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Study of GDF8 polymorphisms and their influence on musculature and strength in weightlifting

Abstract

Background: Further deepening is required in studies that include objective indicators of sports performance and its direct correlation with the genotype in the analysis on variations of the GDF8 Gene and its association to the sports phenotype in the weightlifting discipline in the department of Valle del Cauca, Colombia**.**

Objective: To determine the presence of genetic variants in the GDF8 gene in weightlifters belonging to the Valle del Cauca Weightlifting League.

Methods: Descriptive cross-sectional study of 25 athletes from the Valle del Cauca Weightlifting League, detection of genetic variants by genotyping through next generation sequencing from peripheral blood samples, and use of the IBM® SPSS® Statistics 21 statistical program.

Results: 92% of the participating athletes presented the genetic variant rs1805086 of the GDF8 gene; 57% of the population that presented the variant were male, and 91% of the population that presented the variant were of African descent. It was found that 43% of the population with the presence of the variant obtained more than 5 local medals and more than 1 medal in international tournaments.

Conclusions: A frequency of 92% for the R allele of the rs1805086 polymorphism (MSTN K153R) observed in the study population suggests a possible selection effect associated with the participants' sports profile and highlights the importance of this genetic marker as a potential predictor of performance in strength and power disciplines.

Keywords: weightlifting, GDF8 Gene, myostatin, weightlifting, strength.

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Introduction

Weightlifting is a sports discipline that aims to develop strength in weightlifters,[1] it is classified as an individual Olympic to cyclical sport of low mobility and maximum intensity, which consists of lifting the maximum weight through movement or technical executions.[1,2] Over the years, weightlifting as a sports discipline has become one of the most characteristic and important sports in Colombia.

This sport has various techniques such as the snatch and clean and jerk, its incidence is primarily at the neurological level, thus achieving a greater recruitment of muscle fibers. This is relevant if the execution characteristics of sports gestures are of great speed, because a greater predominance of muscle fiber recruitment is developed, as it is an acyclic exercise.[3,4]

The fundamental motor skills of sports performance, in training where neuromuscular adaptations and metabolic alterations occur improve the performance of athletes prior to the competitive period and the plasticity of muscle tissue, the GDF8 gene, which codes for a protein called myostatin (MSTN) which is a growth factor with action, which is predominant in the fibers of skeletal muscle, generating muscle growth and hypertrophy,[5] this condition is closely related to increased strength and therefore in sports performance.

The protein myostatin (MSTN) was discovered in 1997 and is encoded by the MSTN gene, located on chromosome 2 2q32.2, encodes 375 amino acids in three exons and occupies a site of approximately 8 kb, is also known as growth differentiation factor 8 (GDF-8), and acts as a protein hormone that exerts negative regulation of muscle growth.[6]

Different mutations, polymorphisms and genetic variants have been identified in this gene that are of interest in the sports field, as they could be associated with a significant increase in muscle mass, an aspect of vital importance for the development of strength and other motor skills necessary for the development of sports practice, one of them is rs1805086, a single nucleotide polymorphism (SNP) located in the GDF8 gene.[6]

It has been described that the rs1805086 variant, also known as K153R, in which there is a transition at position 2246 changing a Adenine for a Guanine in the AAG codon (AGG), this variation is more frequent in Afro-descendants compared to other populations.[7]

This polymorphism is associated with variations in muscle mass and strength, especially in response to resistance training. The variant allele (also known as the R allele) of the rs1805086 SNP is associated with possible increased muscle mass and strength and is more prevalent in some populations.[8]

Hence the importance of determining the essence of genetic variants in the GDF8 gene in athletes of the Valle del Cauca weightlifting league and potentiating the use of genetic engineering as a way of application for the development of implicit sports skills in the day to day life of high performance athletes in this sport.

Methods

Type of study

A cross-sectional observational descriptive study was conducted in which the presence of genetic variants present in the GDF8 gene was

determined in a group of elite athletes assigned to the Valle del Cauca Weightlifting League.

This study was approved by the human ethics committee of the National School of Sport University Institution regulated by resolution 008430 of October 04, 1993, issued by the Ministry of Health of the Republic of Colombia

Peripheral blood collection for molecular analysis

The peripheral blood samples were taken by a professional in bacteriology and a clinical laboratory trained for this procedure; After reading and signing the informed consent, venipuncture was performed after the previous disinfection of the area to be punctured by means of a cotton swab impregnated with alcohol, cleaning the area with circular movements from inside to outside the area without reviewing, venipuncture was performed using a No. 21 gauge needle attached to a shirt, which will be connected to the lilac-capped vacutainer with EDTA anticoagulant, to perform the blood extraction.

The samples were transported at 4ºC, to the Molecular Biology and Biochemistry laboratory of the Department of Physiological Sciences of the Faculty of Health of the Universidad del Valle for processing.

DNA extraction

DNA extraction was carried out from blood samples previously taken using the commercial Wizard Genomic ® DNA Purification kit 100, following the manufacturer's protocol. Subsequently, the concentrations and degree of purity of the DNA, absorption (260/280) between 1.8 and 2.0, were calculated with the latest generation spectrophotometer Nanodrop, as well as its quality by means of the amplification of a segment of the constituent gene GAPDH. Sequencing was performed using the BigDyeTM terminatorcycle PCR methodology that incorporates Dye or labeled nucleotides to the PCR product or expansion

Statistical analysis

An analysis of frequency distribution and measures of central tendency of the variables studied was performed, then a bivariate analysis was performed using the chi-square test to establish a statistical association between the variables, bearing in mind that a statistically significant association will be one that has ($p < 0.05$), all statistical analyses were performed using the statistical program IBM® SPSS® Statistics 21.

Results

A relevant piece of information in the characterization process is the sports characteristics of the participants, among which sports performance can be highlighted, measured mainly with the number of medals or achievements obtained at the national and international level.

It is highlighted that at least 60% of the population under study has more than 6 years of sports training, this means that their sports knowledge compared to the rest of the population could be greater given the years of experience they have.

Thus, in the variable of local medals it is shown that 13/25 of athletes count as more than 5 local medals, demonstrating that of 100% at least half and one more have been meeting sports preparation objectives, likewise 52% of the population has managed to obtain more than 2 national medals and only 12 5 has managed to get an international medal as indicated in table 1.

Table 1 Sports characteristics of the population

Variable	n (25)	Percentage %
Years of training		
≤6	10	40%
> 6	15	60%
Local Medals		
≤5	12	48%
>5	13	52%
National Medals		
≤ 2	12	48%
>2	13	52%
International medals		
≤1	22	88%
>1	3	12%

After the genetic analysis carried out, it was possible to determine the rs1805086 variant of the GDF8 gene in the long arm of chromosome 2 in region q32.2, of the 25 athletes analyzed, 92% (23/25) presented the genetic variant mentioned above, this distribution is presented in Figure 1.

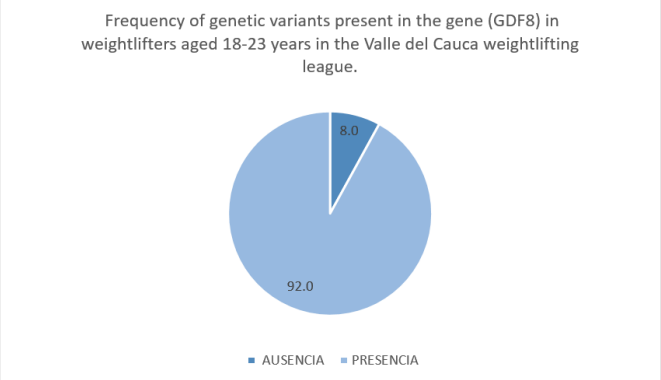

Figure 1 Frequency of genetic variants present in the (GDF8) gene.

The analysis of the genetic interaction showed that the interactions were carried out in 1.40%, the genes converged in 4.35% in the same routes or pathways of action, shared protein domains in 0.59%, co-expression in 13.50%, physical interactions 67.64% and co-localization in 6.17%. See Figure 2.

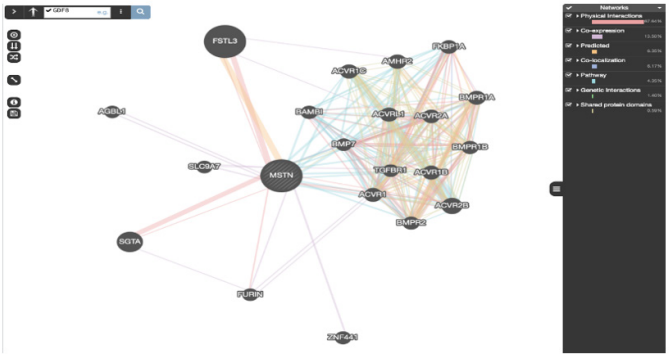

Figure 2 Analysis of the interaction of the GDF8 gene with the GeneMANIA program.

The results show that at least 12 of the 25 athletes have 6 or fewer years of training, these cover 52% of the population, in turn 2 of these athletes do not have the presence of the variant (rs1805086); In turn,

the local medals variable shows us that at least 13 of the 25 athletes count as 5 or less local medals, these are equivalent to 57% of the population and in turn 2 of these athletes present the absence of the variant (rs1805086).

In the national medals variable, it shows us that at least 14 of the 25 athletes have 2 or less national medals, these athletes cover 61% of the population under investigation, without overlooking that 2 of these athletes present the absence of the variant (rs1805086).

Finally, 12 of the 25 athletes have more than 1 international medal, this is equivalent to 43% of the population under investigation, it should be noted that the 12 athletes have the presence of the rs1805086 variant, as shown in Table 2.

Table 2 Association between sports characteristics and the presence of the rs1805086 variant of the GDF8 gene in athletes of the Valle del Cauca weightlifting league.

Variable	Presence of the variant rs1805086		Absence of the variant rs1805086		p valor
	n=23	%	n=2	%	
Years of training					
≤ 6	12	52%	2	100%	0,191
> 7	11	48%	0	0%	
Local Medals					
≤5	13	57%	2	100%	0,229
>5	10	43%	0	0%	
National Medals					
≤2	14	61%	2	100%	0,269
>2	9	39%	0	0%	
International medals					
≤0	11	57%	2	100%	0,157
>1	12	43%	0	0%	

[1] 95% confidence limits testing exclusion of 0 or 1, as indicated.; Statistically significant (p value ≤0.05)).

Discussion

The findings statistically presented in this study are significantly contextual to the extent that the object of research is to determine the presence of genetic variants found in the gene (GDF8) of weightlifters aged between 18-23 years of the Valle del Cauca weightlifting league, in which the presence of the genetic variant rs1805086 found in 23 of the 25 athletes studied was obtained as a covariate. that is, the one corresponding to 98% of the population under investigation.

Authors such as Santiago et al,[9] state that "the gene (MSTN) encodes myostatin, a specific secreted peptide of skeletal muscle that functions mainly to modulate myoblast proliferation and, therefore, muscle mass and strength" (p.1). Similarly, Calbet and Fiol[10] state that "Myostatin is also known as growth and differentiation factor 8 (GDF 8)" (p.5), thus highlighting that myostatin fulfills roles as a negative regulator of muscle growth and in turn also as a muscle regenerator.[11]

In this way, it is understood that the working group based on the results of the variables object of this project and associated with the Gen, allows for greater control of sports planning, since it provides sufficient information for athletes to sustain their sports level at the highest level to guarantee the fulfillment of the proposed objectives.

Genetic variants can contribute to conferring elite athlete status. However, this does not mean that a person with favorable genetic traits would become a champion, "because multiple genetic interactions and epigenetic contributions, along with environmental confounding factors, shape the overall phenotype. The analysis of genetic polymorphisms related to sports performance would provide information on the potential to become an endurance or power elite".[12]

Sometimes it tends to be affirmed that a person is or can aspire to be a high-performance athlete in any sport, because at first glance their anatomical characteristics comply with aspects of competition, without considering relevant, the study or detailed analysis of the subject when involving them in the sports discipline. However, to ensure that a person reaches the highest level of sport, it is not only enough with the discipline and perseverance that sport requires, but also with genetic factors that hereditarily influence sports performance, such as: ethnic group, initiation of sports performance, socio-economic environments, sex, type of muscle fibers, anthropometric measurements, etc.

From the above statement, Ginevičienė, et al [13] reaffirm the analysis that indicates how elite athletes "seem to arise as a result of endogenous processes (such as anatomy, cal, metabolic, functional or behavioral characteristics) that interact with exogenous characteristics influences of environmental factors (such as training, diet, medical care, etc.) In addition, genetic interactions are relevant to physical performance (such as speed, body composition, resistance training, predisposition to injury or behavioural attitude towards competition)" (p.757).

Similarly, Rodríguez [14] states that "the professional growth of an athlete and their chances of becoming part of the sports elite depend to the same extent on two elements of great importance: their genetic inheritance and the sports training they perform" (p.2).

Therefore, the importance of body composition in weightlifting lies in the need to be able to lift the maximum possible weight while maintaining a low fat mass and a high lean mass, since this sport, being regulated according to body weight categories, limits the excessive increase in total body mass.[15] In this way, the detection of talents in the discipline of weightlifting results in a challenge and complexity for sports teams, with the need for a multidisciplinary approach typical of previous professionals, aimed at training and increasing the chances of an athlete reaching their peak, high-performance process.[16]

Finally, Comparing the frequency of 92% for the R K153R allele of the rs1805086 polymorphism in the studied population,[17] there is an unusual increase with respect to the general population where it is less than 7%, while for the Caucasian population an approximate between 3-4% is reported,[9] for the athlete population in strength mode a percentage of 10%, even in African populations, a frequency of around 22% has been reported,[6] This discrepancy could be due to particular characteristics of the population studied such as ethnicity and/or a selection effect by the sports modality, among others, our results ratify that the study group is a highly specialized subpopulation in contrast to the reference population cohorts reported in the literature.[6,9,13,18]

Conclusion

This study developed a conceptual integration, which allows professional training and generation of scientific knowledge from sports sciences, its complement with disciplines from genetic studies from biology, in order to expose greater variables of reading and multidimensional comprehension of athletes in training and high performance, before their training teams, whose purpose is the identification of talents in the sports discipline of weightlifting, their empowerment and sports guidance to optimize the skills and abilities

of each athlete.

The above is exposed, by a correlation between the presence and predominance of the GEN (GDF8) in the athletes of the Valle del Cauca Weightlifting League who have achieved medals in the different competitions that are held at the national and regional level, in relation to variables such as sex, ethnicity and age, being men of Afro-descendant ethnic belonging, who are more likely to maximize their sports performance in what corresponds to the margin of the population sample marked by the study. In this way, understanding athletes from their various dimensions is vital to ensure that training processes and practices are focused on the particularity of the athlete and with this, obtain greater competitive results, without neglecting the variations that may occur, due to the aforementioned variables or others.

The exercise of identification and orientation of sports talents, understanding how the associativity of the GEN (GDF8) can have with its various variations, identified through a process of laboratory and interdisciplinary accompaniment between the various professionals involved in the sports training of the discipline, will allow a better accompaniment in sports, maximize the results that the Valle del Cauca delegation presents in the competitions and co-work with the athlete through biological results, the understanding of his performance in the discipline of weightlifting or in another sport, is related to the opportunities and capacities of sports performance between an athlete where the GEN predominates and another where there is predominance; and with this, the training teams collect the information that leads to strengthen the athlete, according to their aptitudes.

The interdisciplinary and investigative integration in this field of sport, for weightlifting or weightlifting facilitates a result at scale, since it will strengthen in the Leagues the optimization of their processes and efforts to accompany the training and consolidation of more competitive athletes and their results, will leave the organization with greater recognition, benefits and experience in their training methodologies. For the discipline of weightlifting, the findings of the research is a primary and innovative antecedent, in order to be a reference for study to continue its deepening and replication in other disciplines and their sports niches.

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