Commentary Article

iMedPub Journals
www.imedpub.com

Journal of Translational Neurosciences
ISSN 2573-5349

2021
Vol.6 No.5:9830

Disease Has a Genetic Foundation

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Received date: July 30, 2021; **Accepted date:** October13, 2021**; Published date:** October25, 2021

Citation: Gerhard H.W. May (2021) Disease Has a Genetic Foundation, J Transl Neurosci Vol. 6 No: 5.

Abstract

Genetics plays an influence in all diseases, to a greater or lesser level. Disease processes are influenced by variations in our DNA and variances in how that DNA operates (alone or in combination), as well as the environment (which includes our lifestyle). This overview looks at the genetic foundation of human disease, including single gene disorders, chromosomal abnormalities, epigenetics, cancer, and complex disorders, and how current knowledge and technology developments can be used to provide patients with proper diagnosis, management, and therapy.

Keywords: cancer; genetics; genomics; molecular basis of health and disease

Introduction

Most people think of rare, single-gene illnesses like cystic fibrosis (CF), phenylketonuria, or haemophilia, or cancers with an obvious heritable component when they discuss the genetic basis of disease (for example, inherited predisposition to breast cancer). Despite the fact that genetic illnesses are individually rare, they account for roughly 80% of rare disorders. The sheer quantity of uncommon ailments means that one out of every seventeen people is affected by one. Furthermore, due to the plethora of variances in our DNA, our genetic composition plays a role in all disease processes, including common illnesses, to a greater or lesser level. Some of these variations, alone or in combination, may make an individual more vulnerable to one condition (for example, a kind of cancer), while making the same individual less susceptible to another disorder (for example, a type of cancer) (for example, diabetes).

Many illnesses are influenced by the environment (including lifestyle) (for example, nutrition and exercise in connection to diabetes), yet our cellular and physical reactions to the environment may change depending on our DNA. The immune system's genetics, which varies greatly across the population, impacts our response to pathogen infection. Furthermore, most cancers are the consequence of a series of genetic alterations that occur over a person's lifespan and are influenced by environmental variables. Clearly, understanding genetics and the genome as a whole, as well as its variation in the human population, is critical to comprehending disease processes, and this knowledge is the foundation for curative medicines, helpful treatments, and prevention measures.

With so many genetic illnesses, it's impossible to give more than a few examples to demonstrate the principles in this paper. There are a lot of searchable internet resources that provide a plethora of accurate material for further information on certain ailments.

Variation in the human genome

The human genome and the reference sequence for the human genome:

It was recognised from the start of the Human Genome Project that there was a great deal of DNA sequence diversity among healthy people, and that there is no such thing as a "typical" human DNA sequence. However, if we're going to talk about DNA sequence changes, we need to talk about them in terms of a baseline, which is the human reference genome sequence.

Mutation vs. variation

Alleles are two (or more) different variants of a DNA sequence that occur in the population; each allele indicates a different version (or variant) of that sequence. We can compute the frequency at which a given variant appears in the population by analysing several human genomes, which is known as the "minor allele frequency" or MAF. A variant is deemed a polymorphism when the MAF is at least 1%, albeit this is a somewhat arbitrary cut-off.

Variants of a single nucleotide

Single nucleotide variations (SNV) or single nucleotide polymorphisms (SNP) are the most common variants in human genome, which impact only one base pair (bp) and are referred to as single nucleotide variants (SNV) or single nucleotide polymorphisms (SNP) depending on the MAF. The human genome is thought to include at least 11 million single nucleotide polymorphisms (SNPs) (averaging approximately 1 per 300 bp). It also appears plausible that if we sequenced the genomes of everyone on the world, we would find at least one people with an SNV for most locations in our genome, where such variation is compatible with life.

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