

• Visualization of Funseq2 output

- both BED and VCF formats
- 32 features in total
- Among which
 - GERP scores, noncoding features (Promoter, TFP, TMP, Intron, Enhancer, ncRNA, UTR, Pseudogene), mutation type (transition vs transversion), reference and variant genotype, strand +/- information
 - Top N (default N=50) target genes, MOTIFG, MOTIBR, hot regions
 - Variant distribution across and within chromosomes
- More features can be added by the user

• Three steps

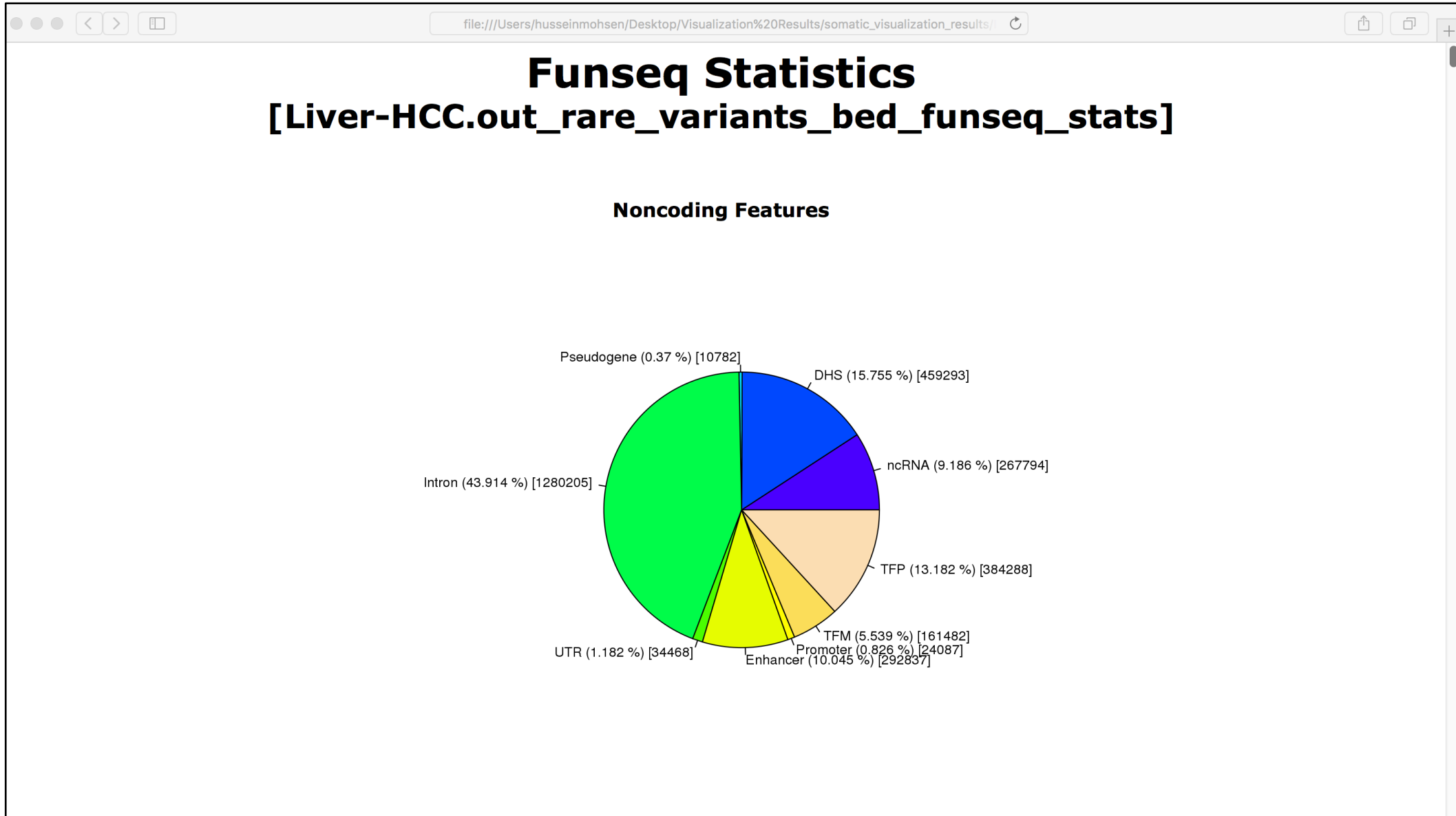
- Statistics
- Plot generation
 - Flexible *w.r.t.* types of plots
- HTML Generation

• Accepts additional parameters to qualify specific variants

- Up to two, unordered ones

```
[hm444@farnam2 ~]$ awk -v regex_args="Promoter" -f cancer-variant-correlation/scripts/funseq_stats.awk funseq_results/Output.vcf > Funseq2_stats_example.txt
```

Example



- Execution time
 - ~ 10 seconds for 100,000 variants
 - Grows linearly
- Tested on 200+ samples
- Part 1 to be followed by part 2 very soon
- What's next
 - Already generated Part 1 statistics for all PCAWG somatic and germline samples
 - 37 x 2 x 3 samples/files
 - To be presented with across-samples statistics as part of the two-hit hypothesis project