

Part 1: Using Genetics Responsibly in a Sports Context

A Case-Based Walkthrough

Purpose of this case study

Genetics is often discussed as if it offers direct instructions for personalisation. In reality, genetic information is biological background information: it describes tendencies in how the body may function under certain conditions. Whether those tendencies are relevant depends entirely on context.

The purpose of this case study is to:

- Explain the biological basis of how genes influence physiological processes
- Show how genetic variation may (or may not) influence nutrition-related decisions
- Demonstrate how genetics should be integrated after foundational assessment

This document is not intended to provide prescriptive advice. Its goal is to strengthen biological reasoning and judgement.



Essential molecular biology background

To use genetics responsibly, it is important to understand what genetic information actually represents.

2.1 DNA, genes and proteins

DNA is the molecule that carries genetic information. Genes are segments of DNA that provide instructions for making proteins. Proteins carry out most biological functions in the body, including:

- Enzyme activity
- Structural support (e.g. muscle and connective tissue)
- Transport and signalling

A genetic variant does not usually create or remove a protein entirely. More commonly, it leads to small differences in how efficiently a protein works, how much of it is produced, or how it responds to environmental inputs such as diet, training, or stress.

2.2 Genetic variation and SNPs

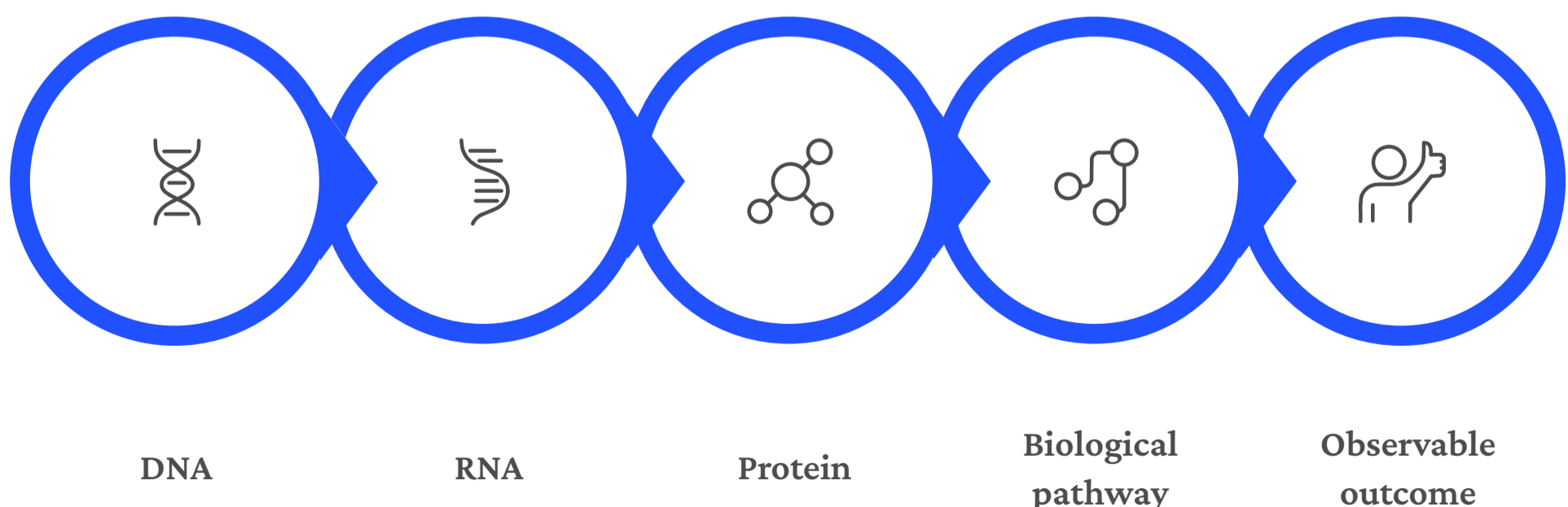
Most nutrigenetic information is based on single nucleotide polymorphisms (SNPs). These are single-letter changes in the DNA sequence.

Important points to understand:

- SNPs are common and normal
- Most SNPs have small effects
- Their influence depends on the surrounding biological and environmental context

2.3 From gene to function: why context matters

Genes influence physiology indirectly. The pathway typically looks like this:



At each step, environmental factors (diet, training, sleep, stress, medication) influence what actually happens. This is why genetic information should be interpreted as background potential, not as a direct instruction.

Case context

1. Case context

The purpose of the case context is not to analyse the sport itself, but to provide enough background to explore whether genetics adds interpretive value.

Illustrative profile:

- Male, 24 years old
- Team sport athlete (football winger)
- High weekly training load including multiple pitch sessions, strength training and competition

Broad goals:

- Improve repeat sprint capacity
- Recover more effectively between sessions and matches
- Reduce recurrence of soft-tissue injuries

Key challenges reported:

- Two hamstring strains within 18 months
- Muscle soreness lasting longer than 48 hours
- Subjective reports of "energy crashes" during matches

The key question is not what to do, but whether genetic background meaningfully changes how we interpret this situation.

2. Reframing the key question

A common but problematic question in applied genetics is: "What should we change based on DNA?" This assumes a direct causal pathway from gene to action. Biologically, this is rarely the case.

A more appropriate framing is:

- Which physiological processes are under strain?
- Where might genetic variation plausibly influence those processes?
- Would that influence meaningfully alter decisions?

This reframing aligns genetic interpretation with biological reality.

What must be considered before genetics

Before genetic information is considered, foundational physiological context must be understood.

Training load

Volume, intensity, frequency and recovery windows

Tissue stress and repair

Cumulative load on muscle and connective tissue

Energy availability

Alignment between intake and expenditure

Recovery inputs

Sleep quality, rest days, stress exposure

If these layers are not addressed, genetic interpretation becomes speculative.

Genetic information: scope and limits

In this case, genetic information was considered with focus on how variation might influence:

- Caffeine metabolism
- Oxidative stress handling
- Tissue repair and connective tissue characteristics

6.1 Caffeine metabolism

Variants influencing caffeine metabolism affect the activity of enzymes involved in caffeine breakdown. Biologically, this may influence:

- How long caffeine remains in the system unmetabolised
- Sensitivity to dose and timing

Training context, habitual intake, and timing remain relevant factors.

6.2 Recovery-related pathways

Some genetic variants are associated with antioxidant enzymes or inflammatory signalling. These may influence susceptibility:

Importantly:

- Increased susceptibility does not guarantee poor recovery
- Adequate recovery strategies can override genetic tendencies

6.3 Connective tissue considerations

Variants related to collagen structure may influence tissue properties over long timeframes. They do not predict injury, but may justify more cautious progression in certain contexts.

Case-specific genetic profile and interpretation

The following genotypes were reviewed as part of the interpretation process:

Gene	Variant	Genotype
CYP1A2	rs762551	C/C
SOD2	rs4880	A/G
COL5A1	rs12722	T/T
ACTN3	rs1815739	R/X

7.1. Interpretation logic: how genetics was used

Key questions included:

- Is there a plausible mechanistic link between the variant and the reported issue?
- Is the pathway currently under strain?
- Would this information change how carefully an intervention is implemented?

When genetics was not used

Equally important is recognising when genetics adds little value. In this case, genetics was not used to:

- Explain injuries where mechanical load was clearly dominant
- Attribute fatigue without evidence of recovery mismatch
- Replace established physiological assessment

From a biological standpoint, these outcomes are better explained by environmental load than by genetic variation.

Strategy employed: how genetics informed decisions

Genetic information was used to modify implementation strategy, not to introduce new interventions.

Caffeine strategy

Emphasis was placed on:

- More conservative dosing
- Earlier timing relative to match play
- Avoidance of repeated dosing late in matches

Rationale: The CYP1A2 C/C genotype increases the likelihood that poorly timed caffeine could exacerbate perceived "energy crashes" and interfere with recovery.

Recovery strategy

Recovery support was prioritised due to:

- High training and match load
- Recurrent soreness
- SOD2 A/G genotype increasing susceptibility under stress

Genetics reinforced the importance of consistency and adequacy, particularly during congested weeks.

Injury-related considerations

COL5A1 genotype did not trigger injury-specific interventions.

Instead, it supported:

- Conservative progression after injury
- Greater attention to cumulative load rather than acute spikes

Genetics here functioned as a long-term lens.

1. Communicating genetic insights responsibly

Genetic information should be communicated as probabilistic and context-dependent. Good communication includes:

- Emphasising interaction with environment
- Avoiding absolute or predictive language

This reduces misinterpretation and supports informed decision-making.

2. Ethical and professional boundaries

Biological data carries perceived authority. This makes ethical boundaries essential. These include:

- Avoiding prediction of performance or injury
- Avoiding diagnostic language
- Remaining within professional scope

Genetics should inform understanding, not create false certainty.

3. Reflection prompts

- Which physiological layers dominate this case?
- Where does genetic variation plausibly matter, and where does it not?
- How would you explain these limits clearly?

Part 2: Common Mistakes in Sports Nutrigenomics

Sport environments place high demands on biological systems and create pressure for optimisation. Genetics here can add an important layer of information if used correctly.

Mistake 1: Treating genetic variants as predictors

Most genetic associations describe statistical relationships, not causal certainty.

Biologically:

- Many variants influence protein efficiency, not presence
- Effects are small relative to training and nutrition
- Outcomes emerge from multiple interacting pathways

Prediction based on single variants is therefore unjustified.

Mistake 2: Using genetics before fundamentals

Environmental inputs regulate gene expression and protein activity. When recovery, sleep, or energy availability are inadequate, genetic differences become largely irrelevant.

Addressing fundamentals should always precede genetic interpretation.

Mistake 3: Confusing nutrition advice with diagnosis

Genetic data used in nutrigenetics is not diagnostic.

Biologically, most variants:

- Do not indicate disease
- Require clinical context for interpretation

Blurring this line risks ethical and professional issues.

Mistake 4: Assuming a genetic variant is actionable without a clear biological link

A genetic variant is only actionable if it sits within a biological pathway that can be influenced by a nutrient or lifestyle factor. If there is no plausible pathway linking the gene to something you can change, the result may be interesting but not actionable.

In practice, a genetic variant is actionable when:

- The gene codes for, or regulates, a protein involved in the pathway of interest
- There is a clear interaction with a nutrient or lifestyle factor
- Changing the nutrient or lifestyle factor could plausibly alter the outcome

This does not mean guaranteeing an effect, but that the biology supports a reasonable expectation of influence.

About Dr. Leta Pilic

I'm a PhD-qualified nutrition scientist and nutrigenetics educator with over a decade of experience in research, academia and applied practice. I believe genetic testing should be useful, not overwhelming, and ethical, not salesy. My goal is to help you unlock meaningful, personalised health strategies using tools that are evidence-based and responsibly delivered.

So what makes me qualified to help you?

- PhD in nutrition and genetics
- Registered Nutritionist with the Association for Nutrition (the only UK Voluntary Register of Nutritionists recognised by Public Health England and the NHS)
- Actively educating future personalised nutrition professionals
- Actively researching in nutrition and genetics, making sure I am up to date and contributing to the latest evidence
- Deep knowledge of laboratory procedures that is essential to understand how your DNA is analysed

Connect with me on [LinkedIn](#) for professional updates and insights.