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A rare-variant association test for non-normal/censored outcomes in family-based designs

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Genome-wide association studies have identified over 6,000 common variants associated with over 500 complex diseases and traits. However, for most phenotypes only a small portion of the heritability is explained by these common variants. With the rapid development of sequencing technologies, it is now possible to identify rare variants that are thought to be associated with complex traits. However, the efficiency of these efforts heavily depends on the use of appropriate statistical methods to analyze the collected data. In this presentation, I will be concerned with rare-variant association tests in the presence of familial dependencies. Two cases will be considered: non-normal and right-censored phenotypes. In both cases, the familial dependence will be modeled via a Gaussian copula and a flexible regression model will be proposed for the marginal behavior of the phenotype. Score-type tests will be derived and several strategies to assess the significance of the resulting test statistics will be proposed and compared. The performance of the proposed tests will be evaluated and compared to existing approaches with simulations and their use will be illustrated with real data association studies.

This is joint work with Karim Oualkacha, Celia Greenwood, Martin Leclerc, and Jacques Simard.

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