Commentary

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KMT2E gene and its role in neurodevelopmental and psychiatric disorders

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DESCRIPTION

The KMT2E gene, which belongs to the KMT2 family of histone methyltransferases, is essential for controlling neurodevelopmental processes and gene expression. Intellectual disability (ID), autism spectrum disorder (ASD), schizophrenia, and epilepsy are among the neurodevelopmental and psychiatric conditions that have been linked in recent research to KMT2E mutations. The importance of KMT2E in brain function becomes more widely acknowledged, a thoroughly review of the body of research is necessary. Our systematic review attempts to summarize recent research on the relationship between neurodevelopmental and psychiatric traits and variations of the KMT2E gene.

Our systematic study shows that KMT2E variations, including deletions and mutations, are strongly linked to a higher risk of epilepsy, schizophrenia, autistic spectrum disorder, and intellectual disability. Individuals with KMT2E mutations frequently exhibit cognitive deficits, developmental delays, and an increased risk of seizures. These results highlight the crucial function of KMT2E in the regulation of gene expression and neurodevelopmental pathways. Beyond the neurological symptoms, individuals with KMT2E-related disorders often present with a spectrum of behavioral and psychiatric manifestations. These may include hyperactivity, anxiety, mood instability, and in some cases, features that overlap with broader psychiatric conditions such as schizophrenia. The phenotypic variability observed among individuals with KMT2E mutations suggests the involvement of additional genetic, epigenetic, or environmental factors that may influence the severity and nature of clinical outcomes.

At the molecular level, KMT2E is known to modulate chromatin structure through histone methylation, thereby impacting the transcriptional regulation of genes involved in neurogenesis, synaptic function, and neuronal migration. Disruption of these processes, due to altered KMT2E activity, likely contributes to the wide range of neurodevelopmental impairments observed. Importantly, KMT2E does not act in isolation but interacts with other epigenetic regulators, further complicating the landscape of its downstream effects.

Recent advancements in high-throughput sequencing technologies and genome-wide association studies (GWAS) have facilitated the identification of pathogenic KMT2E variants in diverse populations. Functional studies using animal models and patient-derived cells are beginning to elucidate the specific pathways and mechanisms through which KMT2E mutations exert their effects. For instance, models have demonstrated aberrant neuronal differentiation and reduced synaptic connectivity associated with KMT2E dysfunction.

CONCLUSION

This systematic review underscores the pivotal role of the KMT2E gene in maintaining normal neurodevelopmental processes and cognitive function. Variants in KMT2E, including both deletions and mutations, are recurrently associated with a spectrum of neurological and psychiatric conditions such as intellectual disability, autism, epilepsy, and schizophrenia. Individuals carrying these variants commonly display developmental delays, learning difficulties, and seizure susceptibility, suggesting that KMT2E is integral to pathways governing brain development and neuronal stability. As research into the genetic underpinnings of neurodevelopmental disorders progresses, a deeper understanding of KMT2E's biological functions may offer valuable insights into the diagnosis and potential treatment strategies for these complex conditions.