





Functional analysis and allelic association

Variants

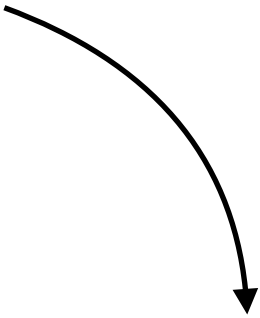
# Functional analysis

Exons

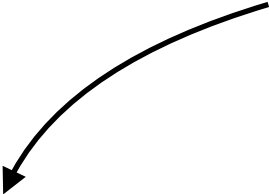
5 kb  
upstream

3 kb  
downstream

Allelic  
association







# Allelic association

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- Popoolation2<sup>1</sup>
  - Uses number of reads for each allele in each population
  - Downsampled the data
  - Fisher's exact test
  - Benjamini-Hochberg procedure to correct for multiple testing

rs12345	Ref (A)	Alt (T)
Healthy	200	120
Diseased	50	270

<sup>1</sup>Kofler et al., 2011. Bioinformatics 27, 3435–3436.

# Functional analysis and allelic association

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