

Genetic Investigation of Laryngeal Papillomatosis: Insights from Whole Genome Sequencing and Transcriptome Analysis

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Introduction: Laryngeal papillomatosis (LP) is a debilitating condition characterized by the formation of benign epithelial tumors in the larynx. While viral etiology, is well-established in a subset of cases, the majority of LP cases lack a clear etiological basis. Here, we present our investigation involving five patients with severe LP, two of whom had tracheostomies due to the aggressive course of the disease. Secondary causes were excluded, prompting consideration of a genetic predisposition.

Laryngeal papillomatosis of unknown etiology



<https://ncicb.nci.nih.gov/xml/owl/EVS/thesaurus/t3/cgi-bin/ncicb/ncitbrowser/ConceptReport.jsp?dictionary=papillomatosis-epi>

Methodology: Whole genome sequencing (WGS) was conducted on DNA samples from all five patients (three females (aged 3, 7 and 20) and two males (aged 7 and 45)) accompanied by comprehensive transcriptome analysis to indicate potential genetic determinants contributing to LP pathogenesis.



Results: Despite rigorous analysis, no pathogenic genetic mutations were identified within the coding or non-coding regions of the genome. Our findings underscore the complexity of LP etiology. The absence of identifiable genetic mutations implicating LP pathogenesis in our study underscores the need for multifaceted approaches encompassing epigenetic, and environmental factors to identify molecular landscape of this disorder.

Discussion: Further exploration employing advanced genomic techniques coupled with functional validation studies is warranted to unravel the genetic foundation of LP and pave the way for personalized therapeutic interventions.

In conclusion, our study highlights the challenges in detecting the genetic basis of LP and emphasizes the importance of a comprehensive understanding that incorporates various genetic and environmental factors. Such insights are crucial for developing targeted therapies and personalized management strategies for individuals affected by this condition.