Integrative Annotation of Variants from 1092 Humans: Application to Cancer Genomics -
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Identifying Important Identifiers

Each of us has millions of sequence variations in our genomes. Signatures of purifying or negative selection should help identify which of those variations is functionally important. Khurana et al. (1235587) used sequence polymorphisms from 1092 humans across 14 populations to identify patterns of selection, especially in noncoding regulatory regions. Noncoding regions under very strong negative selection included binding sites of some chromatin and general transcription factors (TFs) and core motifs of some important TF families. Positive selection in TF binding sites tended to occur in network hub promoters. Many recurrent somatic cancer variants occurred in noncoding regulatory regions and thus might indicate mutations that drive cancer.

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