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Pathway analysis of rare variants using generalized structured component analysis

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Most approaches for rare variants focus mainly on individual gene analysis. However, it has also been recognized that a majority of biological behaviors manifest from a complex interaction of biological pathway. Although several statistical methods for pathway-based analyses using rare variants have been proposed to analyze pathways individually, those methods generally assume that the pathways are independent and do not consider substantial overlapping genes between pathways. In this paper, we propose a novel statistical approach: a robust pathway-based approach using hierarchical components. The proposed method uses generalized structural components and analyzes all pathways from the public databases such as Kyoto Encyclopedia of Genes and Genomes (KEGG) pathway database. To consider the relationship between genes within-pathway, we use both reflective and formative models. Furthermore, our approach uses ridge-regularized method to reduce the effects of the overlapping genes between pathways. Evaluation of the method was performed with the simulation data sets as well as the sequencing data for the phenotype of autism.

Biography

Taesung Park has completed his PhD from University of Michigan and visiting fellow/scholar from National Institutes of Health. He is a full Professor of the department of statistics and the Director of National Creative Research Lab for Bioinformatics and Biostatistics, Seoul National University, Korea. Based on the experience of statistical researches, he founded this lab to extend his research area to bioinformatics and biostatistics. He has published more than 160 papers in SCIE journals and served as Associate Editor and Editorial Board Member of international journals such as *CSDA*, *Biometrical Journal*, *Genetic Epi.*, and Book Editor of *Biometrics*.

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