

Whole-transcriptome oropharynx expression in adulthood sleep-disordered breathing- A case-controlled pilot study

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Objective: Sleep-disordered breathing (SDB) including primary snoring and obstructive sleep apnea (OSA) is associated with many medical comorbidities. The genetic basis of SDB has been poorly characterized. We sought to quantify oropharyngeal gene expression of SDB patients underwent palatal surgery and to establish the yield of modern next generation sequencing (NGS) technologies in this setting.

Method: This pilot study enrolled 10 SDB patients underwent palatal surgery: 5 severe OSAs (apnea-hypopnea index [AHI] \geq 30/h) and 5 controls (AHI $<$ 15/h). Both the groups were matched by age, sex, and body mass index. Fresh tissues of the resected uvula or soft palate were collected for NGS genetic evaluation after extraction of mRNA. Whole-transcriptome oropharyngeal expression was analyzed using RNA-seq.

Results: Differences in median tonsil size and tongue position between the 2 groups were statistically insignificant (both $P > 0.05$). Fifty-eight differentially expressed genes (13 up-regulation and 45 down-regulation) between the 2 groups were identified. Exploratory gene ontology analyses revealed differential expression of 2 components (extracellular region, extracellular region part), 4 functions (cytokine receptor binding, chemokine receptor binding, cytokine activity, receptor binding), and 13 processes (5 related to development, 5 related to immune response, 3 related to differentiation) (adjusted $P < 0.1$). Kyoto Encyclopedia of Genes and Genomes pathway analysis revealed that these target transcripts were implicated in 78 various pathways such as hematopoietic cell lineage, primary immunodeficiency, African trypanosomiasis, B cell receptor signaling pathway, and amoebiasis in sequence of Q values.

Conclusion: Oropharynx gene expression of SDB patients undergoing palatal surgery is heterogeneous and involves multiple genes. NGS technology allows identification of putative pathways and mechanisms of SDB and its future applications in disease prevention and treatment need further study.

中文題目：成人呼吸障礙的口咽全轉錄體表現——一個病例控制先驅研究

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