# GENETIC PRINCIPLES -AN OVERVIEW

Our bodies are complex entities comprised of sophisticated and intricate systems. At any given moment, we make decisions that can impact these systems, our genes and our overarching health to our benefit or our detriment. The choices that are best suited for each of us is dependent on our unique genetic code. If we hope to understand ourselves, our clients and our respective genes, we first need to grasp the fundamental principles which make up the exciting and lifechanging field of genomics.

These include:

- The Upstream approach
- Genetic variation
- Penetrance
- Nutritional genomics
- · Pathway analysis
- Epigenetics
- and our proprietary formula, G+E=H<sup>™</sup>.



Upstream

Positively influencing a client's long-term health benefits means practitioners need to start looking at the biochemical individuality of each human being, based on the uniqueness of their genetics and environment.

When it comes to healthcare, there's the concept of upstream and downstream approaches. Traditional approaches have predominantly involved looking at a client and attempting to fix their symptoms or their disease – what we call downstream. This approach isn't always sustainable or successful, and the strategy of merely patching people up can be so far downstream that we might as well be out to sea. Instead of treating symptoms with hastily prescribed medications or more extreme symptomatic treatments, what we're interested in is identifying the cause of the symptoms. It's the WHY? Treating the 'why' can help practitioners address and manage the root cause of their clients' problems and potentially prevent similar or new conditions from occurring in the future.

What we often fail to take into consideration and what we need to start realizing is that every client has a story, a history – where they live and work, what they eat, how they eat, their exercise regime and of course their unique genetic blueprint. Gaining an unobstructed view of a client requires identifying and understanding these complex behaviours as well as their genes which brings us to the next principle, genetic variation.



### **Genetic Variation**

Genetic variation is the difference in our DNA; it's the ever so small but impactful 0.1% difference that makes us unique. From the colour of our hair, the shape of our faces, the way we manage stress and even how we react to food and exercise– it all comes down to our unique genetic blueprint or code.

#### Let's break that down

The letters in our DNA code for amino acids, and when we put together these amino acids, they form a protein. These proteins are hormones, enzymes or neurotransmitters (a few examples) – all the vital elements we need to function. If we have genetic variations, then some of these changes in our DNA code are going to change our amino acids and ultimately change our protein function.

Now, not every single section in our DNA that's different will change a protein. What we are interested in are the differences that do alter the enzyme or hormone. We want to understand how that change affects how we respond to our environment. What choices are we making in terms of diet and lifestyle and are these even the right choices based on who we are at an individual level...at that 0.1% level.

The ways a genetic variation can appear in our DNA code include substitution, deletion, insertion and copy numbers - the different ways variations occur.

But what does this all mean and why should we care? Well, the purpose of understanding our genes and specific genetic variations is to gather as much information as possible. We need to understand how specific changes impact amino acids, proteins, enzymes, hormones and our overall function at an individual level. Having this understanding can help guide us in terms of what are the necessary changes we need to make daily to lead healthy, long lives. Now, something you might have heard or read about in literature and the media centres around genetic mutations. In our field, we prefer to steer away from the term mutations. A mutation is the same as a genetic variation... a spelling change in our code. However, sometimes the spelling change can be can have a far greater impact.

When the field of medical genetics was born, a severely significant genetic variation was labelled as a mutation. If we take into consideration the concept of high and low penetrance, which we will get to shortly, a change in a high penetrance gene -considered a massively important spelling change -, has the potential to cause a disease or get pretty close to causing it.

In the medical literature, it was the high penetrance changes that were referred to as mutations because they were powerful and easy to see their effect on an individual's health. Understandably, such a significant and negative change would be termed mutation and have a negative connotation. But in the arena of nutrigenomics, we are not dealing with human genetic diseases, where one gene variant or one SNP may equal a disease. In our field, by assigning a value judgement and using a word like mutation, we're prejudging what might be wrong with a client. Remember, genetics is informative, and it's insightful, but it's never judgmental. So you won't catch us using the word mutation, it's just not helpful!



Genetic variations and their potential impact on our biochemistry, and how these changes could have a significant or less significant effect on us and who we are, brings us to penetrance.



### Penetrance

Penetrance refers to how likely a person is to present a specific physical trait based on their genetic blueprint.

Just like we've discussed, previously, when the probability is so high, we talk about it being highly penetrant. High penetrance relates to genetic changes that are powerful and impactful with an increased chance of impacting our biochemistry. In high penetrance, our genes play a significant role and epigenetics, the daily choices we make, have a lesser role.

The terms "high' and 'low' are used when explaining the likelihood that an individual might develop a specific trait dependent on their particular genetic results. For example, an individual who carries a gene variant that is associated with breast cancer and has an 80% penetrance (high), would most likely want to seek regular breast exams and screening, and potentially genetic counselling. However, if the gene variant penetrance is only 1%, the individual's response might be quite different.

Low penetrance is the other side of the coin, where epigenetics and the daily decisions we make have a far more significant impact on our health and the individual gene variants, by themselves, play a lesser role. This doesn't mean we ignore our genes, but rather focus on the right daily choices we make in relation to our unique genetic profile.

A further critical component we need to focus on is how we might increase the power and ability of these low penetrance genes. To achieve this, we need to bring them together and group them into what we call biochemical pathways.

#### **High Penetrance**



#### Low Penetrance





#### Pathways

Biochemical pathway analysis involves taking a series of low penetrance genes and grouping them - based on biochemical or metabolic functiontogether they form a stronger and more precise picture of an individual's genetic susceptibility.

In our field, we focus on low penetrance genes and want to avoid making recommendations based on a single low penetrance gene variant, because individually, they are not very robust. But when grouped into a biochemical pathway, they become more powerful and informative. Think of this as a cumulative effect - It all adds up.

Once we have this, we can make a client recommendation based on the total biochemical pathways that have been impacted. Focusing on a client's core pathways helps us get an unobstructed view of what's happening upstream. It helps build a more vibrant and far more contextual picture of the whole person, and instead of just addressing symptoms, we go to the root of the problem.



# **Epigenetics**

Getting this clear image means considering an individual's environment, their epigenetics. The term 'epigenetics' means 'above the genes' and has become an increasingly exciting area for us as it provides insight into how our way of life and our lived experiences can impact our genetic destiny. Epigenetics tells us how specific elements in our environment and our lifestyle can contribute to the way our genes are "expressed" or behave. All of these changes add up and subsequently have the potential to impact our health, our disease risk and overall well-being. Our daily choices - what we choose to consume, how we react to stress, and what we expose ourselves to have the potential to determine whether specific genes get expressed or not. Our genes and our epigenetics make us who we are today, and who we might be tomorrow and in the future.





### **Nutritional Genomics**

There are plenty of one-size-fits-all, quick-fix approaches to managing our health and diet. However, the missing component in all of these methods involves considering the individual. We believe that an individual's health is not static and one of the ways we can positively impact how genes are expressed (and ultimately our health) is by looking at the interaction between our diet, our lifestyle and our genes. This interaction can be broken up into two fields of study, namely nutrigenetics and nutrigenomics.

Nutrigenetics looks at how we respond to nutrients based on our genetic profile. Nutrigenomics

explores how foods and nutrients change the way our genes express themselves.

Working with both Nutrigenetics and Nutrigenomics allows us to develop plans for personalized, targeted and appropriate dietary recommendations.

Research suggests that our unique genetic makeup influences the outcome of our health when taking our diet, environment and our lifestyle into consideration. Our daily choices and a personalized nutrition approach is a viable and sustainable way to reduce our risk of disease and keep our health in check.







#### G+ E = H™

Society today is experiencing a sharp increase in the number of people who suffer from complex and chronic diseases, from diabetes and heart disease to autoimmune disorders. Unfortunately, the traditional approach to addressing these conditions through mass—made quick fixes have failed to take into account the individual in their entirety.

The areas of health, wellness, and how we understand ourselves is rapidly moving towards a field where understanding our genetic profile, in combination with our environment and taking responsibility for specific behaviour change, is critical. Which brings us to our final principle...the  $G+E = H^{TM}$  equation.

While our genes (G) give us valuable insight into our health, we don't live in a vacuum. At any one moment in our life, we're inundated by multiple elements that can affect our health and change our gene expression. In the same way, only looking at epigenetic (E) factors (our daily choices and exposures) without taking our genetic profile into account often leads to a standard approach to healthcare that only really works for a small percentage of the population.

It's not possible to make informed health decisions that have longevity and impact if we take an either-or approach to diet and lifestyle versus our unique genetic profile. When we know our G and our E, we can make the right decisions and choices which positively impact our H (health).



## **Principles in Action**

Each of us is unique and complex, which means understanding our genetic blueprint and how our daily choices influence us is necessary to lead happier and longer lives. Thanks to advances in the field of genomics, we're able to make impactful and practical recommendations and promote personalized approaches to health and wellness.

Understanding each of these principles, as well as how they work together, allows us to gain valuable insight into our clients' at an individual level. Insight and understanding of each principle accelerates our potential to make a positive impact on their health, assist them in making the best daily choices and most importantly, helping them achieve their greatest human potential!



