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Perspective

Targeted Sequencing in Duchenne and Becker Muscular Dystrophy: Unraveling Novel Variants

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DESCRIPTION

Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD) are X-linked recessive disorders characterized by progressive muscle weakness and degeneration. Both conditions are caused by mutations in the dystrophin gene but differ in terms of severity and age of onset. Next-generation Sequencing (NGS) has revolutionized genetic research and diagnostic approaches, enabling the identification of disease-causing variants with high accuracy and efficiency. Next-Generation Sequencing (NGS) is a high-throughput DNA sequencing technology that has revolutionized genetic research and clinical diagnostics. NGS enables the simultaneous sequencing of millions of DNA fragments, allowing for rapid and cost-effective analysis of entire genomes, exomes, or specific targeted regions. NGS-based targeted sequencing is a focused approach that selectively captures and sequences specific genomic regions, such as genes or gene panels associated with a particular disease or condition. NGS-based targeted sequencing offers several advantages over traditional sequencing methods. It allows for the simultaneous analysis of multiple genes or genomic regions in a single experiment, reducing the time and cost associated with analyzing individual genes separately. Targeted sequencing is particularly useful when studying diseases with known genetic associations, such as cancer or genetic disorders like (DMD) and Becker Muscular Dystrophy (BMD). DMD and BMD are caused by mutations in the dystrophin gene which codes for the dystrophin protein involved in maintaining the integrity of muscle cells.

Targeted sequencing allows for the efficient and accurate identification of variants within the dystrophin gene, aiding in the diagnosis, prognosis, and genetic counseling of affected individuals. When performing NGS-based targeted sequencing for DMD and BMD, researchers and clinicians typically design gene panels specifically targeting the dystrophin gene or a set of genes associated with muscular dystrophies. The design of these panels involves selecting and capturing relevant genomic regions using complementary DNA probes or primers. The analysis of NGS data involves several steps, including sequence alignment to a reference genome, variant calling, and annotation. Bioinformatics tools and pipelines are used to identify genetic variants, such as point mutations, insertions, deletions, or large structural variations within the targeted genomic regions. These identified variants are then evaluated for their pathogenicity, potential impact on protein function, and correlation with the phenotype. NGS-based targeted sequencing has led to the discovery of numerous novel variants in the dystrophin gene associated with DMD and BMD. These novel variants provide valuable insights into the genetic basis of the disorders and contribute to the expanding knowledge of the phenotypic spectrum. Furthermore, the identification of specific variants aids in improving the accuracy of genetic diagnoses and enables personalized treatment approaches. While NGS-based targeted sequencing offers significant advantages, it is not without limitations.

Targeted sequencing focuses only on pre-selected genomic regions, potentially missing variants located outside the targeted regions. General overview of the characteristics that can be associated with novel variants are identified in the dystrophin gene. The location of the variant within the dystrophin gene can provide valuable information about its potential impact. Variants can occur in different regions of the gene, such as exonic (coding regions), intronic (non-coding regions), or regulatory regions. Novel variants can encompass different types of genetic alterations, including Single Nucleotide Variants (SNVs), insertions, deletions, or complex structural variations. Each type of variant can have specific implications for the resulting protein structure and function. Variants within the dystrophin gene can lead to various effects on the resulting dystrophin protein. Some variants may disrupt the reading frame, resulting in a truncated or nonfunctional protein.

Evaluating the pathogenicity often involves considering multiple lines of evidence, such as population frequency, computational predictions, functional studies, and correlation with the phenotype. Some variants in the dystrophin gene can be

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associated with specific disease phenotypes or severity. Variants that are rare or unique to specific individuals or families may require further investigation to determine their clinical significance. Some potential implications of variants on disease severity and progression: Variants in disease-associated genes can contribute to phenotypic heterogeneity, influencing the spectrum and severity of clinical manifestations. Different variants within the same gene can lead to distinct phenotypes, ranging from mild to severe forms of the disease. Variants in protein-coding regions of genes can result in altered protein structure and function. These changes can disrupt important protein-protein interactions, enzymatic activity, or protein stability, leading to functional impairment. Variants that severely impact protein function are more likely to result in a severe disease phenotype, whereas variants with milder effects may manifest as a milder form of the disease.

Variants that disrupt regulatory elements may lead to reduced or increased gene expression, impacting disease phenotype. Disease-associated genes often participate in complex molecular networks and pathways. Variants in these genes can disrupt the normal functioning of these pathways, leading to disease development and progression. Variants in different genes can interact with each other, modulating disease severity and progression. Modifier genes are genes that can influence the phenotype of individuals with specific disease-causing variants. Identifying modifier genes can explain the variability in disease severity observed among individuals with the same primary disease-causing variant. Modifier genes may have a direct or indirect effect on disease progression and response to treatment.

CONCLUSION

NGS-based targeted sequencing has revolutionized the field of genetics, enabling rapid and accurate analysis of specific genomic regions associated with diseases like DMD and BMD. It has played a critical role in identifying novel variants, improving diagnostic accuracy, and guiding personalized management strategies. As NGS technologies continue to advance and become more accessible, targeted sequencing will continue to shape the field of genetic research and contribute to the understanding and management of genetic disorders.