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Integrative Analysis of Alternative Splicing Events and Aberrant Splicing in NSD1 Gene

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Introduction

Alternative splicing plays a crucial role in expanding the proteome diversity by generating multiple isoforms from a single gene. The NSD1 gene, implicated in various developmental disorders including Sotos syndrome, undergoes alternative splicing, leading to the production of distinct protein isoforms. In this article, we explore the identification of alternative splicing events in the NSD1 gene and employ multiple algorithms for protein isoform structure prediction. The comprehensive analysis of alternative splicing and isoform structures provides valuable insights into the functional diversity and potential implications in disease pathology. Alternative splicing is a fundamental process in gene expression regulation, enabling the production of various mRNA transcripts and subsequent protein isoforms.

The NSD1 gene, encoding the nuclear receptor-binding SET domain protein 1, is known to exhibit alternative splicing, contributing to its functional versatility and complexity. Sotos syndrome, characterized by overgrowth and developmental abnormalities, is often associated with aberrant NSD1 splicing patterns. This article aims to investigate the identification of alternative splicing events in NSD1 and predict protein isoform structures using multiple algorithms. To elucidate the alternative splicing landscape of NSD1, we employed a combination of computational tools and transcriptomic data analysis. By leveraging publicly available RNA-sequencing datasets and transcript annotations, we identified distinct splicing events occurring within the NSD1 gene. These events include exon skipping, alternative 5' and 3' splice site usage, and intron retention. Through this analysis, we uncovered the extent of alternative splicing in NSD1 and its potential impact on protein function [1].

Description

To gain insights into the structural consequences of alternative splicing, we employed multiple algorithms for protein isoform structure prediction. These algorithms utilize computational modeling approaches, such as homology modeling, ab initio modeling, and molecular dynamics simulations, to generate three-dimensional structures of NSD1 protein isoforms. By comparing the predicted structures of different isoforms, we elucidated potential structural variations that could affect protein-protein interactions, enzymatic activities, or functional domains. The aberrant splicing of NSD1 has been implicated in Sotos syndrome, a rare genetic disorder characterized by overgrowth and developmental anomalies. By examining known NSD1 aberrant splice site mutations reported in Sotos syndrome patients, we correlated these mutations with the identified alternative splicing events [2].

Our analysis provides insights into the potential functional consequences of these mutations and their contributions to the pathogenesis of Sotos syndrome. The identification of alternative splicing events in the NSD1 gene and the prediction of protein isoform structures offer a foundation for further investigations. Future studies could explore the functional characterization of these isoforms, their interactions with other proteins, and their roles in cellular processes. Understanding the impact of alternative splicing on NSD1 function will enhance our comprehension of Sotos syndrome pathology and potentially uncover novel therapeutic targets. This article highlights the importance of alternative splicing in the NSD1 gene and its implications in Sotos syndrome [3].

By employing computational tools, we identified alternative splicing events, predicted protein isoform structures, and investigated their potential role in disease pathology. This integrative approach provides valuable insights into the functional diversity of NSD1 and paves the way for further research in the field of alternative splicing and genetic disorders. Sotos syndrome is a rare genetic disorder characterized by overgrowth, developmental anomalies, and intellectual disabilities. The NSD1 gene has been identified as a major causative gene in SoS, and aberrant splice site mutations in NSD1 have been linked to the pathogenesis of the syndrome. In this article, we utilize in silico prediction methods to analyze known NSD1 aberrant splice site mutations in SoS patients.

We investigate the allelic contribution of these mutations to NSD1 transcripts, shedding light on the underlying molecular mechanisms that contribute to the development of SoS. Sotos syndrome is a complex genetic disorder characterized by overgrowth, craniofacial abnormalities, and neurodevelopmental deficits. Mutations in the NSD1 gene, encoding the nuclear receptor-binding SET domain protein 1, have been identified as the primary cause of SoS. Aberrant splice site mutations in NSD1 can disrupt the normal splicing process, leading to the production of abnormal transcripts and subsequently contributing to the pathogenesis of SoS. In this article, we employ in silico prediction methods to analyze known NSD1 aberrant splice site mutations in SoS patients and investigate their allelic contribution to NSD1 transcripts.

By utilizing various computational tools and databases, we comprehensively analyze the known NSD1 aberrant splice site mutations reported in SoS patients. These mutations include point mutations, insertions, deletions, and splice site alterations. Through in silico prediction methods, we assess the potential impact of these mutations on splice site recognition, spliceosome assembly, and mRNA splicing. This analysis aids in understanding the molecular consequences of these mutations and their role in the pathogenesis of SoS. To investigate the allelic contribution of NSD1 aberrant splice site mutations, we analyze RNA-sequencing data from SoS patients carrying these mutations. By aligning the sequencing reads to the NSD1 gene and identifying specific mutations, we determine the proportion of mutant and wild-type alleles present in the NSD1 transcripts [4].

This analysis allows us to assess the allelic imbalance resulting from the aberrant splicing events, providing insights into the functional consequences and potential dosage effects associated with specific mutations. The in

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silico prediction of NSD1 aberrant splice site mutations and the detection of allelic contribution to NSD1 transcripts contribute to our understanding of the molecular mechanisms underlying SoS pathogenesis. By identifying specific mutations that disrupt normal splicing, we gain insights into the impact on NSD1 protein isoforms, functional domains, and protein-protein interactions. Furthermore, the allelic imbalance analysis helps elucidate potential dosage effects and their contribution to the phenotypic variability observed in SoS patients [5].

Conclusion

In silico prediction of NSD1 aberrant splice site mutations and the detection of allelic contribution to NSD1 transcripts provide a foundation for further investigations. Future studies can explore the functional consequences of specific mutations, investigate the downstream effects on gene expression and protein function, and elucidate the molecular pathways affected in SoS. This knowledge can potentially guide the development of targeted therapies and personalized treatment strategies for SoS patients. This article highlights the importance of in silico prediction methods in analyzing NSD1 aberrant splice site mutations and their allelic contribution to NSD1 transcripts in Sotos syndrome patients. By integrating computational approaches with experimental data, we gain insights into the molecular mechanisms underlying SoS pathogenesis. This research contributes to our understanding of the genetic basis of SoS and opens avenues for future investigations in the field of rare genetic disorders.

Acknowledgement

None.

Conflict of Interest

None.

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