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The Impact of Genetics on Human Life

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Abstract: The rapid advancement of genetic testing over the past three decades has transformed modern medicine by enabling precise diagnosis, targeted therapies, and the rise of personalized healthcare. This article explores the evolution and current landscape of genetic testing, focusing on its applications in diagnosing monogenic, chromosomal, and multifactorial disorders. It highlights the significance of novel technologies such as next-generation sequencing (NGS) and whole-exome sequencing (WES) in improving diagnostic accuracy and treatment outcomes. A special emphasis is placed on the Nemours Precision Medicine Initiative, which integrates genetic testing, pharmacogenomics, and biomedical informatics to revolutionize pediatric healthcare through personalized and data-driven approaches. The article also examines the genetic basis of diseases, emphasizing how identifying disease-associated genes aids in the development of targeted therapies and preventive strategies. Furthermore, it discusses the future prospects of genetic testing, including the integration of genomics into routine clinical practice, improvement in genetics education, and addressing disparities in access to testing. Ultimately, understanding the genetic underpinnings of diseases offers immense potential to enhance patient outcomes, reduce healthcare costs, and drive innovation in precision medicine.

Keywords: Genetic testing, Precision medicine, Monogenic disorders, Pharmacogenomics, Genomics, Personalized healthcare

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