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A step to diagnosis of sleep apnea with next generation sequencing

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Abstract

The introduction of next generation sequencing (NGS) has led to an exponential increase of elucidated genetic causes in both extremely rare diseases and common but heterogeneous disorders. It can be applied to the whole or to selected parts of the genome (genome or exome sequencing, gene panels). NGS is applied in both research and clinical settings, and there is a rapid transition of research findings to diagnostic applications. These developments may greatly help to minimize the "diagnostic odyssey" for patients as whole-genome analysis can be performed in a few days at reasonable costs compared with gene-by-gene analysis based on Sanger sequencing following diverse clinical tests. Despite the enthusiasm about NGS, one has to keep in mind its limitations, such as a coverage and accuracy of < 100%, resulting in missing variants and false positive findings. Therefore, there is an urgent need to define standards for NGS with respect to run quality and variant interpretation, as well as mechanisms of quality control.

Aberrant respiratory control mechanisms have been implicated in dentofacial deformities, such as long face syndrome or adenoid facies. Obstructive sleep apnea (OSA) is a potentially life-threatening condition in which the patient suffers periodic cessation of breathing during sleep and is the most important etiological factor in the long face syndrome. The symptoms include loud snoring, irregular breathing patterns and restless movements during sleep which impairs the quality of life. The aim of this study is to determine the genetic association of obstructive sleep apnea associated genes (ACE, TNF- α , IL-6, 5-HTR2A, 5-HTR2C, 5-HTT, LEPR, PPAR- γ , ADRB, and APOE) with specific primers in polymerized chain reaction through an extensive genome search in Odisha population.

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