





Functional analysis and allelic association

Variants

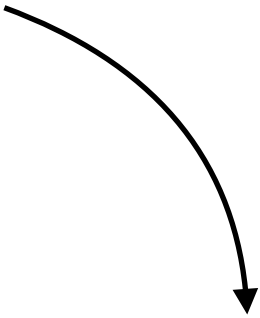
# Functional analysis

Exons

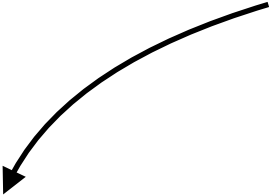
5 kb  
upstream

3 kb  
downstream

Allelic  
association







# Missense mutations

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- Polyphen2
  - Aligns homologues and identifies conserved AAs
  - 8 sequence based, 3 structure based features
  - Sens: 88 %
  - Spec: 62 %
- SIFT
  - Aligns homologues and identifies conserved AAs
  - Sens: 85 %
  - Spec: 69 %

# Functional analysis and allelic association

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