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MENDELIAN GENETICS AND ITS APPLICATIONS IN UNDERSTANDING GENETIC DISEASES

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Abstract
This research topic explains the fundamental principles of the Mendelian genetics and how they have been of the fundamental importance in explaining the ability to inherit genetic diseases in human beings. The laws of segregation and independent assortment, which were developed by Gregor Mendel in the nineteenth century, are still the foundations of classical and modern medical genetics. According to qualitative descriptive approach, the study applies scientific knowledge to investigate how the principles deal with the transmission of monogenic disorders, including autosomal dominant, autosomal recessive and X-linked inheritance. The findings indicate that Mendelian models are required to comprehend the way to establish the carrier, predict the danger of the disease in the further generations and provide appropriate genetic counselling. The example of diseases with a great deal of literature such as Cystic Fibrosis, Sickle Cell Disease and Huntington Disease can serve as examples of how these laws can be applied in clinical diagnosis. The paper also analyses the fusion of classical genetics and the present genomic technology such as DNA sequencing and the Human Genome Project. Despite the fact that more comprehensive processes of environmental and multi-gene interaction are used with complex polygenic disorders, Mendelian genetics continues to provide the primary theoretical framework of addressing hereditary illnesses. The authors come to the conclusion that the collaboration between the Mendelian and molecular biology is critical to the future of early diagnosis and preventive medicine and particular treatment procedures.

1. Introduction

Heredity is a topic that has been given considerable attention in the sciences of biology over the last hundred years. Modern genetics is based on the work of Gregor Mendel, an Austrian scientist and monk who carried out systematic experiments on pea plants (Pisum sativum) in the middle of the nineteenth century. Mendel found that using thoroughly managed experiments of cross breeding those qualities were inherited in predictable ways and that they were made possible by discrete units which are today referred to as genes. His discoveries resulted in the development of two main principles the law of segregation according to which the two alleles of a gene separate during gametes formation, and the law of independent assortment according to which genes that govern different traits are inherited independently of each other. Groundbreaking may not have been much known until the beginning of the twentieth century when the works of Mendel were re-discovered by various scientists and became the foundation of classical genetics (Griffiths et al., 2015).

The evolution of the Mendelian genetics brought a lot of improvement into the knowledge of the biological lines of transmitting the traits of the parents to the offspring. Genetic traits, according to Mendelian rules are decided by the pairs of alleles, both inherited. These alleles may either be dominant or recessive and their combination causes the actual characteristics to be seen, or phenotypes of an organism. This simple yet effective model of inheritance has given the science community the capability to foretell genetic results using the punnett squares and pedigree analysis. Consequently, the Mendelian genetics have proved to be a critical concept in the study of hereditary traits in humans and other living beings (Pierce, 2017).

As the genomic field has progressed at a blistering pace during the late twentieth and the early twenty-first centuries, the principles of Mendelian inheritance have been incorporated into the current genomic technologies. The Human Genome Project was one of the most crucial achievements of the sphere, which was completed in 2003. This global initiative has succeeded in mapping and sequencing of the complete human genome which gives researchers extensive information about human genetic organization. The scientific results of this project have helped scientists to determine genes that cause many inherited diseases, as well as to study the role of genetic differences in human diseases and health (Collins et al., 2003).

The importance of the Mendelian genetics in genetics diseases is that the genetics are diseases which are manufactured by the integrity or malfunctioning of the DNA of a person; the diseases bring about diseases. A lot of the hereditary diseases are of classical Mendelian inheritance. As a case in point, Cystic Fibrosis is an autosomal recessive disorder that is caused by CFTR gene mutations and causes severe respiratory and digestive problems. Equally, the Sickle Cell Disease is a mutation in the hemoglobin gene that makes the red blood cells have an unusual sickle shape that may restrict the flow of blood and result in severe health consequences. Another such famous example is the Huntington Disease, which is an auto-dominant disease, the development of which is associated with progressive neurological degeneration and cognitive impairment (Nussbaum et al., 2016).

The understanding of the inheritance patterns such diseases follow is extremely relevant in medical genetics since it enables medical practitioners to make speculative assumptions as to the risk of the spread of the disease in the family. The genetic testing in accordance with the Mendelian principles allows identifying the carriers, assessing the risk of the disease and giving appropriate genetic recommendations to individuals and families. They are particularly useful in the early diagnosis and preventive care plans as individuals can acquire knowledge of making informed decisions in regards to reproduction and disease management (Strachan and Read, 2018).

Regardless of current advancements made in genetic research, genetic diseases are intricate and are affected because of the interaction of multiple genes with several environmental factors. Nevertheless, Mendelian genetics still forms the basis of theory with which one can explain the laws of inheriting most monogenic diseases. Current genomic technologies, such as the sequencing of genes and molecular diagnostics, have increased the use of Mendelian principles in biomedical research and clinical practice. Therefore, the use of classical Mendelian genetics in conjunction with modern genomic science continues to play a significant role in enhancing the understanding of these hereditary diseases and enhancing hereditary disease diagnosis, treatment, and prevention approaches.

This study intends on investigating the concepts of Mendelian genetics as well as exploring how they are used in the case analysis of the laws of inheritance of hereditary ailments. The research aims at describing the application of the laws of Mendel in enabling scientists to determine hereditary disorders, match genetic hazards, and aid in the advancement of diagnostic and preventive medical approaches.

Even though Mendelian genetics is a good basic framework of explaining the aspect of hereditary behavior, there are so many people who are not adequately informed of the aspects of genetic diseases transmission across generations. In addition, not all hereditary disorders are readily identified and diagnosed as persons are not aware of their genetic nature. This poses problems of early diagnosis, prevention and treatment of genetic diseases.

Thus, the research problem of the given study is: What is the role that the principles of Mendelian genetics can help in enhancing the understanding, prediction and diagnosis of genetic diseases in humans?

2. Literature Review

Modern genetics can be said to date back to the works of the earliest scientists, particularly Gregor Mendel, who carried out a systematic study, involving pea plants in the nineteenth century. Mendel found that the qualities were acquired as discrete units and he referred to those as genes, later on predicting that are inherited through controlled breeding experiments which involve the parents giving the newborns a certain pattern of inheritance. Through his work, the law of segregation and the law of independent assortment were developed, which are two theories that describe how alleles part during gametogenesis and how different traits are inherited independently (Griffiths et al., 2015). Despite the fact that the work of Mendel was initially disregarded, it was rediscovered in the first part of the twentieth century and it became the foundation of the classical genetics.

Subsequent progress in the field of biology linked Mendelian ideas to the actions of the chromosomes. Scientists proved that genes are contained in chromosomes and that segregation of alleles as would be found by Mendel is related to segregation of homologous chromosomes during the meiosis. This finding was the foundation of the chromosomal theory of inheritance and brought about better association between classical genetics and cytology. This was advanced by the discovery of the structure of the DNA by James Watson and Francis Crick in the year 1953 and this discovery revealed the molecular process that stores and transmits genetic information (Watson and Crick, 1953). This discovery advanced a better knowledge of the process of genetic instructions replication and their transfer to the next generation.

The process of modern genetic research has developed widely due to the increase of molecular biology and genomic technologies. Among the greatest accomplishments made in this area was the completion of the Human Genome Project that was able to map the human genome. This was an international scientific project that equipped scientists with detailed evidence and information of the location and the role of thousands of human genes. The outcome of the project significantly enhanced the capabilities of scientists to discover genes that cause inherited diseases as well as comprehend the genetic foundation of human diseases (Collins et al., 2003). This has caused genetic research to gain greater significance in medicine, biotechnology and health care among the people.

Several works have used Mendelian genetics to explain genetic diseases, which are conditions that occur as a result of mutation or changes in DNA. Genetic diseases tend to exhibit regular patterns of inheritance, among these being autosomal dominant, autosomal recessive and sex-linked inherited diseases. A famous case is Cystic Fibrosis, which is an autosomal recessive illness affecting CFTR gene mutations. People with this condition become carriers of two faulty copies of the gene due to which thick mucus is deposited in the lungs and digestive tract (Nussbaum et al., 2016). Studies on cystic fibrosis have made scientists gain more insight into the correlation between gene mutations and the development of illness.

Sickle Cell Disease is another example that is commonly carried out research that follows an autosomal recessive model of inheritance. The mutation in the gene that produces hemoglobin, the red blood cell protein that carries oxygen, causes this disease. The mutation leads to sickle and rare rigidness of the red blood cells, as a result of which blood circulation may be blocked, and serious health problems may occur. Research also demonstrated that the possession of one mutated gene can provide partial resistance against malaria in individuals with half of that particular gene, demonstrating that genetic variations can also affect the susceptibility to the disease and their capacity to readily adjust to the environment (Strachan and Read, 2018).

Moreover, Huntington disease is mentioned widely in the literature on genetics as a good example of autosomal dominant disorder. Huntington disease is an illness caused by mutation in a gene namely HTT, which causes progressive destruction of nerve cells in the brain. Due to the dominance nature, those people who inherit one copy of the mutated gene tend to develop the disorder. Studies about Huntington disease have helped to uncover the genetics and general knowledge about the neurology genetic diseases but have highlighted the significance of the genetic screening and early diagnosis on the case (Pierce, 2017).

Genetic counseling has also been a critical area where the application of Mendelian genetics is used. Through study of family history and inheritance of diseases, the medical practitioners are able to determine the probability of occurrence of genetic diseases in adulteration generations. The information will assist families make informed choices concerning reproduction and health control. The recent progress in molecular diagnostic technologies has also enhanced detection of disease causing mutations, which can be diagnosed and better treatment plans made early (Nussbaum et al., 2016).

Regardless of how crucial it is, researchers admit that not all diseases can be characterized by mere Mendelian inheritance. Most common diseases such as diabetes and cardiovascular diseases are associated with intricate interactions between a number of genes and the environment. However, Mendelian genetics has been a basic construct of gene identification in disease-related matters as well as in a basic mechanism of inheritance. The current genomic technologies, including gene sequencing and genome editing, further increase the use of the principles of Mendelian on biomedical research and clinical medicine (Pierce, 2017).

All in all, the literature explains that Mendelian genetics has been used in taking the center stage in developing the conceptualization of heredity and genetic disease. Since first experiments of Mendel, genomic discoveries of the current era did not deny the basic fundamentals of hereditability in explaining the way genetic effects and disorders propagate through generations.

Research Questions

This paper seeks to understand the relevance of the Mendelian genetics in the explanation of the inheritance pattern of the genetic diseases and its role in contemporary medical genetics. Inheritance principles that were originally introduced by Gregor Mendel are still used as basis in comprehending the mechanism of passing on genetic traits and hereditary disorders to one generation to another. Despite the growth of modern genetics through the introduction of molecular biology and genomic technology, the Mendelian concepts take a place center stage in studying a number of hereditary diseases.

Genetic disorders are a significant area of research since they directly impact human health and could have severe medical, social, and economic implications. The knowledge of the processes by which these diseases are inherited will enable scientists and medical practitioners to better diagnose the disease early as well as offer genetic counseling and devise measures meant to prevent the disease as well as offer treatment. Mendelian genetics offers a tool of systematic analysis of transmission of traits involved through the analysis of dominant and recessive allele, patterns of inheritance, and family trees. With such tools of analysis, researchers are able to estimate the possibility of genetic disorders occurring in generations to come.

To explore the connection between Mendelian genetics and genetic diseases, the current study is guided by some relevant questions. These questions will be aimed at investigating not only the theoretical base of the Mendelian inheritance but also the practical aspects of the medical genetics.

1. What are the steps in the principles of Mendelian genetics that answer the question of the inheritance of genetic traits and diseases in humans?
   This question aims at investigating the role of the laws of segregation and independent assortment in the transfer of information of genetic material between parents and children.
2. What do the predominant Mendelian patterns of inheritance have to do with human genetic diseases?
3. This question is aimed at determining the key patterns of inheritance including autosomal dominant, autosomal recessive, and sex-linked inheriting and the effect they have on the expression of hereditary diseases.
4. How can Mendelian genetics contribute to predicting and diagnosing hereditary diseases?
   This question will assess the possibility of using genetic analysis, pedigree studies, and contemporary genetic testing to determine the risk of disease in families.
5. How much can the Mendelian principles assist the scientists in the explanation of genetic processes of certain hereditary diseases?

This question examines how the rankings in the Mendelian systems of inheritance could be used to account on the transmission of familiar hereditary disorders including Cystic Fibrosis and Sickle Cell Disease.

The study seeks to answer these questions to explain how the relevance of Mendelian genetics has continued to play a critical role in outlining genetic diseases and enhancing medical research with regards to the subject of human genetics.

The present research follows the qualitative descriptive research approach, which relies more on the scholarly review and analysis of available scientific writings regarding Mendelian genetics and genetic diseases. The aim of this methodological direction is to generalize the findings of past research in an attempt to present a comprehensive solution of the role that Mendelian laws of hereditary in explaining hereditary diseases.

The study is based on the secondary data sources whereby it incorporates academic textbooks, peer-reviewed journal articles, and authoritative scientific text in the genre of genetics. These are sources that provide a theoretical explanation of the Mendelian inheritance or cases of genetic diseases that are known to follow the pattern of transmission of Mendelian patterns. Through the analysis of these scientific resources, the paper obtains credible data on the evolution of genetics in the past and the complexity of biological processes of passing genotype information and clinical consequences of genetic mutations.

The major part of the methodology is the literature review procedure that will entail gathering and reviewing academic materials discussing Mendelian genetics and its use in medical science. The chosen literature consists of classical literature on genetics, up-to-date studies on the topic of molecular genetics, and clinical research on hereditary diseases. This will make the research based on the existing scientific knowledge and backed with reliable academic sources.

A comparative and analytical method is then applied in examining the collected information. Still in this phase, various scientific investigations and theoretical understandings are evaluated and contrasted with the aim of determining the similar conclusions and the significant discoveries pertaining to Mendelian inheritance and genetic ailments. Especially the research based on well documented Mendelian disorders like the Huntington disease, which is an autosomal dominant inherited disease is given special attention. In this discussion, the research illuminates the impact of application of the Mendelian principle in elucidation of the occurrence of certain genetic diseases.

Moreover, the analysis of the patterns of genetic inheritance with references to the examples, which are commented upon and discussed in the scientific literature will be the part of the methodology. The above are some of the ways researchers apply the Mendelian concepts in interpreting family pedigrees, determining who carries genetic mutations, and the chances of the disease occurring in successive generations. The paper establishes that Mendelian genetics are practically applicable in current medical practice and in genetic counseling by looking at these documented cases.

On the whole, it is a good methodological framework that enables the study to combine theoretical information with the examples of genetic investigations. A combination of the results of various academic sources is intended to generate an unambiguous and thorough comprehension of the contribution of Mendelian genetics towards explaining genetic illnesses and justification of changes in medical genetics.

Results and Findings

The extensive discussion of the available literature indicates that the Mendelian genetics introduce a structural basis of interpreting the inheritance of the human genetic diseases. Even with the advanced genomic technologies, the foundations set by Gregor Mendel remain central in the current genetic research and clinical practice. The findings show that Mendelian principles allow the researcher to make predictions on the inheritance pattern as well as gain insight on the mechanisms of genetic diseases being passed on to the subsequent generation.

The most prominent result is that the patterns of Mendelian inheritance are quite common with numerous hereditary diseases. These are autosomal dominant patterns, autosomal recessive patterns and X-linked pattern. Autosomal recessive diseases such as autosomal dominant diabetes include a mutated gene, which an individual inherits two copies of one gene that is inherited from each parent. Carriers who are not displaying any symptoms but with one mutated allele are those who can transmit the mutation to the human offspring. It is found in diseases like Cystic Fibrosis that are a result of mutations in CFTR gene. According to literature, such patterns are important to acknowledge the carriers and determine the risk of transmitting the disease within the family (Nussbaum et al., 2016).

In the same way, Sickle Cell Disease is an autosomal recessive inherited disease. The gene encoding hemoglobin mutation results in a production of rigid and sickle-shaped red blood cells that block the normal circulation of blood and may cause extreme health problems. It has also been identified that epidemiological studies indicate that carriers that have one copy in the mutated gene might have some malaria resistance, proving the evolutionary importance of specific genetic mutations (Strachan and Read, 2018). Such a discovery shows the twin significance of Mendelian genetics: it does not only explain the importance of hereditary diseases, but the overall biological significance of genetic variation.

Autosomal dominant disorders, on the contrary, do not need more than one mutated allele to develop the disease. A good example is the Huntington disease whereby one defective copy of the HTT gene causing progressive degeneration of the neurological system occurs. Those individuals who inherit the gene by a diseased parent stand a fifty percent probability of contracting the disease. The certainty of dominant inheritance means that genetic counselors and clinicians are able to give families accurate risk evaluation, which can get early intervention and surveillance (Pierce, 2017).

Another important issue raised in the literature is the importance of X-linked inheritance, which leads to increased occurrence of some diseases in the male population, including hemophilia and Duchenne muscular dystrophy. In such instances, X chromosomal mutations are manifested in males, because they have only one X chromosome; however, females are usually carriers unless both X chromosomes are mutated. Intelligence effects on X-linked inheritance have been found to aid in informed family planning and proactive diagnostic strategies, which supports the practical significance of Mendelian genetics in clinical practice (Griffiths et al., 2015).

Furthermore, a review of the current literature indicates that Mendelian genetics plays a crucial role in genetic counseling and risk assessment. Through family pedigrees and using the principles of Mendelian, medical practitioners can correctly estimate the likelihood of disease transmission, asymptomatic carriers and offer evidence-based advice to families on matters of reproductive decision making. As the literature indicates, it is vital to highlight that early identification of genetic risks is necessary to achieve effective disease control, preventive health care and minimize the occurrence of hereditary diseases among populations.

Nevertheless, the results have some limitations as well. Genetic inheritance does not all follow the principles of Mendel. Polygenic and complex diseases, including diabetes mellitus, cardiovascular diseases and some cancers, are those that have many genes and environmental factors and thus can not be entirely explained using the Mendelian laws. Nevertheless, in spite of this complexity, Mendelian genetics continues to form the basis by which modern genetic research and studies, such as genome-wide association studies (GWAS) and molecular diagnostics, is built. The results confirm that classical Mendelian principles are still paramount in the interpretation of the occurrence of monogenic disorders and are fundamental to clinical practice and research activities.

3. Discussion

This study finding validate that the Mendelian genetics remains an important theoretical foundation of the hereditary diseases and the medical practice. These law of segregation and independent assortment as postulated by Gregor Mendel allows scholars to define the process of passing genetic traits between parents and the offspring. Not only are these essential laws important in prediction of inheriting traits but also explain genetic information in relation to human disease.

One of the main points which were emphasized in the discussion is the practical uses under the field of medicine to Mendelian genetics. Knowing heritage patterns helps the clinicians and researchers to:

1. Determine genetic mutation carriers - The family pedigrees can be analyzed to determine the people who are carriers of the alleles, which cause the disease but are not expressed. The families which have a history of auto recessive diseases like Cystic Fibrosis should have this knowledge.
2. Anticipate disease risks - Autosomal dominant diseases such as Huntington's Disease could be anticipated using the genotype of one parent and this would play a crucial role in ensuring that interventions and follow ups are made on the basis of a disease at risk.
3. Do: encourage genetic counseling through the delivery of precise estimates of likelihood of a disease occurring- Mendelian genetics can support decision-making processes in families by supporting them in making informed reproductive and medical choices.

The second aspect discussed is the application of the Mendelian genre of genetics using the contemporary genomic technologies. DNA sequencing, polymerase chain reaction (PCR), and gene editing methods including the CRISPR-Cas9 system have increased the uses of the principles of Mendelian genetics. These technologies enable the researchers to identify the specific mutations that cause genetic disorder and come up with specific therapies. Indicatively, the discovery of CFTR mutation in cystic fibrosis patients has seen the generation of the use of genetic methods in treating the condition, as opposed to merely treating the disorder (Collins et al., 2003).

Besides, although the monogenic disease can be explained in terms of Mendelian theory, the discussion does not ignore the difficulty of polygenic and multifactor disorder. Such diseases as type 2 diabetes or hypertension have complicated interactions between various genes and environmental factors, which necessitate higher orders of statistical and genomic model to predict. However, even in such instances Mendelian principles can still be useful to explain the risk of involvement of individual gene mutations in a disease.

The other significance is the research and educational importance of the Mendelian genetics. In instructing such principles, teachers give the students and upcoming researchers a manual approach to comprehend the issue of genetic inheritance. Also, the principles govern the studies of genetic mutations, creation of diagnostic instruments, and methods of the population health that focus on the prevention of hereditary illnesses.

Altogether, it is stated that Mendelian genetics do not only justify past patterns of inheritedness, but also contribute to present-day genetics, medical research and clinical practice. Its combination with the genomic technologies has reshaped the research of the hereditary illnesses allowing to conduct a more accurate diagnosis and early identify the disease as well as provide therapeutic interventions

4. Conclusion

The study shows that Mendelian genetics remains as a solid foundation of genetic science and medical research. The concepts expressed by Gregor Mendel, with the most notable being the laws of segregation and independent assortment, continue to provide a well-organized and consistent model of understanding how genetic traits in human beings inherit.

The findings reveal that many hereditary diseases follow the pattern of the Mendelian inheritance which incorporates autosomal dominant, autosomal recessive, and X-linked. These developed trends enable researchers and health care practitioners to predict the likelihood of disease occurrence, detect the vectors of genetic mutations, and provide specific genetic advice to impacted families. Cystic Fibrosis, Sickle Cell Disease and Huntington, Diseases among others are examples of situations where the application of the principles of Mendelian principles has a direct impact on diagnostic procedures and clinical decision making.

Even though progress has been made in research that reveals the complexity of polygenic and multifactorial disorders, Mendelian genetics is essential in exploring the monogenic diseases. Its concepts are the foundations upon which modern genomic research, molecular diagnostics and personalized medicine are based. Through the combination of the scientifically established classic laws of Mendelian and modern technological progress, scientists are better prepared now to more deeply understand the genetic basis of the disease, increase their efforts in the early diagnostics, and come up with specific treatment methods.

Overall, Mendelian genetics is not only a certain milestone in the olden year in the annals of historical achievement, but also a perennial cornerstone of current medical science. Its values provide theoretical and viable means of understanding, forecasting, and controlling genetic disorders among human populations.

Recommendations

Based on the findings and discussions made, recommendations to the same are provided as follows:

Strengthen genetic education -Academic programs ought to focus on the concepts of the Mendelian genetics to reify the understanding of inheritance, heredity, and disease processes among students, researchers, and medical practitioners. Introduce genetic screening schemes - The health care organizations should promote preventive genetic screening schemes to the families with a known history of inherited diseases, especially the autosomal recessive and X-linked conditions. Increase genetic counseling facilities - Governments and health care organizations are supposed to expand availability of genetic counseling services in order to provide family members with well informed understanding about reproductive options, disease prevention and early intervention measures. Encourage research in genomics and Mendelian genetic research -Money must go towards studies that combine the principles of classical Mendelian with the latest genomic methods, such as DNA sequencing and gene editing, to inspire new diagnostic and therapeutic developments. Create awareness at the population level -The public health program needs to educate the population on hereditary disorders, genetic factors, and the importance of early detection and preventive health service. Bring Mendelian genetics into clinical practice by Health facilities should implement Mendelian based risk assessment tools to better personalized patient care, as well as to inform interventions against hereditary disorders. Foster multi-disciplinary research- Geneticists, clinicians, and molecular biologists are advisable to collaborate to advance the translation of the findings of Mendelian studies to clinical implementation and greater health population policy.

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