

UNIVERSITI TEKNOLOGI MARA

**AN ASSOCIATION STUDY
BETWEEN SPORTS-RELATED
GENETIC POLYMORPHISMS AND
PERFORMANCE
CHARACTERISTICS AMONG THE
ELITE ATHLETES**

NOURUL EMMILIA BINTI MOHD FAZLI

Thesis submitted in fulfillment
of the requirements for the degree of
Master of Science
(Sports Science and Recreation)

Faculty of Sports Science and Recreation

March 2022

ABSTRACT

Human physical performance is a multigenic trait. Variations in athletic performance may be attributed to a polygenic profile instead of a single genetic variant. This research aims to determine the frequencies of the allele and genotype of ten (10) sports-related genetic polymorphisms (*ACE*, *ACTN3*, *PPARA*, *PPARGC1A*, *ADRB2*, *IL6*, *AGT*, *NOS3*, *MTHFR* and *PPARG*) in Malaysian elite athletes. Cross-sectional study was incorporated to investigate the associations between physical performance and sports-related genetic polymorphisms which are represented by power and endurance TGS. A total of 184 elite athletes comprised of 26 power athletes, 28 endurance athletes, 84 adolescent football players, as well as 46 U19 and U21 football players were genotyped for the polymorphisms. The athletes' genotype data and physical performance data that were collected in prior by professional fitness coaches involving the football players were analysed using Student's t-test, ANOVA, and standard multiple regression (which includes Pearson Correlation) analyses using SPSS software. The study on U19 football players found that players who had better postural stability, power and muscle strength performances had higher power TGS. Significant positive correlations were discovered between muscle strength performance and power TGS. Significant positive correlation was also revealed between the power performance and TGS of power. Standard multiple regression analysis indicated that the power performances parameters are significant in explaining the variation in power TGS. Significant negative correlations between postural stability and TGS of power were also discovered. Standard multiple regression analysis indicated that postural stability parameters are significant in explaining the variation in endurance power TGS. The study on adolescent football players found that players who had significantly better speed and acceleration, as well as muscle strength and endurance performances were revealed to have significantly higher mean of power TGS. Significant positive correlation between 20 m sprint and endurance TGS, as well as negative correlation between handgrip and endurance TGS were also found. Standard multiple regression analysis indicated that muscle strength and endurance parameters are significant in explaining the variation in endurance TGS among the players. Therefore, the research suggested that genetic factors are useful in personalizing athletes' regular training programs to improve sports performances.

ACKNOWLEDGEMENT

Firstly, I would like to express a special gratitude to my supervisor, Prof. Dr. Teh Lay Kek, who expertly guided me throughout the study and kept me constantly engaged with my research; thus, challenging me to live up to my potential. Her unwavering enthusiasm for science and dedication as both a teacher and scientist who has given me guidance, encouragement, criticism, advice and also intellectual freedom are truly inspiring and appreciated. My appreciation also extends to my co-supervisor, Associate Prof. Dr. YM Raja Mohammed Firhad Raja Azidin and the Director of Integrative Pharmacogenomics Institute (iPROMISE), Prof. Dato' Dr. Mohd Zaki Salleh for their valuable suggestions, time and efforts to guide me from the beginning until the end of this project. I would also like to express my sincere thanks to the Supervisory Committee of Postgraduate at Integrative Pharmacogenomics Institute (iPROMISE) which was set up to monitor progress of my study and had provided me with technical and scientific input to improve the study. I also thank the Faculty of Sports Science and Recreation of UiTM community for their constant support, guidance and encouragement. Above ground, I am fully indebted to my parents, family, and friends, whose value to me only grows with time.

TABLE OF CONTENT

	Page
CONFIRMATION BY PANEL OF EXAMINERS	ii
AUTHOR'S DECLARATION	iii
ABSTRACT	iv
ACKNOWLEDGEMENT	v
TABLE OF CONTENT	vi
LIST OF TABLES	ix
LIST OF FIGURES	xii
LIST OF SYMBOLS	xv
LIST OF ABBREVIATIONS	xvii
CHAPTER ONE INTRODUCTION	1
1.1 Research Background	1
1.2 Research Scope and Delimitation	3
1.3 Problem Statement	4
1.4 Research Questions	5
1.5 Research Objectives	6
1.6 Research Hypotheses	6
1.7 Significance of Study	7
1.8 Operational Terms	7
CHAPTER TWO LITERATURE REVIEW	9
2.1 Genetic Polymorphisms	9
2.2 Sports Genomics	10
2.3 Sport-related Genetic Polymorphisms	10
2.4 Endurance-related Genetic Polymorphisms	11
2.5 Power/strength-related Genetic Polymorphisms	14
2.6 Polygenic Scores	19
2.7 Predictors of Talent in Football	20
2.7.1 Anthropometric Measurements	21

CHAPTER ONE

INTRODUCTION

1.1 Research Background

In recent years, correlations between genetic factors and human physical performance are widely studied. These findings have led to the emergence of a new scientific discipline that focuses on the genomic architecture of elite athletes known as sports genomic. Therefore, numerous association studies have been conducted in the past few years to unravel the genetic endowments that might explain the differences in the levels of athletic performance among individuals, particularly elite athletes, by profiling naturally occurring genetic polymorphisms in their genomes (Maciejewska et al., 2012; Ahmetov & Fedotovskaya, 2015; Jacob et al., 2021). The components of the athletic performance include endurance, strength, power, cardiovascular capacity, and neuromuscular coordination (Dror et al., 2015).

Sport-related genetic polymorphisms are heritable influence over components of the athletic performance (Williams & Folland, 2008). Given that the human physical performance is a multigenic trait (Lucía et al., 2010; Guilherme et al., 2014), the variation in athletic performance may be attributed to a polygenic profile instead of a single genetic variant. Variants in genes that are involved in many biological systems of the body may be potentially associated with the elite athletes' phenotypes. To date, there are over 120 sport-related genetic polymorphisms associated with elite athlete status. These include endurance-related genetic markers (Ruiz et al., 2009; Ahmetov & Fedotovskaya, 2015) and power/strength-related genetic markers (Ruiz et al., 2010; Ahmetov & Fedotovskaya, 2015; Maciejewska-Skrendo et al., 2019). Among those sport-related genetic markers, positive associations with elite athlete status have been replicated in a number of those sport-related genetic markers (Ahmetov & Fedotovskaya, 2015). As of now, positive associations with athlete status have been shown in at least ten of these polymorphisms (*ACE*, *ACTN3*, *PPARA*, *PPARGC1A*, *ADRB2*, *IL6*, *AGT*, *MTHFR*, *NOS3*, and *PPARG*) in several studies.

Football is considered as the world's number one sport. A large number of people in the world are actively involved in the game of football (Kunz, 2007). According to the latest statistics based on the findings of a large-scale FIFA survey