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Detection of rare causal DNA mutations from common markers: sample applications

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We demonstrate the applicability of inconsistent ranking techniques to the detection of rare and distributed genomic mutations associated with complex diseases. The idea is to identify signatures of such mutations by linkage to inconsistent signals from common markers in their vicinity. We focus on examples of analysis of large genome wide association studies (GWAS with $10^4 - 10^5$ samples) for breast cancer, celiac and primary sclerosing cholangitis.

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