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Perspective

Aortic Aneurysm Progression: A Novel Frontier in Disease Research

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DESCRIPTION

The aorta, the largest artery in the body, collapses and swells in aortic aneurysms, which can be fatal vascular disorder. This condition often develops without noticeable symptoms until it reaches to a critical stage, at which point it becomes a medical emergency. Understanding the mechanisms and regulatory factors contributing to aortic aneurysms is significant for early diagnosis and the development of effective treatment strategies. In recent years, there has been a continuous focus on the genetic and epigenetic factors that play a significant role in the pathogenesis of aortic aneurysms.

Genetic factors in aortic aneurysms

Familial aggregation: Aortic aneurysms have been observed to cluster within families, suggesting a genetic component. Several genetic syndromes, such as Marfan syndrome, Ehlers-Danlos syndrome, and Loeys-Dietz syndrome, are associated with an increased risk of aortic aneurysms.

Candidate genes: It has been identified that various candidate genes associated with aortic aneurysms. These include genes that encode components of the extracellular matrix, such as Fibrillin-1 (*FBN1*) and Transforming Growth Factor-beta (TGF-β) receptor genes (*TGFBR1* and *TGFBR2*).

Genetic variants: Single Nucleotide Polymorphisms (SNPs) have been associated with an increased risk of aortic aneurysms. For example, certain SNPs in the *TGFBR1* gene have been linked to a higher susceptibility to thoracic aortic aneurysms.

Heritability: Combined studies have suggested that aortic aneurysms have a significant heritable component. The heritability of thoracic aortic aneurysms has been estimated at approximately 60%.

Epigenetic factors in aortic aneurysms

DNA methylation: Epigenetic modifications, such as DNA methylation, play a role in regulating gene expression. Abnormal DNA methylation patterns have been identified in aortic aneurysm tissues, suggesting their involvement in the disease pathogenesis.

Histone modifications: Alterations in histone modifications can impact the expression of genes related to vascular tissue remodeling and inflammation. Research has shown that histone modifications, such as histone acetylation and methylation, are implicated in aortic aneurysms.

Non-coding RNAs: Non-coding RNAs, including microRNAs (miRNAs), have emerged as key epigenetic regulators in aortic aneurysms. Certain miRNAs have been found to target genes associated with extracellular matrix degradation and vascular smooth muscle cell dysfunction.

Interaction between genetic and epigenetic factors

The development of aortic aneurysms is often the result of a complex interplay between genetic and epigenetic factors. Epigenetic modifications can influence the expression of genes with known genetic variants associated with aortic aneurysms. For example, epigenetic changes may enhance or suppress the effects of genetic mutations, thereby contributing to disease progression.

TGF-β signaling pathway

The TGF-β signaling pathway is central to the pathogenesis of aortic aneurysms. Both genetic mutations and epigenetic modifications can impact the components of this pathway. Genetic variants in genes encoding TGF-β receptors, ligands, or downstream signaling molecules can lead to the overactivation of the TGF-β pathway. Epigenetic changes, such as histone modifications or miRNA dysregulation, can further exacerbate this abnormal signaling.

Epigenetic modifications and aneurysm progression

Vascular Smooth Muscle Cells (VSMCs): Epigenetic alterations in VSMCs can promote phenotypic switching, causing these cells to become more migratory and less contractile. This phenotypic switch contributes to the structural degeneration of the aortic wall.

Extracellular matrix remodeling: Abnormal epigenetic changes can lead to excessive extracellular matrix degradation and

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insufficient matrix synthesis. This imbalance results in aortic wall weakening, leading to aneurysm formation and progression.

Therapeutic implications

Understanding the genetic and epigenetic factors involved in aortic aneurysms is essential for the development of targeted therapies.

Gene-based therapies: Targeting specific genes associated with aortic aneurysms, such as those involved in the TGF-β pathway, may help mitigate disease progression.

Epigenetic modulation: Epigenetic therapies, including DNA methylation inhibitors and histone deacetylase inhibitors, can be explored to correct abnormal epigenetic modifications in aortic aneurysms.

Combined approaches: A combination of gene-based and epigenetic therapies may offer a more comprehensive strategy to address the multifactorial nature of aortic aneurysms.

Understanding the interplay between genetic and epigenetic factors is vital for the development of novel diagnostic and therapeutic strategies. Targeted gene-based therapies and epigenetic modulation may hold the potential of more effective interventions to prevent the devastating consequences of aortic aneurysms and improve patient outcomes.