

# Athletic Performance Genomics

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# OVERVIEW

Strong evidence exists for the role that genetics play in athletic performance. Olympic and elite level athletes carry clear genetic markers that create a propensity for remarkable performance. It is important to point out however, that there are also many Olympic level and elite athletes that possess genetics that are not indicators of elite performance. This seemingly disparate occurrence can be attributed to several factors. First, athletic traits are generally expressed through a combination of genes as opposed to one specific variation in a single base pair out of 3 billion. Second, epigenetics provides the ability to alter the expressions of genes; we can upregulate or down-regulate expression through our environment and lifestyle. (look to see if HFE expression is altered by altitude)

The ACTN3 gene variant as one example; a certain variant of this gene will result reduced or no actinin-3 in muscle fibers, elite sprinters mostly have the ACTN3 variant that codes for its presence in muscle. When it is missing, there is not an upregulation possible, however, with appropriate training, those missing the protein, can up-regulate production of a similar protein called actinin-2 through the gene ACTN2. Despite not having the gene for sprinting, there are still elite level sprinters and power athletes that have the variant without actinin-3.

Finally, the alteration of specific genes also occurs when we take strategic action through the use of nutrigenomics (nutrition and supplementation).





Keep in mind that genetics is about propensities or probabilities, not absolutes. Having ideal genes for elite performance will not make you an elite athlete and not having optimal performance genetics does not make it out of reach.

This translation guide is not designed to provide direct to consumer guidance, but as a way to open a discussion with an Apeiron Academy certified epigenetic coach who understands the art and science of genetics, epigenetics, and the interactive component of using the personal genetic blueprint to guide success in alignment with your specific goals.



The areas we consider in this report are:

- VO2 max the amount of oxygen you can take in and utilize
- Muscle fatigability how quickly your muscles exhaust with exertion
- Recovery how long should you for recovery after intense workouts
- Strength/hypertrophy genes related to the ability to build strength and muscle mass
- Power vs. endurance what is your genetic propensity
- Injury propensity genetic predispositions to soft tissue injury





# VO2 MAX OVERALL

## GENETIC DATA

## GENERAL

GENE	TYPE
BTAF1	СТ
TSHR	Π
GRIN3A	GG
KCNH8	СТ
C9	GG
ZIC4	Π
CAMTA1	СТ
RGS18	AG
BIRC7	GG
NDN	СТ
TTC6	Π
APOE (1)	Π
APOE (2)	CC
HFE(1)	CC
HFE(2)	GG

VO2 max is a common measure of athletic performance and can be used to guide training programs for optimal outcomes. Elite athletes consider optimal VO2 max an essential area to optimize. The is a strong genetic component to VO2 max that can predict 25-50% of the observed variability. This variability falls into multiple physiologic categories that comprise VO2 max.

VO2 max is the highest rate of oxygen consumption that an individual can obtain during maximal exertion. VO2 max is calculated by the Fick equation (cardiac output x arterial O2 – cardiac output x mixed venous O2). So, VO2 max takes into account the following physiologic parameters:

• Cardiac.

- Heart stroke volume (contractility)
- Heart rate
- Vascular tone
- Muscle fiber type slow twitch fibers have
- greater oxidative capacity
- Metabolic machinery of the cell

#### Overall VO2 Max Propensit





# VO2 MAX CARDIOVASCULAR

#### CARDIOVASCULAR

GENE	GENO TYPE
ADRB1	CC
ADRB2 (1)	GG
ADRB2 (2)	CC
NFIA-AS2	GG
EDN1	GG
DBX1	СТ
HIF1A	AG
CREB1	AG
KIF5B	CG
NPY	TT
NOS3(1)	GG
NOS3(2)	СТ



Our genes are involved in the outcomes of each of these areas.

Below is the schematic of how the genetics are involved:





# VO2 MAX METABOLIC

## METABOLIC

GENE	GENO TYPE
DEPTOR	AA
MIPEP	AG
ACSL1	AA
NRF1 (1)	CC
NRF1 (2)	AA
HIF1A	AG
PPARGC1A	CC
СКММ	TT
UCP3	AG
KIF5B	CG
AMPD1	AA

### MUSCLE

TTN	CC
DAAM1	TT N
AMPD1	AA
	At the work

#### Factors that increase VO2 max:

- Increase cardiac output
  - Increase heart rate
  - Increase stroke volume (contractility)
- Dilate capillaries in the muscle
- Increase capillary density in muscle
- Increase utilization and transport of oxygen



#### INTERPRETATIVE NOTES:

You are strong in all aspects of VO2 max except the muscular component and primarily AMPD1. This has to due with the recycling of ATP so it can lead to fatigue during workouts and after. D-ribose powder 3 grams each morning and 1 gram added to water during workout.



# MUSCLE FATIGABILITY

## GENETIC DATA

GENE	GENO TYPE
NATO	
NATZ	AA
HNF4A	AG
AMPD1	AA
COL5A1(1)	
IL15Ra(1)	n/a
AGTR2	
ACE	СТ

In athletic performance, an important parameter to consider is muscle fatigue. Muscle fatigue is defined as the decline in a muscles ability to generate force. Genetics play an important role in guiding optimized protocols but even with the best genetic profiles, without training, there cannot be improvement.

- Genetics
  - Muscle fiber types
  - Metabolic function
  - Oxygen delivery
  - $\circ\,$  Lactate clearance
- Epigenetics Adaptive training

The genetics will provide probabilities or predispositions that can guide interventions which can potentially upregulate the time to fatigue and the epigenetics can be modified by specific training protocols.

Understanding your physiologic propensity for muscle fatigue is essential to optimal performance.



#### **INTERPRETATIVE NOTES:**

Again the AMPD1 is playing into the high fatiguability and the D-ribose should help a great deal with this.



## RECOVERY

## GENETIC DATA

GENE	GENO TYPE	
CRP (1)	CC	
TNF	GG	
SOD2	AG	
IL1B	GG	
IL6	CG	
NAT2	AA	
СКММ	Π	
COL5A1(1)		

Routine intense exercise is not a natural process, in fact, fitness training only became part of human life in the past century. To early hominoids, exercise was part of survival. Hunting required brief bursts of intense activity followed by long periods of rest, agriculturalists generally had prolonged low intensity without the bouts of intense exertion.

How much rest do you require between intense workouts is a common question?

There is significant variability within populations and genetics play a key role. The body requires a certain amount of time to repair the damage incurred by intense workouts and understanding your genetic propensities can guide strategic planning to achieve the highest impact from your routine.

Probable beneficial rest interval after intense exertion:



#### INTERPRETATIVE NOTES:

Your genetics indicate that you may develop a fair amount of inflammation with intense workouts. Monitor your HRV and see how quickly it recovers after intense workouts.



# STRENGTH/HYPERTROPHY

## GENETIC DATA

GENE	GENO TYPE	
ACTN3	СТ	
ACE del	AG	
AGTR2		
IGF1R		
CNTF	GG	
DIO1	СТ	
IGF2(1)	СТ	
IGF2(2)	CG	
IGF2(3)	GT	
IL6	CG	
BMP2	CC	
SHBG	СТ	
CCL2(1)	AA	
CCL2(2)	AA	
CCR2	Π	
IL15Ra(1)	n/a	
IL15Ra(2)	AT	
IL15Ra(3)	AC	

Endomorph, mesomorph, or ectomorph; we are all aware that we possess certain genetic predispositions toward a specific body type. This genetic predisposition resides in the genetic variations that deal do with muscle strength and hypertrophy (muscle size).

It has been estimated across multiple genomic studies that >50% of muscle strength and muscle mass is attributable to heritable genetics.

We can investigate genetic variants that specifically code for hypertrophy and ones that code for strength development. There is no doubt that genetics will gift us with a specific proportion of muscle fiber types but through lifestyle approaches we have an ability to create different varying body type outcomes.

Strength

Hypertrophy



#### **INTERPRETATIVE NOTES:**

Excellent propensity to gain mass and strength when you can identify the training routine that optimizes that for you.



## POWER/SPRINT

## GENETIC DATA

POWER VS ENDURANCE	
GENE	GENO
	TYPE
AGTR2	
ACE del	AG

CT

ACTN3

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An individual's ability to produce short burst explosive power can be strongly influenced by their genetic code. Looking at the genetics for muscle type, energy production, metabolic capacity, and neuromuscular response, can provide insight into your potential. Response to training is another important marker.

Knowing the code will also provide guidance for the techniques to improve response. Genetics will provide the clues to the hardware but the epigenetics provides us with the ability to modify expression.

Your coach can utilize this information to assist you in enhancing the potential within your genetics.

Power/Sprint is one component of athletic performance and can provide guidance into your best mode of training.

Power/Sprint



#### INTERPRETATIVE NOTES:

ACTN3 indicates mixed. Genetically decent power and sprint capabilities.



## ENDURANCE

## GENETIC DATA ENDURANCE GENE GENO TYPE

BDKRB2	СТ
GNB3	СТ
ADRB2 (1)	GG
ADRB2 (2)	CC
ADRB2(3)	CC
PPARGC1A	CC
PPARD (1)	TT
PPARD (2)	AA
ADRA2A	GG
EPHX1	Π
LTBP4	GG
SCGB1A1	GG
UCP2	GG
VEGFA	n/a

Endurance genetics falls on the other end of the athletic spectrum from power & sprint. Genetically speaking, elite athletes will lean toward one end of this spectrum and will tend to excel in sports that rely on a particular genotype.

In the endurance genetics, consideration for muscle type, metabolic capacity, efficiency of the use of specific nutrients for fuel, energy turn-over, and cardiac response all play into optimal performance. Knowing which areas are strong and which could benefit from training is important in planning training routines.

Some athletes will be balanced in both power and endurance but elite athletes are usually shifted to one end of the spectrum.



#### INTERPRETATIVE NOTES:

Definite strength in the genes for endurance. VO2 max will still limit that component. D-ribose should help to boost this response.



## SPRINT/POWER VS. ENDURANCE

# GENETIC DATAPOWER/SPRINTGENEGENOGENEGENOIGF1R--DMDAAAGTAG

What is your genetic gift? Are you predisposed to better performance in sprint and power athletics or are you designed more for distance and endurance? Most people fall somewhere in the middle, referred to as a mixed athlete.

We look at wide ranging genetic factors that will contribute to these various outcomes but possessing a specific predisposition does not relegate you to staying within that spectrum.

Knowing your genetics can guide your training to suggest a focus on enhancing your strengths or training your less optimal propensities.

Overall



#### INTERPRETATIVE NOTES:

Mixed type training seems to fit your genetics best. Short burst high intensity.



# SOFT TISSUE

## GENETIC DATA

GENE	GENO TYPE
COL1A1	CC
COL3A1	GG
COL12A1	AA
COL5A1 (1)	
COL5A1 (2)	AC
GDF5	n/a
MMP3(1)	СТ
MMP3(2)	СТ
FAM46A	Π
CILP	GG
COL11A1	GG
CRP	GG
ADAM12	CC
ESRRB1	Π
FGFR1	СТ
SASH1	CC
SAP30BP	AA

Genetic predispositions toward connective tissue injuries is an important consideration whether you are an athlete or a weekend warrior. These genes look at specific tissue types and based on population studies can provide valuable information regarding the potential for injuries. Knowing your genetic predispositions can guide risk mitigation.



#### **INTERPRETATIVE NOTES:**

Excellent genetics for connective tissue. Most common injury risks are below normal for you except the risk of rotator cuff injury.