## Variants Statistics in Beacon

## 1 General Statistics

Statistics are calculated using Query by region -by names/aliases and Filters.

## 1.1 Stats on sequences (maybe table only, already in help page?)

- 1. Number of runs: total runIds
- 2. Number of runs with variants (we are keeping WT?)
- 3. Number of unique sequence/haplotypes: ?

```
option Split by sequencing technology (platform): Illumina, Nanopore (for now only Illumina)
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```
option Split by sequence database: SRA, GISIAD, GWH
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option Split by Individual geographic region: Australia, China, USA, Singapur, etc

option Split by Biosample collection date (month)

option Split by Biosample sample.type

option Split by Individual sex

## 1.2 Stats on variants

- 1. Number of genomic positions with variants: 28073/ Number of genomic positions: 29903
- 2. Variance per position: fig Needle plot (counts of unique sequences/haplotypes or counts of runs having them?)
- 3. Number of unique variants in database: 34951
- 4. Graph: Number of unique variants, split by options, e.g fig 1

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option Split by sequencing technology (platform): Illumina, Nanopore (for now only Illumina)
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option Split by variant frequency (quartiles)

option Split by variant type field: SNP, indels (although for now there are only SNPs?)

option Split by genomic region field: coding: all with genomic region=CODING, non-coding: the rest

 $option \ \ Split \ by \ molecular \ consequence \ (grouped: \ SYN: SILENT, NON-SYN: MISSENSE+NONSENSE, \\ NONCODING: the \ rest)$ 

option Split by Biosample sample.type

option Split by Individual sex

option See Per region Statistics: Distribution in genomic regions: non-coding, gene, cds/mature peptide, stem loops. Number of unique variants are aggregated in regions, show also split by syn/non syn, as in fig 2.

- 5. Number of positions with aminoacid substitutions in database/ Number of coding positions
- 6. Number of variants producing unique aminoacid substitutions in database: 18308

option See Per region Statistics: Distribution in genomic regions: non-coding, gene, cds/mature peptide, stem loops. Number of unique aminoacid substitutions (aminoacid change) (eg. "G507C") are aggregated in regions

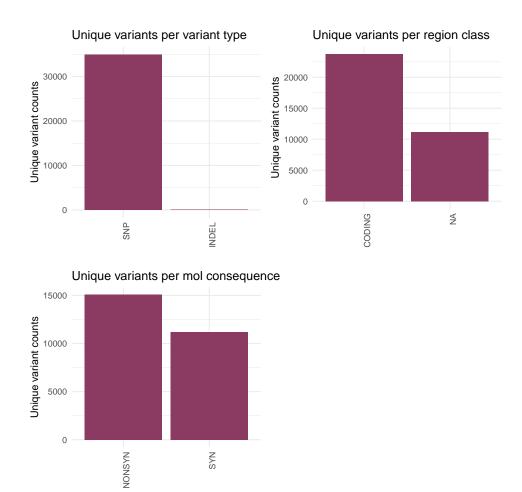
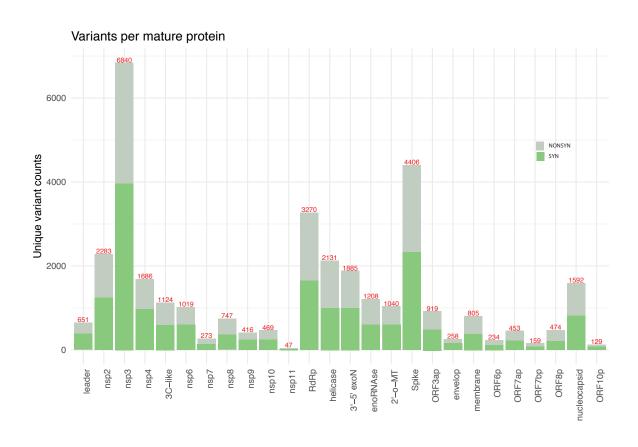


Figure 1: General statistics: Number of unique variants split by variant type, region class and mol consequence



 $\label{eq:sars-cov2} \mbox{Figure 2: Unique variants in coding regions of SARS-CoV2, shown per mature proteins, shown on clicking cds/mature peptide.}$