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Genome-wide significance

In genome-wide association studies, **genome-wide significance** (abbreviated **GWS**) is a specific threshold for determining the <u>statistical significance</u> of a reported <u>association</u> between a given <u>single-nucleotide</u> <u>polymorphism</u> (SNP) and a given <u>trait</u>. The most commonly accepted threshold is $p < 5 \times 10^{-8}$, which is based on performing a <u>Bonferroni correction</u> for all the independent common SNPs across the <u>human genome</u>. If a *p*-value is found to be lower than this threshold in a genome-wide association study, the <u>null hypothesis</u> of no true SNP-association will typically be rejected. However, there has been some criticism of this threshold, with a 2012 study suggesting that a significant number of associations with *p*-values just above this threshold are genuine, replicable associations. The authors of this study concluded that their finding "...suggests a possible relaxation in the current GWS threshold." More recently, it has been suggested that the conventional threshold should be modified to take into account the increasing prevalence of low-frequency genetic variants in genome-wide association studies. [4]

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