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Diagnosing Chronic Granulomatous Disease: Advances and Challenges in the Genomic Era

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Introduction

Chronic Granulomatous Disease (CGD) is a primary immunodeficiency caused by defects in the NADPH oxidase complex, essential for reactive oxygen species production in phagocytes. This impairment leads to increased susceptibility to infections, particularly by catalase-positive bacteria and fungi, and the formation of granulomas. Historically, diagnosis relied on clinical presentation and functional assays, but genomic technologies have revolutionized the diagnostic process. This review discusses advancements in CGD diagnosis, the role of genomic techniques, and the challenges faced in implementing these technologies effectively. Initial diagnosis often involves identifying recurrent infections and characteristic granulomas in patients. Historically, diagnosis was confirmed through assays like the Nitroblue Tetrazolium test and Dihydrorhodamine (DHR) flow cytometry, which assess the ability of phagocytes to produce reactive oxygen species.

Description

Early genetic testing focused on known CGD-related genes, such as CYBB, CYBA, NCF1, NCF2, and NCF4, but was limited by the availability and cost of sequencing technologies. NGS has transformed the genetic diagnosis of CGD by enabling comprehensive screening of multiple genes simultaneously. This approach reduces diagnostic time and improves accuracy. WES focuses on coding regions of the genome, providing detailed information on mutations affecting protein function. WGS offers a broader view of the genome, including non-coding regions and structural variants, potentially revealing novel mutations. Advanced bioinformatics tools and databases facilitate the interpretation of genetic variants, helping to distinguish pathogenic mutations from benign polymorphisms. Custom gene panels specific to CGD can streamline the diagnostic process by targeting known disease-associated genes [1].

CGD is caused by mutations in several different genes, leading to a wide spectrum of genetic variations. This diversity can complicate the interpretation of genetic tests. The identification of novel or rare mutations not covered by standard panels can delay diagnosis and require additional research. NGS and WGS may miss small variants or structural abnormalities if coverage is insufficient. Ensuring high-quality sequencing and accurate variant calling is crucial. The vast amount of data generated by genomic technologies necessitates sophisticated tools and expertise to accurately interpret results and distinguish pathogenic mutations. While genomic technologies have become more accessible, disparities in healthcare infrastructure can affect the availability of advanced diagnostic tools. The high cost of genomic testing can be a barrier to widespread adoption, particularly in resource-limited settings. Development of more comprehensive gene panels that include newly identified CGD-related genes can enhance diagnostic accuracy [2,3].

Combining genetic testing with functional assays can confirm the clinical significance of identified mutations and improve diagnostic certainty. Efforts to reduce the cost of genomic testing and improve accessibility will facilitate broader use and earlier diagnosis. Building and maintaining comprehensive databases of genetic variants and their associated phenotypes will support better interpretation of genomic data. Understanding the specific genetic mutations in CGD patients can lead to more personalized treatment strategies and improved management of the disease. Ongoing research into gene therapy offers potential for future therapeutic interventions that address the underlying genetic defects in CGD [4,5].

Conclusion

The genomic era has brought significant advancements in the diagnosis of Chronic Granulomatous Disease, offering more precise and comprehensive methods for identifying pathogenic mutations. While these advancements present exciting opportunities, challenges related to genetic heterogeneity, technical limitations, and accessibility remain. Continued progress in genomic technologies, coupled with efforts to address these challenges, will enhance diagnostic accuracy and patient care for individuals with CGD.

Acknowledgement

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Conflict of Interest

None.

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