelle Unterschiede (so genannte genetische Varianten oder Polymorphismen) sichtbar gemacht werden. Die Häufigkeit der gefundenen Unterschiede wird zwischen den verschiedenen Untersuchungsgruppen (SpitzensportlerInnen und untrainierte Personen) mittels statistischer Verfahren verglichen.

Die Studie verfolgt rein wissenschaftliche Zwecke und wird anonymisiert durchgeführt. Das bedeutet, dass Proben im Nachhinein nicht mehr mit dem/der SpenderIn in Verbindung gebracht werden können. Deshalb können individuelle Studienergebnisse nicht mitgeteilt werden. Informationen über allgemeine Studienergebnisse können Sie jedoch bei Univ.-Prof. Dr. med. Norbert Bachl oder DI Dr. Barbara Wessner erhalten.

3. Was sind Genvarianten (Polymorphismen)? Welche Gene werden untersucht?

Die Studie dient zur Erforschung möglicher erblicher Faktoren, die Einfluss auf die Leistungsfähigkeit haben.

Figure 36:Probandeninformation Seite 3 (Brandstetter, 2009, p. IV)

Obwohl der Großteil der genetischen Information bei gleichgeschlechtlichen Personen identisch ist, treten kleine Unterschiede, so genannte Genvarianten oder Polymorphismen, auf. Diese sind angeboren und können im Lebensverlauf nicht mehr verändert werden. Interessanter Weise können sie aber die Wirkungsweise von Eiweißstoffen im Körper und somit unter anderem die Leistungsfähigkeit beeinflussen.

Diese wird in jedem Fall von vielen Faktoren, wie Training, Ernährung und Umwelt, beeinflusst. Die Vererbbarkeit ist ein weiterer Einflussfaktor und wird nach derzeitigen Schätzungen für 25-40% der Leistungsfähigkeit verantwortlich gemacht.

Es ist anzunehmen, dass bestimmte Genvarianten, die sich günstig auf die Leistungsfähigkeit auswirken, vermehrt bei Hochleistungssportlerinnen auftreten. Dies soll durch den Vergleich mit einer untrainierten bzw. Gesundheitssportgruppe nachgeprüft werden.

Die sportliche Leistung als auch die Wirkung von Training und die Bereitschaft zu sportlicher Aktivität werden von vielen Faktoren bestimmt. Es gibt schon zahlreiche Studien, die den Zusammenhang zwischen einzelnen genetischen Varianten und der Leistungsfähigkeit untersuchten. Da letztere jedoch ein sehr komplexes System darstellt, sollen im Rahmen dieses Projekts insgesamt 22 verschiedene Genvarianten, die als Einflussfaktoren in Frage kommen, untersucht und verglichen werden. Diese können in die Gruppen Ausdauer, Kraft, Ausdauer&Kraft, Sportliche Betätigung und Kohlenhydratstoffwechsel unterteilt werden:

Getestete Genvarianten	Funktion
Ausdauer	
Adrenozeptor α-2A	Rezeptor für Epinephrin (Stresshormon)
Adrenozeptor β-1	Steuerung der Herzkraft und -frequenz
Adrenozeptor β-2	Entspannung glatter Muskulatur
Nuclear respiratory factor 2	Transkriptionsfaktor (spezifische Aktivierung des Proteinaufbaus)
PPARG coactivator 1α	Bildung von Mitochondrien (Kraftwerke der Zelle)
Kraft	
Myostatin	Negativ regulierender Wachstumsfaktor
Insulin-ähnlicher Wachstumsfaktor 1	Stimulation der Zellteilung
Insulin-ähnlicher Wachstumsfaktor 2	Zellteilung und -entwicklung
Hepatische Triglycerid Lipase	Fettabbauendes Enzym
Vitamin D Rezeptor	Vermittlung der Vitamin D Wirkung
Ausdauer & Kraft	
Angiotensin I-konvertierendes Enzym	Blutdruck-, Wasser- und Elektrolythaushaltregulierung
α-Actinin 3	Ermöglicht kräftige Kontraktionen bei hohen Geschwindigkeiten in Muskelfasern
Adenosine-Monophosphat-Deaminase	Energiebereitstellung bei intensiven Belastungen
Kreatinkinase	Schnelle, kurzzeitige Erholung von Energielieferanten

Figure 37: Probandeninformation Seite 4 (Brandstetter, 2009, p. V)

Sportliche Betätigung	
Leptin receptor	Wirkungsvermittlung des Sättigungshormons Leptin
Melanocortin 4 receptor	Signalvermittlung an das Sättigungszentrum im Gehirn
Kohlenhydratstoffwechsel	
Adrenergic β-2 receptor	Regulierung des Energiestoffwechsels
Adrenergic β-3 receptor	Fettstoffwechsel und Wärmebildung
AMP-activated protein kinase γ3	Energiebereitstellung
Leptin	Steuerung der Nahrungsaufnahme
Peroxisome proliferative activated receptor γ	Regulierung des Fett- und Zuckerstoffwechsels
Uncoupling protein 2	Wärmebildung und erhöhter Energieverbrauch

Falls sie genauere Informationen zu den einzelnen Genvarianten wünschen, klären wir etwaige Fragen gerne in einem persönlichen Gespräch.

4. Worin liegt der Nutzen einer Teilnahme an dem Forschungsprojekt?

Es ist nicht zu erwarten, dass Sie aus Ihrer Teilnahme an dieser Studie einen unmittelbaren gesundheitlichen Nutzen ziehen werden.

Die Ergebnisse dieser Studie können dazu beitragen, in Zukunft individuelle Trainingspläne für jeden einzelnen zu gestalten. Der Vorteil darin liegt in einer besseren Ausschöpfung der maximal möglichen Leistungsfähigkeit im Spitzensport als auch dem gezielten Entgegenwirken von Risikofaktoren und der Planung effizienterer Gesundheitsmaßnahmen für untrainierte Personen bzw. HobbysportlerInnen (z.B. bessere Wirkung von Kraft- oder Ausdauertraining für die entsprechende Person).

5. Gibt es Risken, Beschwerden und Begleiterscheinungen?

Es gibt keine gesundheitliche Risken, Beschwerden und Begleiterscheinungen, die durch die Teilnahme an der Studie entstehen.

6. In welcher Weise werden die im Rahmen dieses Forschungsprojekts gesammelten Daten verwendet?

Das Untersuchungsmaterial wird ausschließlich für die hier angeführten Untersuchungen herangezogen. Es werden keine anderen Tests damit durchgeführt. Nach Abschluss der Untersuchungen und Veröffentlichung der Ergebnisse wird das Probenmaterial verworfen (voraussichtlich Ende 2011). Während der Untersuchung wird das Probenmaterial am Institut für Sportwissenschaften, Universität Wien, unter der Verantwortung von Univ.-Prof. Dr. med. Norbert Bachl aufbewahrt. Der Schutz vor dem Zugriff Unbefugter ist durch einen beschränkten Zugang zum Labor gewährleistet.

Figure 38: Probandeninformation Seite 5 (Brandstetter, 2009, p. VI)

Die Datenaufarbeitung erfolgt in anonymisierter Form, das heißt, nachträglich ist für niemanden eine Verknüpfung des genetischen Materials mit der Identität der entsprechenden Person möglich. Auch der Prüfarzt und Studienleiter kann die Probe nicht der Identität einer Person zuordnen. Spätere Änderungen einer einmal getroffenen Entscheidung sind daher nicht möglich, die Anonymisierung bietet das höchste Maß an Sicherheit. Auch individuelle Studienergebnisse können daher nicht mitgeteilt werden.

Die Weitergabe der Daten im In- und Ausland erfolgt ausschließlich zu statistischen Zwecken und Sie werden ausnahmslos darin nicht namentlich genannt. Auch in etwaigen Veröffentlichungen der Daten dieses Forschungsprojektes werden Sie nicht namentlich genannt. Weiters ist es nicht möglich, Sie auf Grund anderer Informationen als Person zu identifizieren.

7. Entstehen für die Teilnehmer Kosten? Gibt es einen Kostenersatz oder eine Vergütung?

Durch Ihre Teilnahme an diesem Forschungsprojekt entstehen für Sie keine zusätzlichen Kosten. Von unserer Seite gibt es keinen Kostenersatz und keine Vergütung.

8. Möglichkeit zur Diskussion weiterer Fragen

Für weitere Fragen im Zusammenhang mit diesem Forschungsprojekt stehen Ihnen Ihr Prüfarzt und seine MitarbeiterInnen gern zur Verfügung. Auch Fragen, die Ihre Rechte als ProbandIn und TeilnehmerIn an diesem Forschungsprojekt betreffen, werden Ihnen gerne beantwortet.

Studienleiter: Univ.-Prof. Dr. med. Norbert Bachl

Erreichbar unter: 0664/602 77-488 70

Name der Kontaktperson: DI Dr. Barbara Wessner

Ständig erreichbar unter: 0664/3154930

Figure 39: Probandeninformation Seite 6 (Brandstetter, 2009, p. VII)

Einwilligungserklärung

Name des/der ProbandIn in Druckbuchstaben:

.....

Ich erkläre mich bereit, an dem Forschungsprojekt **„Einfluss genetischer Varianten auf Leistungsfähigkeit und Fitness - ein Vergleich zwischen Spitzenathleten, Hobbysportlern und Untrainierten“** teilzunehmen. Ich bin von Dr. med. Norbert Bachl oder einem seiner Mitarbeiter ausführlich und verständlich über genetische Analysen, mögliche Risiken, sowie über Wesen, Bedeutung und Tragweite des Forschungsprojektes aufgeklärt worden. Ich habe darüber hinaus den Text dieser Probandenaufklärung und Einwilligungserklärung, die insgesamt 7 Seiten umfasst, gelesen. Aufgetretene Fragen wurden mir vom Prüfarzt verständlich und genügend beantwortet. Ich hatte ausreichend Zeit, mich zu entscheiden. Ich habe zurzeit keine weiteren Fragen mehr.

Ich werde den ärztlichen Anordnungen, die für die Durchführung des Forschungsprojektes erforderlich sind, Folge leisten, behalte mir jedoch das Recht vor, meine freiwillige Mitwirkung jederzeit zu beenden. Nach Probenabnahme kann ich meine Entscheidung nicht mehr ändern, da meine Probe anonymisiert wurde. Die Anonymisierung bietet für mich als ProbandIn jedoch die höchste Sicherheit des Datenschutzes und ich kann danach nicht mehr als Person identifiziert werden.

Ich bin zugleich damit einverstanden, dass meine im Rahmen dieses Forschungsprojektes ermittelten Daten aufgezeichnet werden.

Beim Umgang mit den Daten werden die Bestimmungen des Datenschutzgesetzes beachtet.

Eine Kopie dieser Probandeninformation und Einwilligungserklärung habe ich erhalten. Das Original verbleibt beim Prüfarzt.

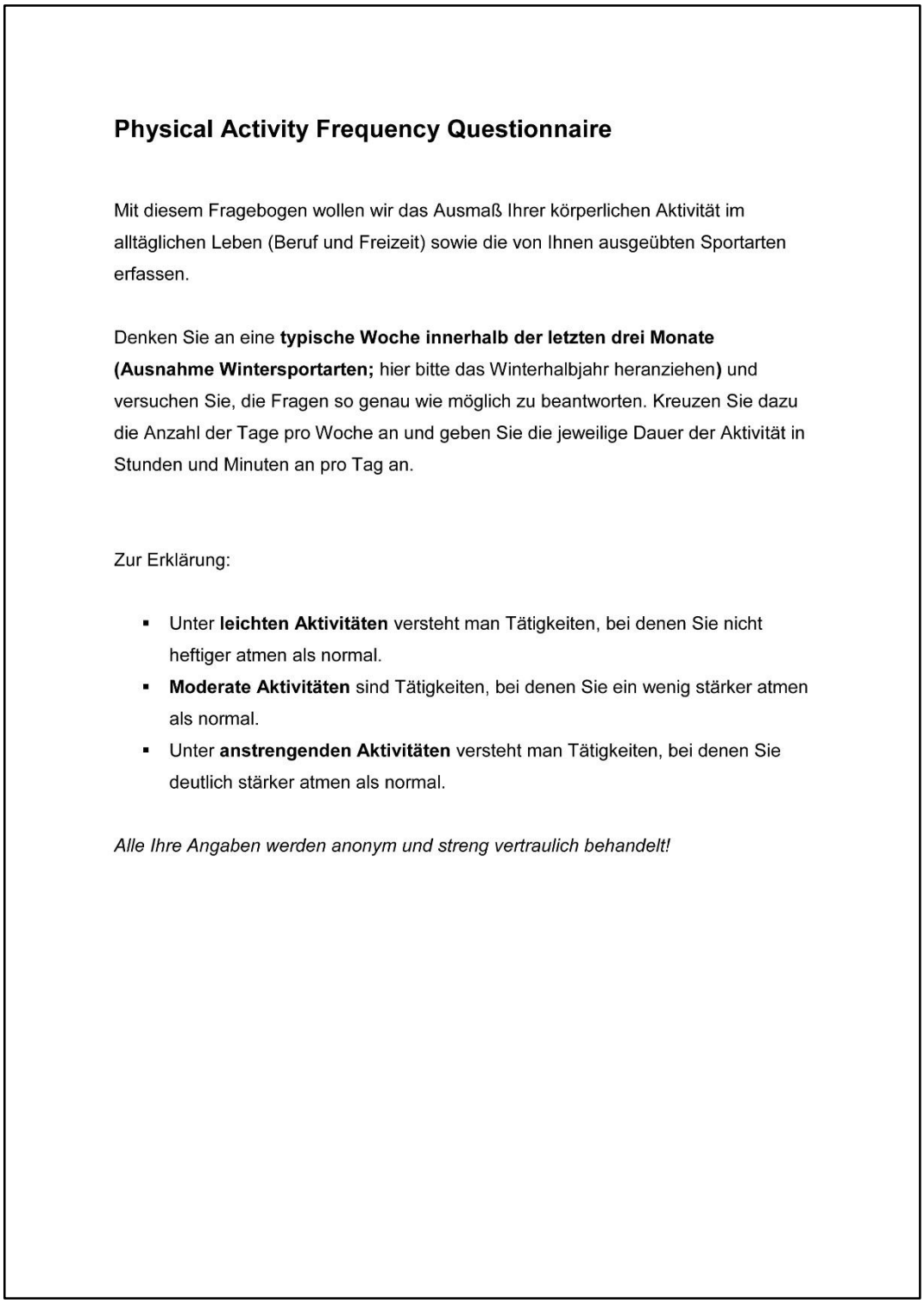
.....
(Datum und Unterschrift des/der ProbandIn)

.....
(Datum, Name und Unterschrift des verantwortlichen Arztes)

(Der/Die ProbandIn erhält eine unterschriebene Kopie der Probandeninformation und Einwilligungserklärung, das Original verbleibt im Forschungsprojektordner des Prüfarztes.)

Figure 40: Probandeninformation Seite 7 (Brandstetter, 2009, p. VIII)

9.2. Fragebogen



Physical Activity Frequency Questionnaire

Mit diesem Fragebogen wollen wir das Ausmaß Ihrer körperlichen Aktivität im alltäglichen Leben (Beruf und Freizeit) sowie die von Ihnen ausgeübten Sportarten erfassen.

Denken Sie an eine **typische Woche innerhalb der letzten drei Monate (Ausnahme Wintersportarten;** hier bitte das Winterhalbjahr heranziehen**)** und versuchen Sie, die Fragen so genau wie möglich zu beantworten. Kreuzen Sie dazu die Anzahl der Tage pro Woche an und geben Sie die jeweilige Dauer der Aktivität in Stunden und Minuten an pro Tag an.

Zur Erklärung:

- Unter **leichten Aktivitäten** versteht man Tätigkeiten, bei denen Sie nicht heftiger atmen als normal.
- **Moderate Aktivitäten** sind Tätigkeiten, bei denen Sie ein wenig stärker atmen als normal.
- Unter **anstrengenden Aktivitäten** versteht man Tätigkeiten, bei denen Sie deutlich stärker atmen als normal.

Alle Ihre Angaben werden anonym und streng vertraulich behandelt!

Figure 41: Physical Acitivity Questionnaire Seite 1 (Brandstetter, 2009, p. X)

Teil 1: Persönliche Daten

Code _____	*(vom Interviewer auszufüllen)*
Geschlecht	☐ Männlich
	☐ Weiblich
Geburtsdatum (TT/MM/JJ)	
Gewicht (kg)	
Größe (m)	

Teil 2: Körperliche Aktivität am Arbeits-/Studienplatz

Aktivität	Tage pro Woche								Durchschnittliche Dauer pro Tag	
	0	1	2	3	4	5	6	7	Stunden	Minuten
Vorwiegend sitzende Tätigkeit (z.B. Bürotätigkeit)										
Vorwiegend stehende oder gehende Tätigkeit (z.B. VerkäuferIn, LaborantIn)										
moderate körperliche Tätigkeit (z.B. RaumpflegerIn)										
Anstrengende körperliche Tätigkeit (z.B. BauarbeiterIn)										

Teil 3: Körperliche Aktivität zur Fortbewegung

Fortbewegungsart	Tage pro Woche								Durchschnittliche Dauer pro Tag	
	0	1	2	3	4	5	6	7	Stunden	Minuten
Auto, Moped, Motorrad										
Öffentliche Verkehrsmittel										
Zu Fuß gehen										
Fahrrad fahren										
Sonstiges: _____										

Figure 42: Physical Acitivity Questionnaire Seite 2 (Brandstetter, 2009, p. XI)

Teil 4: Hausarbeit und Familienfürsorge

Aktivität	Tage pro Woche								Durchschnittliche Dauer pro Tag	
	0	1	2	3	4	5	6	7	Stunden	Minuten
Hausarbeit										
Kinderbetreuung										
Gartenarbeit										
Sonstiges: _____										

Teil 5: Freizeitaktivitäten – Bewegung/Sport

Sportliche Aktivität	Tage pro Woche								Durchschnittliche Dauer pro Tag	
	0	1	2	3	4	5	6	7	Stunden	Minuten
Leichte körperliche Aktivität (z.B. Spazieren gehen)										
Moderate körperliche Aktivität (z.B. langsames Laufen / Fahrrad fahren / Schwimmen)										
Anstrengende körperliche Aktivität (z.B. schnelles Laufen / Fahrrad fahren / Schwimmen / Aerobic)										

Teil 6: Sport – Nähere Angaben *(bitte ankreuzen, Mehrfachnennungen möglich)*

Welche der folgenden Sportarten üben Sie regelmäßig aus?

Sportarten	Regelmäßigkeit		Seit wann?
	ja	nein	Jahre
☐ Laufen			
☐ Radfahren			
☐ Schwimmen			
☐ Rudern			
☐ Wandern			
☐ Skifahren (Wintermonate)			
☐ Snowboarden (Winterm.)			

Figure 43: Physical Acitivity Questionnaire Seite 3 (Brandstetter, 2009, p. XII)

Sportarten	Regelmäßigkeit		Seit wann?
	ja	nein	Jahre
☐ Skiwandern/-langlaufen (Wintermonate)			
☐ Krafttraining/Gymnastik (Kleingeräte, eigenes Körpergewicht)			
☐ Krafttraining (Hanteltraining)			
☐ Aerobic			
☐ Yoga			
☐ Fußball			
☐ Tennis			
☐ Sonstiges: _____ _____ _____ _____			

Teil 7: Inaktiv verbrachte Freizeit

	Durchschnittliche Dauer pro Wochentag	
	Stunden	Minuten
Im Sitzen verbrachte Zeit		
Schlafdauer		

Vielen Dank für Ihre Teilnahme!

Figure 44: Physical Acitivity Questionnaire Seite 4 (Brandstetter, 2009, p. XIII)

10. Lebenslauf

10.1. Persönliche Daten

Zuname:	Teynor
Vorname:	Thomas
Geboren am:	28. Oktober 1987
Geburtsort:	Neunkirchen
Staatsbürgerschaft:	Österreich
Eltern:	Mag. Rita Teynor-Bark (AHS-Lehrerin, geb. am 15. Oktober 1964)
	Dipl.-Ing. Mario Teynor (Architekt, geb. am 9. Dezember 1962)
Geschwister:	Mag. Sandra Teynor (AHS-Lehrerin, geb. am 28. August 1984)
	Mag. Jana Teynor (Bildungsreferentin, geb. am 15. März 1990)

10.2. Schulbildung

September 1994 – Juni 1998	Dr. Adolf Schärf Volksschule Pottschach
September 1998 – Juni 2002	Gymnasium Sachsenbrunn
September 2002 – Juni 2006	Bundesoberstufenrealgymnasium Wiener Neustadt unter besonderer Berücksichtigung der sportlichen Ausbildung
Juli 2004 – Jänner 2005	Brown County Highschool, Nashville, Indiana, USA
Oktober 2006 – November 2010	Bakkalaureatstudium Sportwissenschaften Universität Wien
Seit Oktober 2006	Lehramtsstudium „Bewegung und Sport“ und „Geschichte, Sozialkunde und Politische Bildung“ Universität Wien
Seit November 2010	Magisterstudium Sportwissenschaften Universität Wien

10.3. Aus-, Fort- und Weiterbildungen

2005	Skilehrer-Anwärter (Snowsports Academy)
2006	Rettungsschwimmschein
2006	Staatlich geprüfter Fitlehrwart für Erwachsene (BSPA Wien)
2010	Staatlich geprüfter Triathloninstruktur (BSPA Wien)
2011	Pilates Basic Coach (SPORTUNION)
2011	Trainergrundkurs (BSPA Wien)
2013	Staatlich geprüfter Triathlontrainer (BSPA Innsbruck)

10.4. Berufliche Tätigkeiten

seit 09/2013	SV-Lehrer für „Bewegung und Sport“ und „Geschichte, Sozialkunde und politische Bildung“ am Karl Sonnweber ORG Guntramsdorf
seit 05/2013	Sportwissenschafter in der Gemeinschaftspraxis „die Sportpraxis“
seit 06/2011	selbstständiger Sportwissenschafter und (Triathlon-)Trainer (Athletiktrainer und Krafttrainer der Werner Schlager Academy, Trainingspläne, Personaltrainings, Seminare, etc.)
01/2011 – 09/2014	Sportwissenschafter der Multitraining Schwechat GmbH (zunächst als Freier Dienstnehmer, ab September 2012 im fixen Dienstverhältnis)
10/2012 – 02/2013	Studienassistent am Institut für Sportwissenschaften der Universität Wien in der Abteilung „Sport- und Leistungsphysiologie“ (a.o. Univ. Prof. Dr. Gerhard Smekal)
11/2010 – 08/2011	Trainer des Talenteförderzentrum Eisenstadt (Laufteam Burgenland Eisenstadt)
09/2008 – 09/2011	Laufstilanalysen und Beratertätigkeiten für New Balance Österreich

03/2009 – 11/2011	Mitglied des Organisationskomitees des BEWEG-Energy-Run in Eisenstadt
04/2010 – 05/2010	Praktikum am Österreichischen Institut für Sportmedizin (ÖISM) mit dem Aufgabengebiet der Durchführung von Laufstilanalysen und Trainingsberatung von Hobbysportlern im Rahmen der Ausstellung „2500 Jahre Mythos Marathon“
01/2007 – 10/2008	Trainingsbetreuung im Post-Reha-Bereich des Physiopoint Neunkirchen