Genome Sequencing and Its Impact on Rare Disease Diagnosis

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Abstract. This article examines the transformative role of genome sequencing in the diagnosis and management of rare diseases. By enabling detailed genetic analysis, genome sequencing facilitates the identification of rare genetic disorders that might otherwise go undiagnosed. The article reviews recent advancements in sequencing technologies and discusses their implications for patient care, including personalized treatment plans. The challenges associated with data interpretation and ethical considerations are also explored, offering a comprehensive view of the future of genomics in medicine.

Keywords: genome sequencing, rare diseases, genetic diagnosis, precision medicine, genomics

Introduction

Genome sequencing has revolutionized the field of medical genetics, offering unprecedented insights into the human genome and its role in disease. This article delves into recent advancements in genome sequencing technologies, focusing on their application in diagnosing rare genetic disorders. The ability to sequence an individual's genome provides a powerful tool for identifying genetic mutations responsible for rare diseases, which often remain undiagnosed through traditional diagnostic methods. The discussion highlights the integration of sequencing data into clinical practice, enabling the development of personalized treatment plans tailored to the genetic profile of the patient. Additionally, the article addresses the challenges of data interpretation, storage, and the ethical considerations surrounding genetic information. As technologies continue to advance, genome sequencing is poised to play a critical role in the future of precision medicine and rare disease management.

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