Bayesian analysis of allelic penetrance models for complex binary traits

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Abstract

Complex binary traits result from an intricate network of genetic and environmental factors. To aid their genetic dissection, several generalized linear models have been used to detect interaction between genes. However, it is recognized that these models have limited genetic interpretation. As an attempt to overcome this problem, we have previously proposed the allelic penetrance approach to model dominance and recessiveness at a single locus and to describe independent, inhibition and cumulative actions between two diallelic loci. Classically, recessive inheritance requires the expression of both recessive alleles at homozygotes to obtain the phenotype (type I recessiveness). In previous work, we alternatively define recessiveness as a situation where a recessive allele could express the phenotype when the dominant allele is not active (type II recessiveness).

Here the allelic penetrance models are revisited under both definitions of recessiveness. We apply Bayesian methods to analyze two data sets: one regarding the effect of a given haplotype on the inheritance of two immunoglobulin deficiencies (IgD and IgG4) in humans and the other related to joint action of two loci on Listeria infection susceptibility in mice resulting from an intercross between two strains. Our results indicate that IgD and IgG4 deficiencies are dominant and type I recessive traits with respect to the given haplotype, respectively. Regarding Listeria infection, susceptibility appears to be controlled by an independent action between a locus with a dominant allele inherited from the resistant strain and a locus at another chromosome with a type I recessive allele derived from the susceptible strain.