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Abstract Title: - Exploring the Potential Association of THAP9 and THAP9-AS1 in Cockayne Syndrome Type B and Neurodegenerative Diseases

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Aims: - THAP9 is a transposable element-derived gene that expresses a protein homologous to the Drosophila P element transposase. The THAP9-AS1 lncRNA is transcribed from the antisense strand of the THAP9 gene via a putative bidirectional promoter. THAP9 and THAP9-AS1 have been reported to be involved in various cancers, and their expression is sometimes strongly correlated with patient prognosis. Aberrant activation of transposable elements has been found in many neurological disorders, including Alzheimer's and Parkinson's. It is reported that microglia and astrocytes respond to the abnormal presence of tau protein with induced transposable element transcription in Alzheimer's disease. We want to look further into how these genes interact with THAP9, a DNA transposon and THAP9-AS1, a lncRNA in conditions like neurodegeneration. According to GeneCards, THAP9 is associated with Cockayne Syndrome B, a rare genetic disorder characterised by growth failure, premature ageing, and sensitivity to sunlight.

The aim is to study the THAP9 and THAP9-AS1 gene locus and its interaction with other genes that could contribute to the related diseased conditions.

Methods: - Using RNA-seq and microarray data, the identification of THAP9 and THAP9-AS1 as potential modifier genes in diseases will be achieved through a multi-pronged approach. This encompasses various techniques, including differential gene expression analysis, co-expression analysis, and functional assessments. By employing this comprehensive methodology, we will gain valuable insights into the plausible contributions of these genes to diseases like neurodegeneration and Cockayne Syndrome (CSB).

Results: - This study examines whether THAP9 and THAP9-AS1 interact with the genes involved in neurodegenerative diseases or rare genetic conditions and the consequences of this interaction.

Conclusions: - Genetic analysis will uncover associations between THAP9 and THAP9-AS1 with disease outcomes, which could provide valuable insights into disease mechanisms and the functions of these genes. We focus on the possible association of THAP9 and THAP9-AS1 in neurodegenerative diseases and some rare conditions.

Keywords: - This study examines whether THAP9 and THAP9-AS1 interact with the genes involved in neurodegenerative diseases or rare genetic conditions and the consequences of this interaction.