Characterization of mutational hotspots across cancer genomes

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Background

Data & Methods

Results

1) How many hotspots are there?

2) SNVs, MNVs and indels form hotspots at different frequencies

3) Top mutated hotspots are known drivers and variants of unknown significance

4) SNV hotspots are correlated with inactive and late replicating regions similarly as out-hotspot mutations

5) SNV hotspots are differentially distributed across genomic elements

6) Towards the identification of mutational processes generating hotspot mutations

Next steps

How do mutational signatures contribute to hotspots' formation? Which is the interplay between mesoscale chromatin features and hotspots? Can we prioritize candidate non-coding driver hotspots?