Special Issue

Statistical Developments and Applications in Rare Disease Diagnostic Testing and Classification

Message from the Guest Editor

Rare inborn genetic diseases, due to their low birth rates and commonality of symptom presentation, have been a challenge to diagnose and classify. The increasing use of 'big data' and proliferation of various screening techniques have allowed for recent advances in the diagnostic tools and statistical methods vital to the accurate identification of the disease type. This Special Issue therefore aims to collect research articles related to disease diagnosis and classification, including original developments, novel applications, as well as review articles and short commentary or editorials. While rare inborn genetic disorders and diseases are the motivating topic for this Special Issue, diagnostic challenges among other types of conditions are also of interest. Specific topics for consideration include, but are not limited to, multivariate normal limits, artificial intelligence and/or machine learning tools, Bayesian models, multinomial modelling, missing data analysis, clinical trials, retrospective analyses, and simulation designs, etc.

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