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Medical Genetics: It's Significance in Studying of a Disease

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Abstract

All diseases, to some extent, are influenced by genetics. Disease processes are influenced by variations in our DNA and variances in how that DNA operates (individually or in combination), as well as by the environment (which includes lifestyle). In this review, the genetic basis of human disease including single gene disorders, chromosomal abnormalities, epigenetics, cancer, and complex disorders is examined. It also considers how scientific knowledge and technological advancements can be used to provide patients with the best possible diagnosis, treatment, and care.

Keywords: Epigenetics, Cancer, DNA, Disorders, Human disease.

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Introduction

The majority of people may think of uncommon, single-gene illnesses like cystic fibrosis (CF), phenylketonuria, or haemophilia, or even malignancies with a definite heritable component when they examine the genetic foundation of disease (for example, inherited predisposition to breast cancer). However, even though genetic illnesses are uncommon on their own, they make up around 80% of all rare disorders, of which there are thousands. Due to the sheer quantity of uncommon ailments, 1 in 17 people worldwide are thought to be impacted by them. And since our DNA contains so many variations, our genetic make-up affects every disease process, even common illnesses, to a greater or lesser level. Some of these variations may increase an individual's susceptibility to one condition (such a certain form of cancer), either alone or in combination, while decreasing that individual's risk of developing an unrelated disorder (for example, diabetes) [1]. Despite the fact that our physiological and physical reactions to the environment might vary depending on our DNA, the environment (including lifestyle factors like nutrition and exercise in connection to diseases like diabetes) is a major factor in many disorders. Our immune system's genetic makeup, which varies greatly across people.

The majority of human DNA, also known as the nuclear DNA or nuclear genome, is found in the chromosomes of the nucleus, but there is also a minor quantity of DNA in the mitochondria (the mtDNA or mitochondrial genome). Most people have 23 pairs of chromosomes, thus the majority of our DNA is present in two copies, one from each of our parents, one from each of our mothers.

A geneticist defines a mutation as "any heritable alteration to the DNA sequence," where "heritable" includes both germline inheritance and somatic cell division (the proliferation of cells in tissues) (from parent to child). Although these DNA alterations can result in obvious variations in the person (the "phenotype"), they might also have no effects. As a result, such changes in the human population were formerly referred to as "mutations," especially when they were linked to a disease condition. However, this language conjures up images of the "mutants" in science fiction and zombie movies for many people, who also associate it with negative connotations. Therefore, it is now standard practise to refer to deviations from the reference sequence as "variants," especially in the context of medical genetics within a health care [2].

There are growing numbers of human DNA variations found for which are still unsure of the effect; these are referred to as "variants of unclear significance" or VUS. Variants can also be categorised as benign (not related with disease) or pathogenic (associated with disease).

It was made possible to use minisatellites in forensic analyses and paternity testing to create distinctive patterns (similar to store barcodes) for each individual, a process known as "DNA fingerprinting" because they are highly variable within the population but inherited steadily from parent to child. Micrograms of DNA were needed for this method, which also required the use of radioactive labels and was time-consuming (usually taking 1-2 weeks). When microsatellites were initially discovered around

the end of the 1980s, the older DNA fingerprinting method was swiftly supplanted by 'DNA profiling' utilising microsatellites, which required just a little amount of sample DNA-roughly 1 nanogram.

The majority of the genetic variations in our genome came from one of our parents. However, human DNA is continually exposed to chemicals that might damage it, and there is also a chance for mistakes every time a cell's DNA is reproduced before it divides. In trios (child and both parents), genomic sequencing has shown that each person possesses, on average, 74 de novo SNVs that were not present in either parent, as well as around three de novo insertions/deletions. A de novo CNV larger than 100 kb in size will occur in 1-2% of kids. With a gain or loss of a repeat unit happening in about 1 out of every 1000 microsatellites per gamete every generation, microsatellites have a rather high mutation frequency [3].

The term "karyotype" refers to a species' typical chromosomal complement, which includes chromosome size, number, and shape. The "typical" human karyotype, as defined by the ISCN, is either 46, XX (female) or 46, XY (male). Human chromosomes are made up of DNA that is encased in chromatin, which is a core of histone proteins. In a cell's nucleus, chromatin is typically dispersed, but the chromosomes condense during the metaphase of the cell division cycle [4]. These condensed chromosomes may be dyed using a number of chemicals, and the distinctive banding patterns can then be seen by looking at them under a light microscope. The bands represent chromatin areas with various properties and, consequently, various functional components.

Polyploidy is a condition in which a cell has at least 69 chromosomes and more than two full sets of the human haploid genome. One to three percent of pregnancies result in triploidy (three haploid sets of chromosomes), which is often caused by the fertilisation of a single egg by two sperm or, less frequently, by the fertilisation of a diploid gamete (egg or sperm). While tetraploidy (four haploid sets of chromosomes) is much less common and not compatible with life, triploidy (three sets of chromosomes) is often very poor viability and results in early spontaneous abortion during pregnancy. Aneuploidy, on the other hand, is a condition in which the number of chromosomes is not a precise multiple of the number of haploid chromosomes [5].

Conclusion

The field of genetics in medicine has seen a significant transformation in recent decades, transitioning from a specialty treating ailments that were relatively uncommon in the population to a science that supports advancements in broader patient care. Genetics will be important to every member of the population, a better knowledge of the contributions of genetic predisposition and epigenetic modifications to prevalent diseases, as well as the function of genetic alterations in response to therapy. Education will be crucial in ensuring that new breakthroughs in genetics and genomics are adequately translated and put into clinical practise, both for health professionals and their patients. Clinical scientists, genetic technologists, and bioinformaticians will be in greater demand to provide laboratory services, as well as physicians, genetic counsellors, and nurses knowledgeable in the newest genetic and genomic technology.

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