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Review article

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Genetics and epigenetics of primary Sjögren syndrome: implications for future therapies

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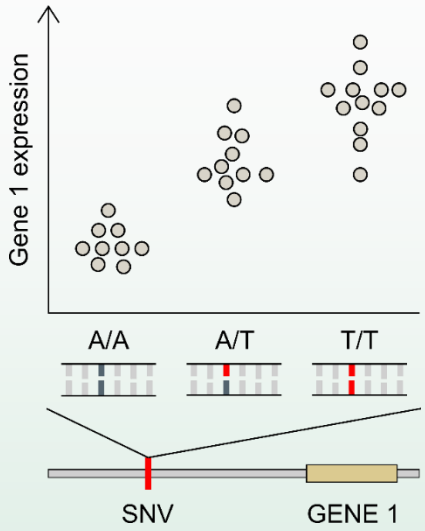
Supplementary box 1. Quantitative Trait Loci

Quantitative trait loci (QTL) refer to genetic loci (such as single nucleotide variants (SNVs)) that are associated with differences in quantifiable phenotypes, including most often differences in gene expression (eQTL; as shown in the figure), but also differences in DNA methylation (meQTL) or protein levels. eQTLs are identified by correlating the genotype of an individual with the expression of genes present within a certain distance on the chromosome of the genetic variant (cis-QTLs) or genes at any location, regardless of the position (trans-QTLs). No constraints are set in terms of the direction of the genotype-to-expression relationship. eQTLs are often context-dependent, meaning that they can be dependent on the cell-type and tissue and can be influenced by the sex of the carrier



eQTLs can be measured individually by comparing the genotype in one position to the expression of one or more nearby genes, or in a genome-wide manner, utilizing genotyping of the whole genome and array expression or RNA sequencing of all genes to systematically scan for these effects. In eQTL analyses that span the whole genome, multiple testing should be appropriately accounted for. In the case of trans-eQTL analyses, the weight of multiple testing makes weaker effects difficult to detect.