

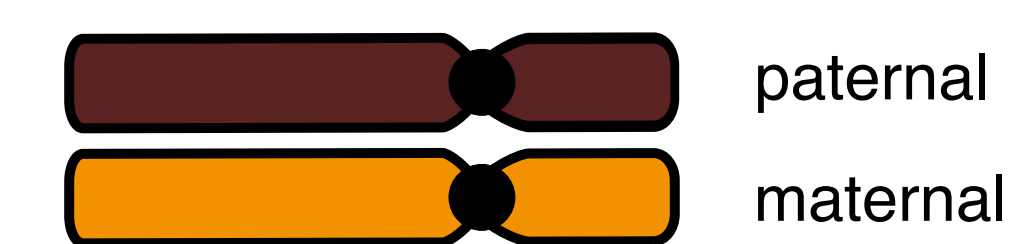
SPICE: Selection Patterns In somatic Copy-number Events



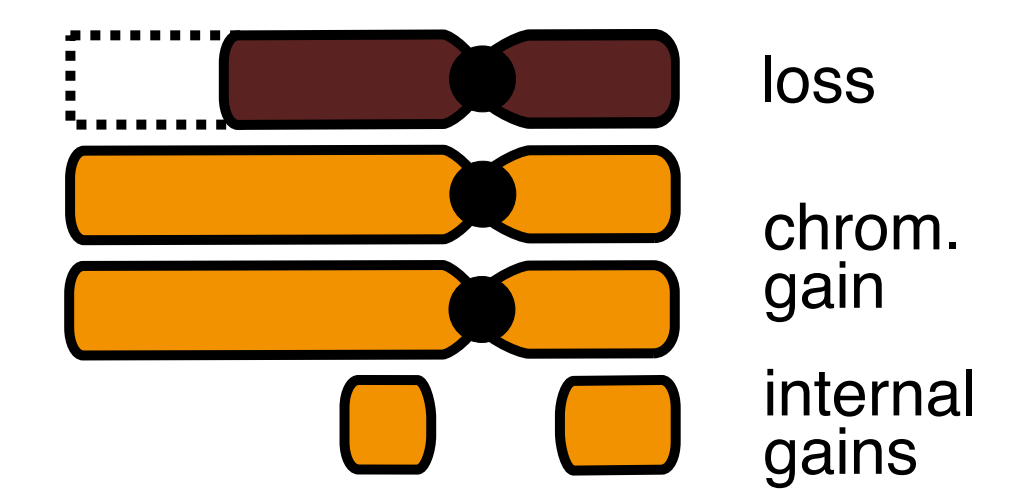
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1 Copy-number events

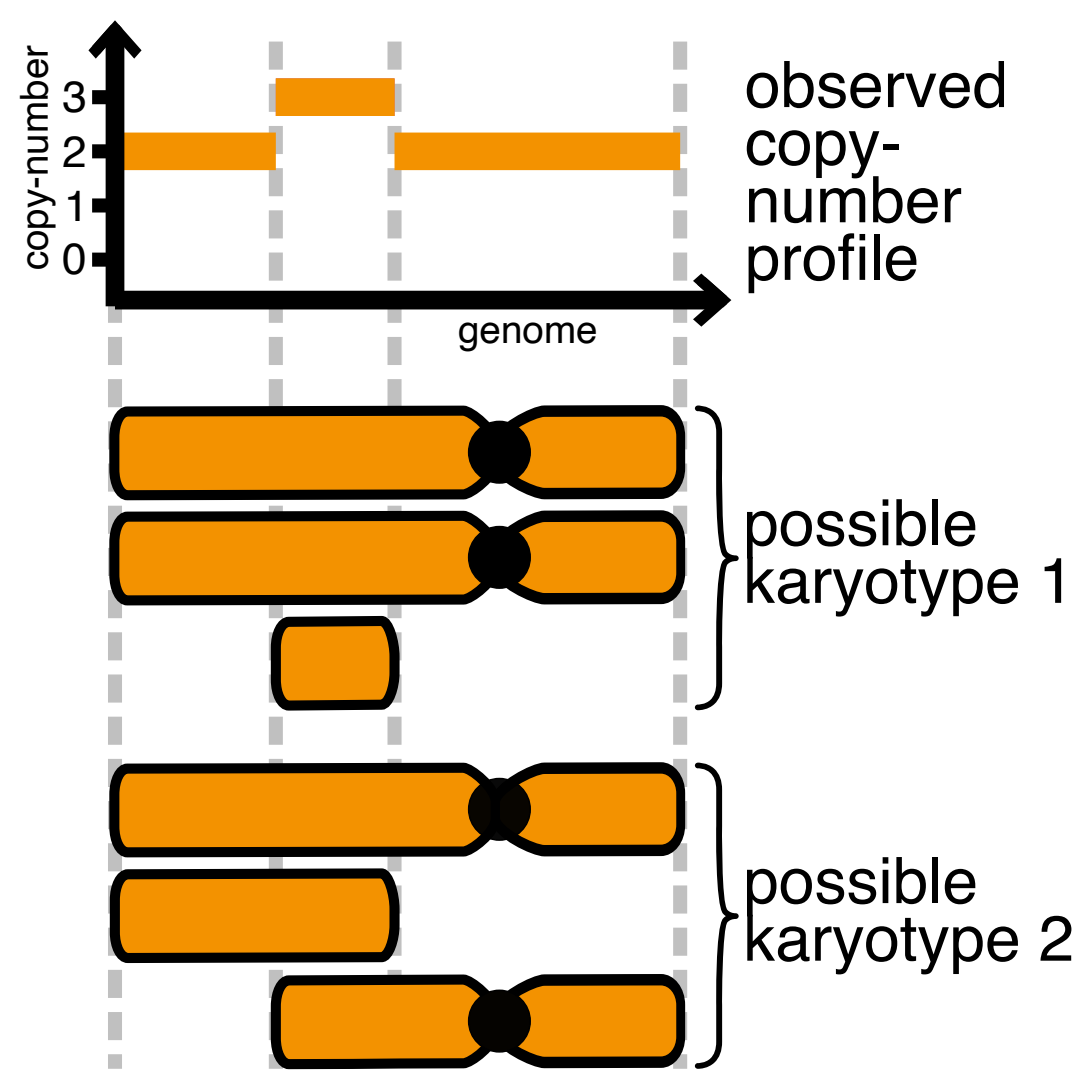
Healthy genome
2 copies per chromosome



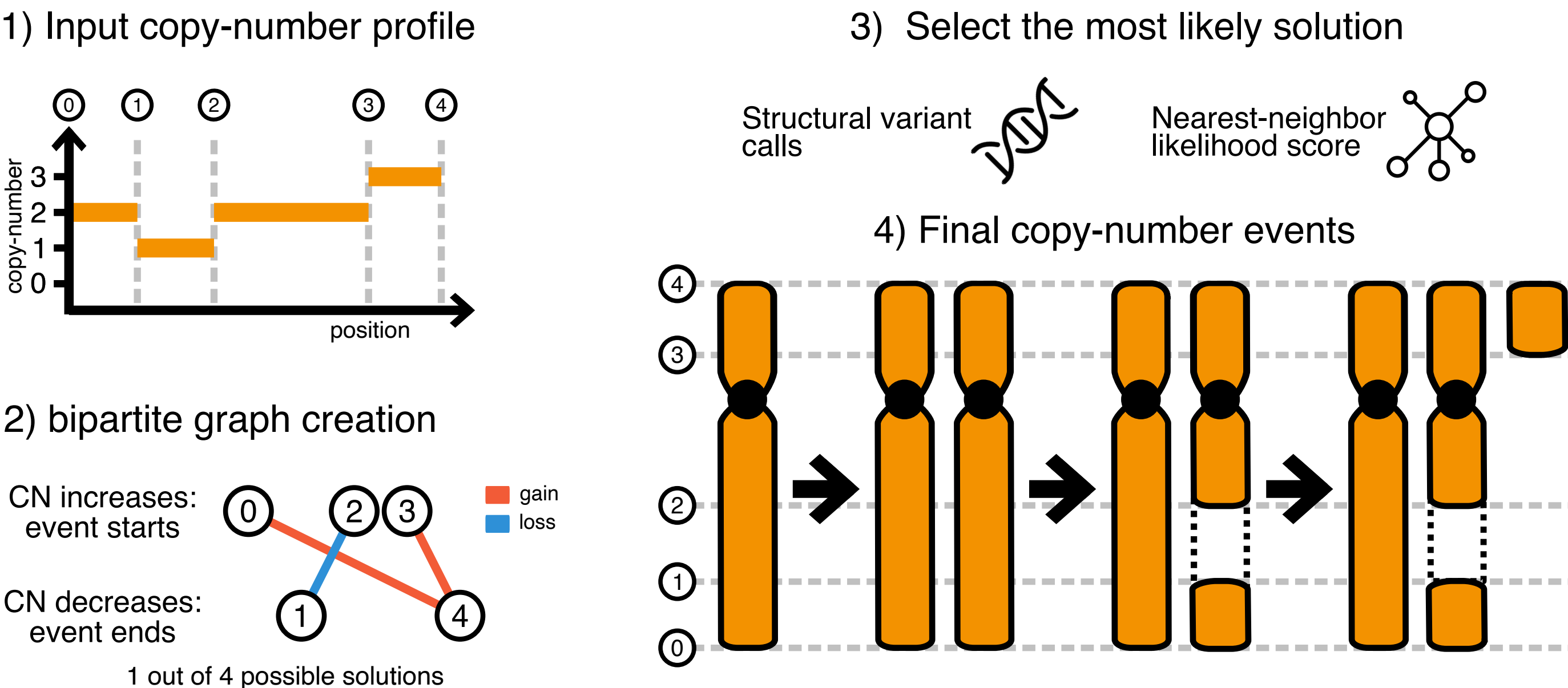
Cancerous genome
Complex copy-number changes



CN profiles are ambiguous



Inferring individual copy-number events

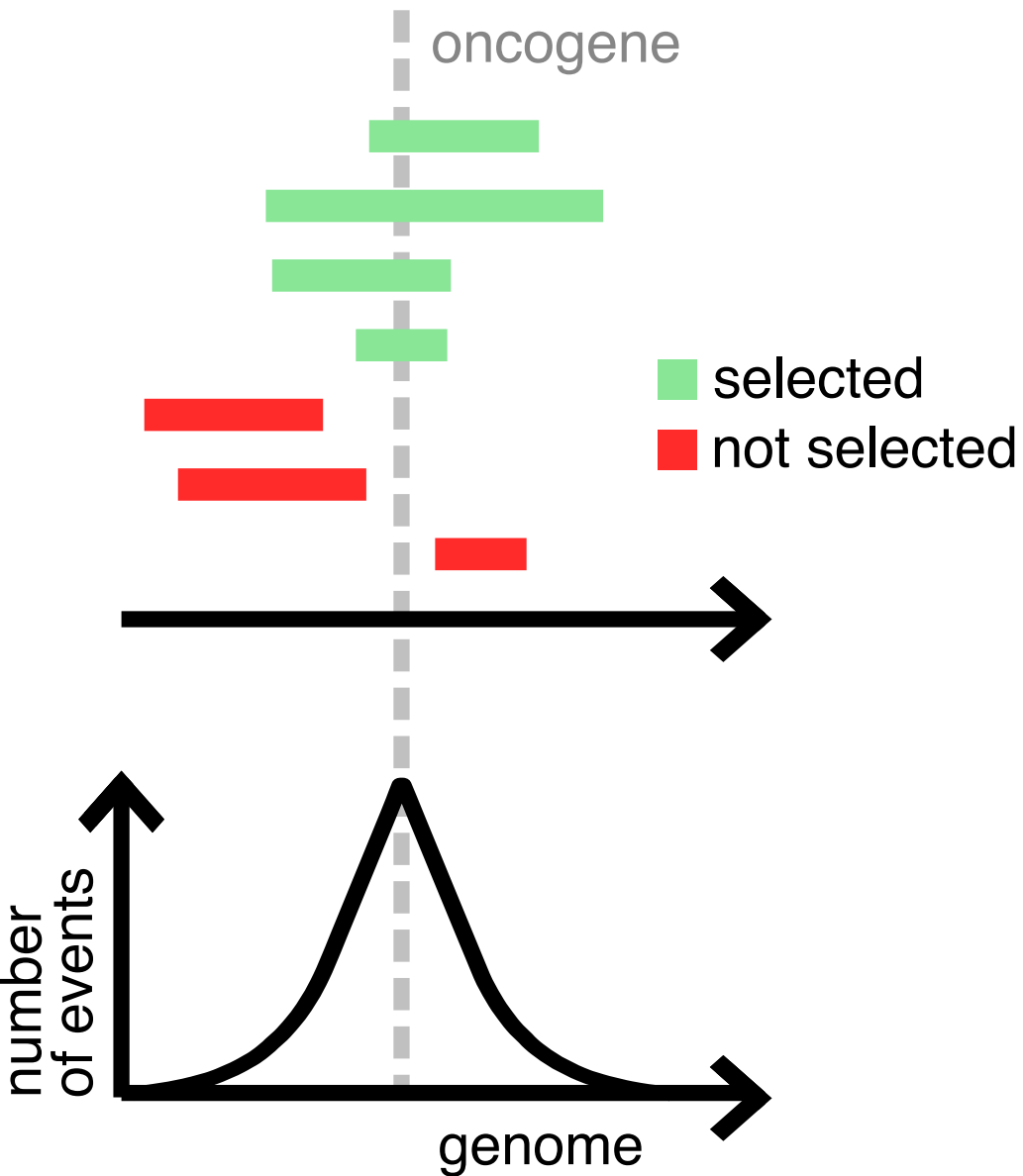


2 Loci of selection

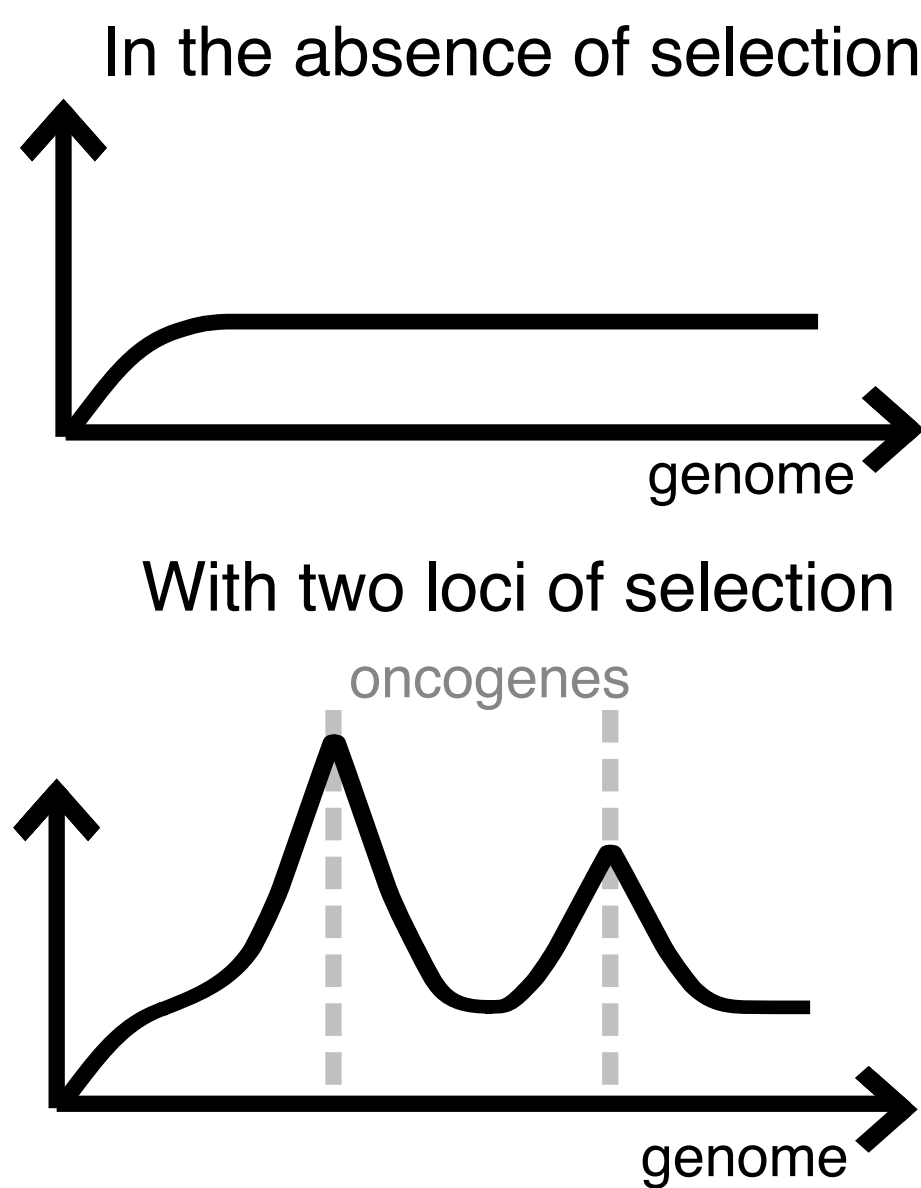
2 simple assumptions

- 1) Uniform breakpoint formation across the genome
- 2) Focal selection around loci that gain oncogenes and lose tumour-suppressors

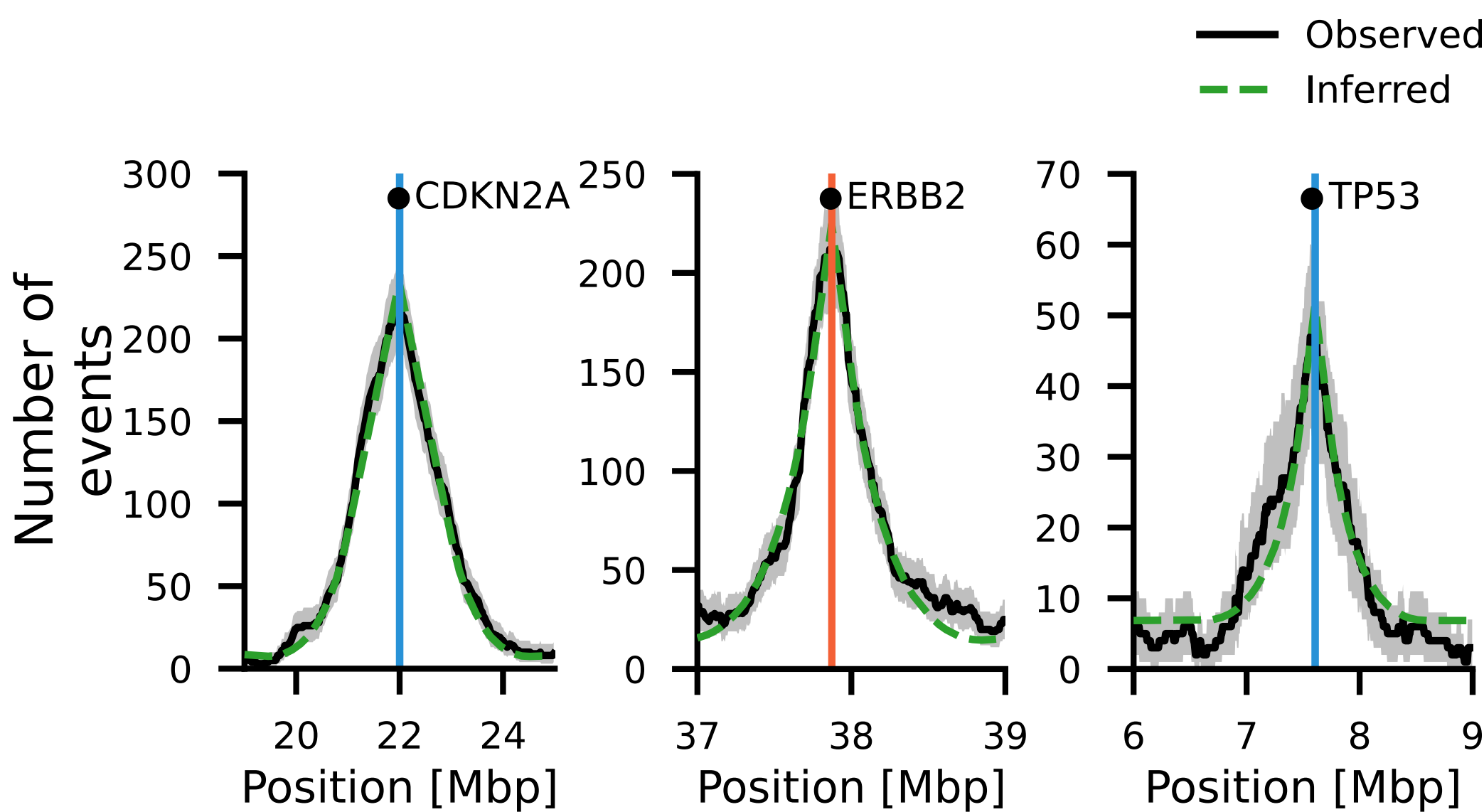
Focal selection



Generative model

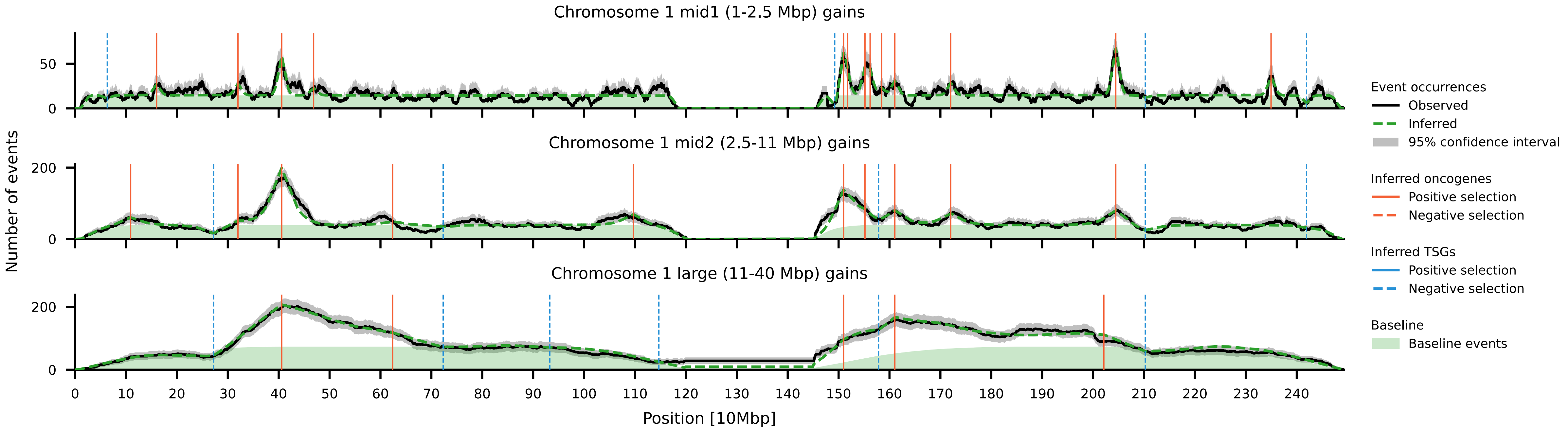


Remarkably good fit to data



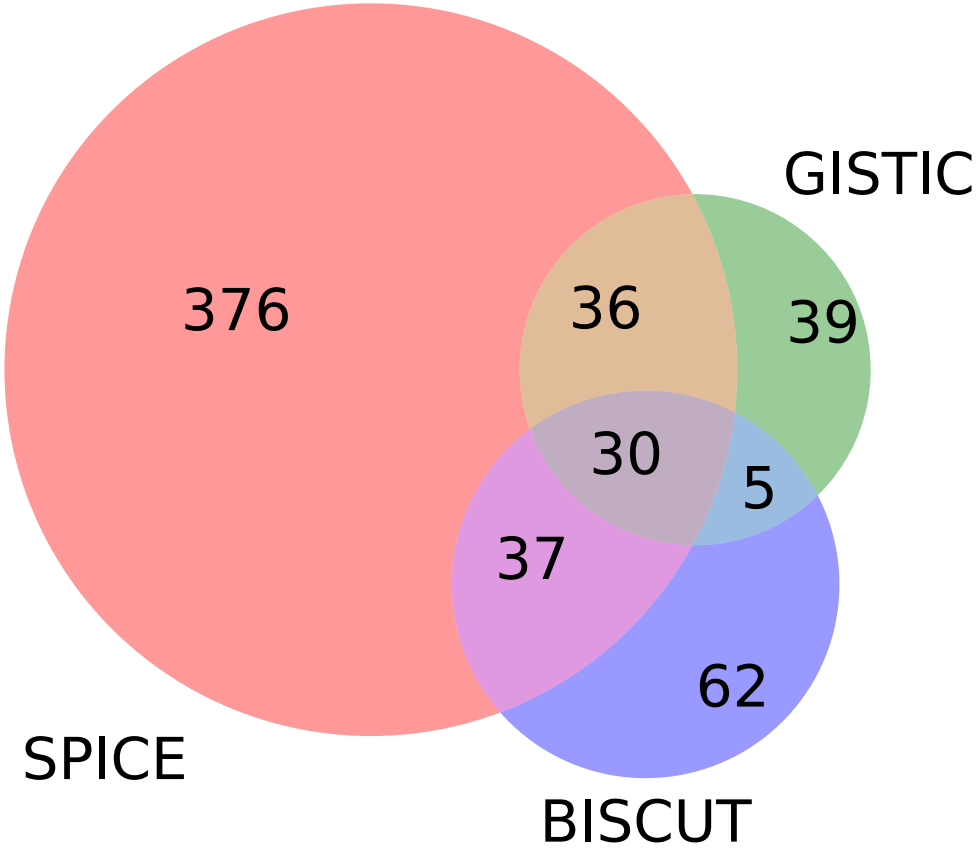
3 Results

89% of the genome is explained by our model (model prediction falls within 95% confidence interval of the data)



Comparison to previous methods

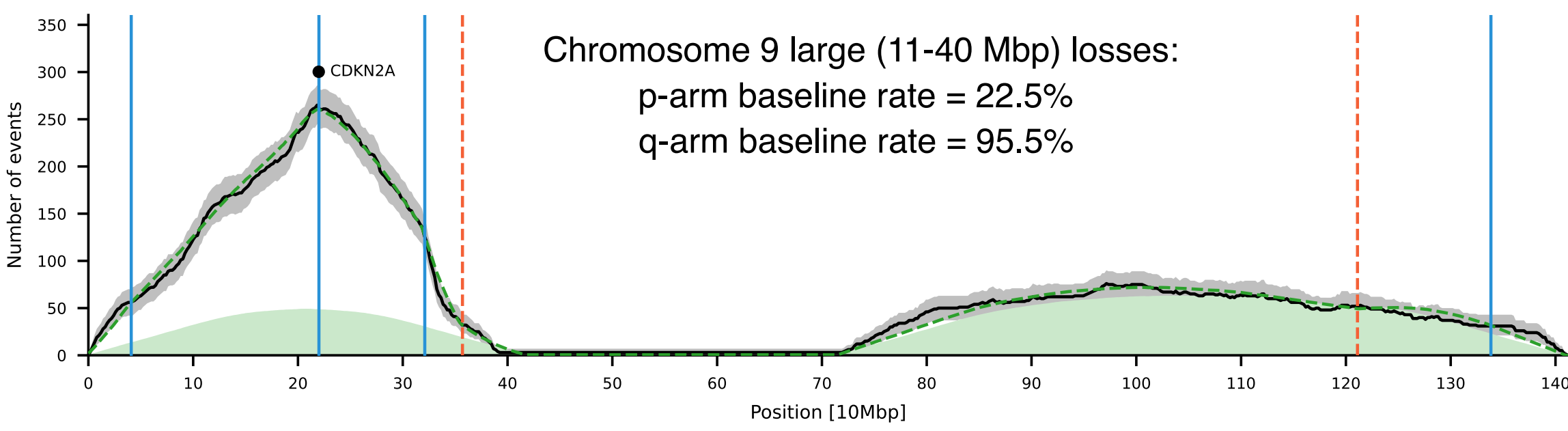
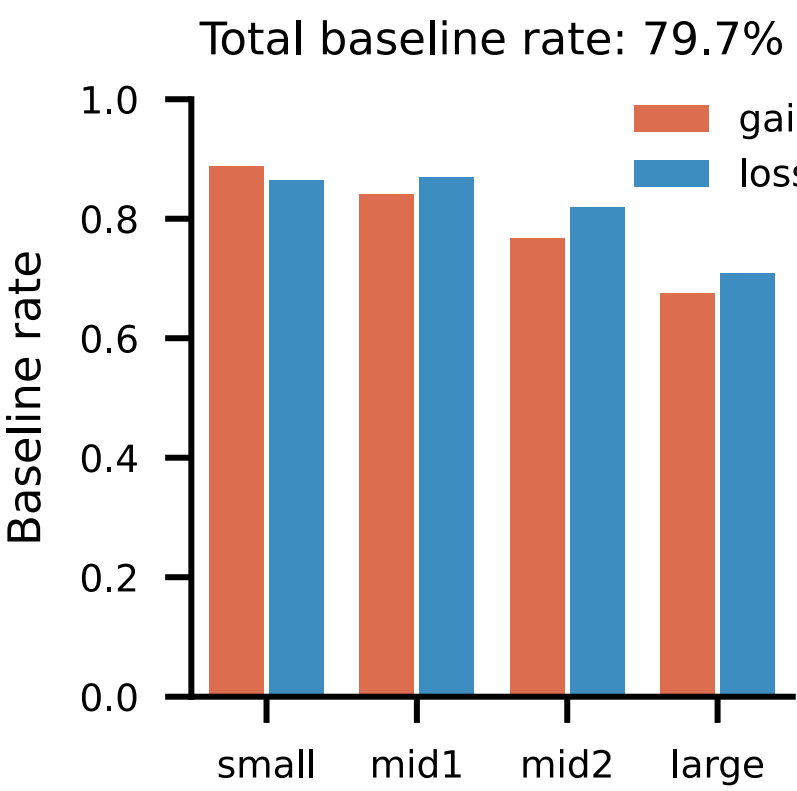
We recover most previously published loci of selection. We also find hundreds of novel loci. Novel loci are enriched for known oncogenes and tumour-suppressors.



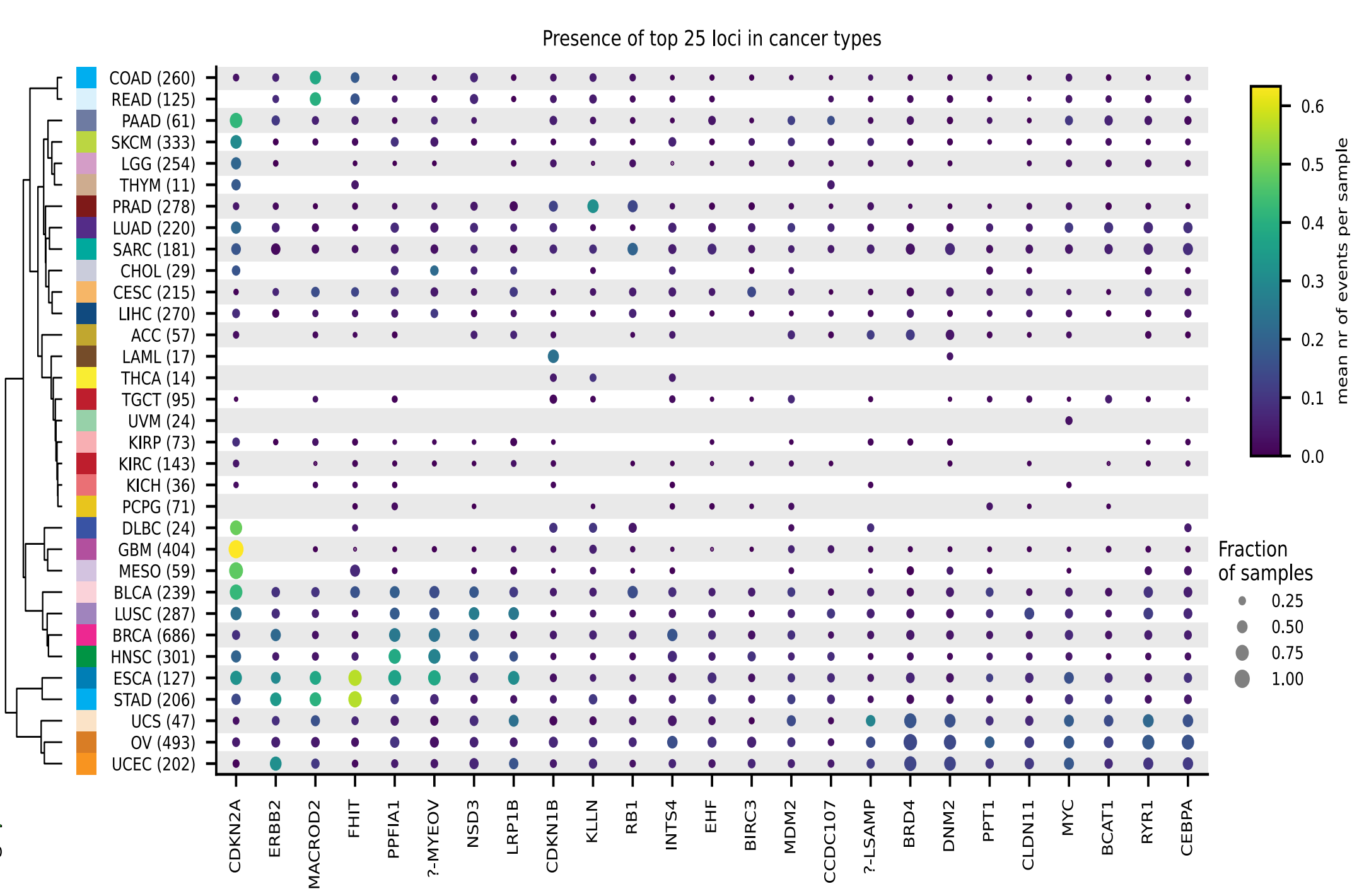
Baseline rate

79.9% of internal copy-number events are not subject to focal selection and likely have no effect on fitness.

Instead they are baseline events similar to passenger mutations.



Cancer-type specific occurrences



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