J Mol Genet Med 2018, Volume 12
DOI: 10.4172/1747-0862-C3-031

A genetic study of hereditary gingival fibromatosis patients by regulatory network analysis connecting exomic variants and transcriptomic alterations

Jae Hoon Lee
Yonsei University, Korea

Hereditary gingival fibromatosis (HGF) is a rare oral disease characterized by either localized or generalized gradual, benign, non-hemorrhagic enlargement of gingivae. Although several genetic causes of HGF has been known, genetic etiology of HGF from the non-syndromic and idiopathic entity is still uncertain. Here, we analyzed exomic and transcriptomic alterations from the familial-case study of idiopathic HGF patients and non-diseased persons. Our regulatory network analysis finds that exomic variants and differentially expressed genes were connected by the member of TGF-β/SMAD signaling pathway or craniofacial development processes, which explain molecular mechanism of fibroblast overgrowth mimicking HGF and distinctive face that is comorbid cases of HGF, respectively. Moreover, a data-mining resource of gene-phenotype mapping and semantic analysis of disease phenotype similarity support that genes derived from our network analysis have pathogenic roles in fibromatosis-related and craniofacial diseases. Our study suggests that the computational approach of connecting exomic and transcriptomic alteration through the regulatory network is applicable for the clinical interpretation of genetic variant in HGF patients.

jaehoon115@yuhs.ac

Notes: