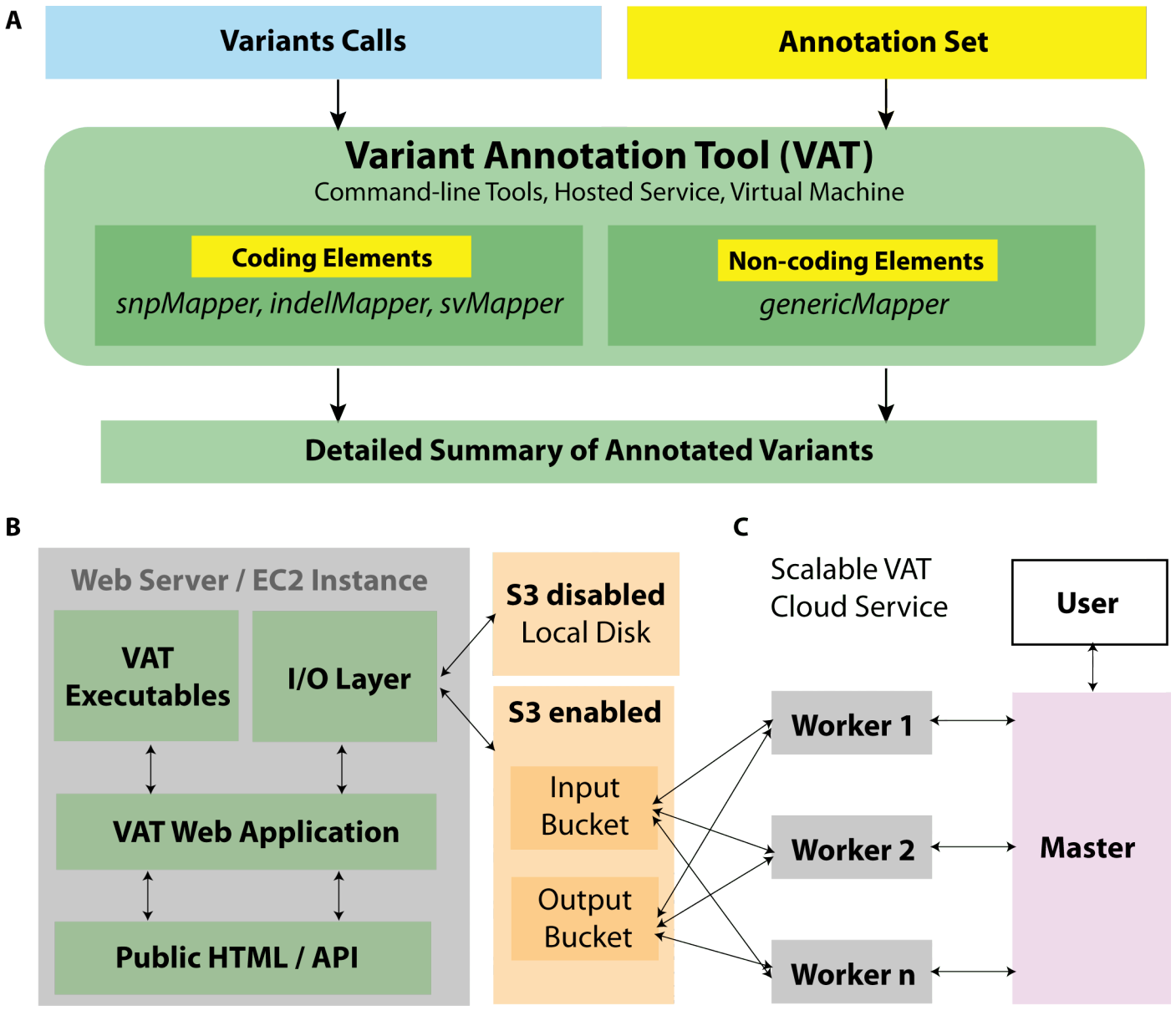
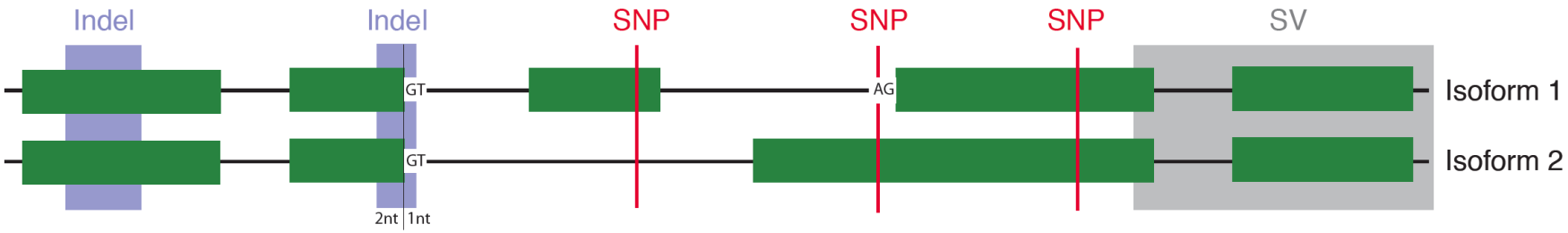
VAT Figures

11/30/11

LH


A
Variants Calls
Annotation Set
Variant Annotation Tool (VAT)
Command-line Tools, Hosted Service, Virtual Machine
Coding Elements
snpMapper, indelMapper, svMapper
Non-coding Elements
genericMapper
Detailed Summary of Annotated Variants
B
Web Server / EC2 Instance
VAT Executables
I/O Layer
VAT Web Application
Public HTML / API
S3 disabled
Local Disk
S3 enabled
Input Bucket
Output Bucket
C
Scalable VAT
Cloud Service
User
Worker 1
Worker 2
Worker n
Master



6nt non-frameshift indel

3nt indel partially overlapping an exon: causes a frameshift due to a 2nt exon overlap and disrupts the splice site

SNP affecting only one transcript

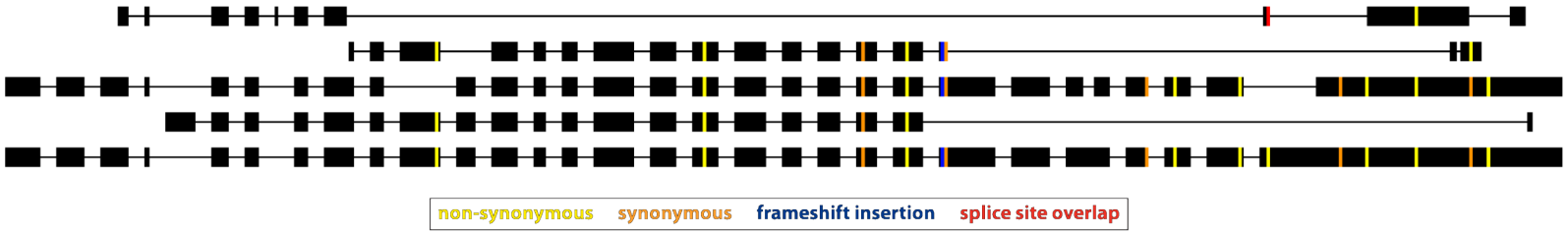
SNP affecting two transcripts in different ways: splice site disruption vs. coding SNP

SNP affecting both transcripts

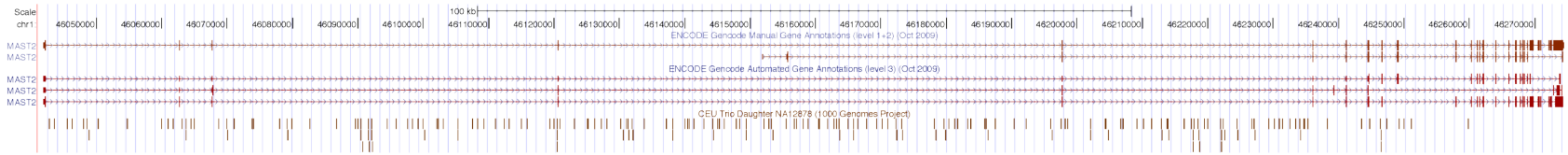
SV truncating more than one exon

VAT

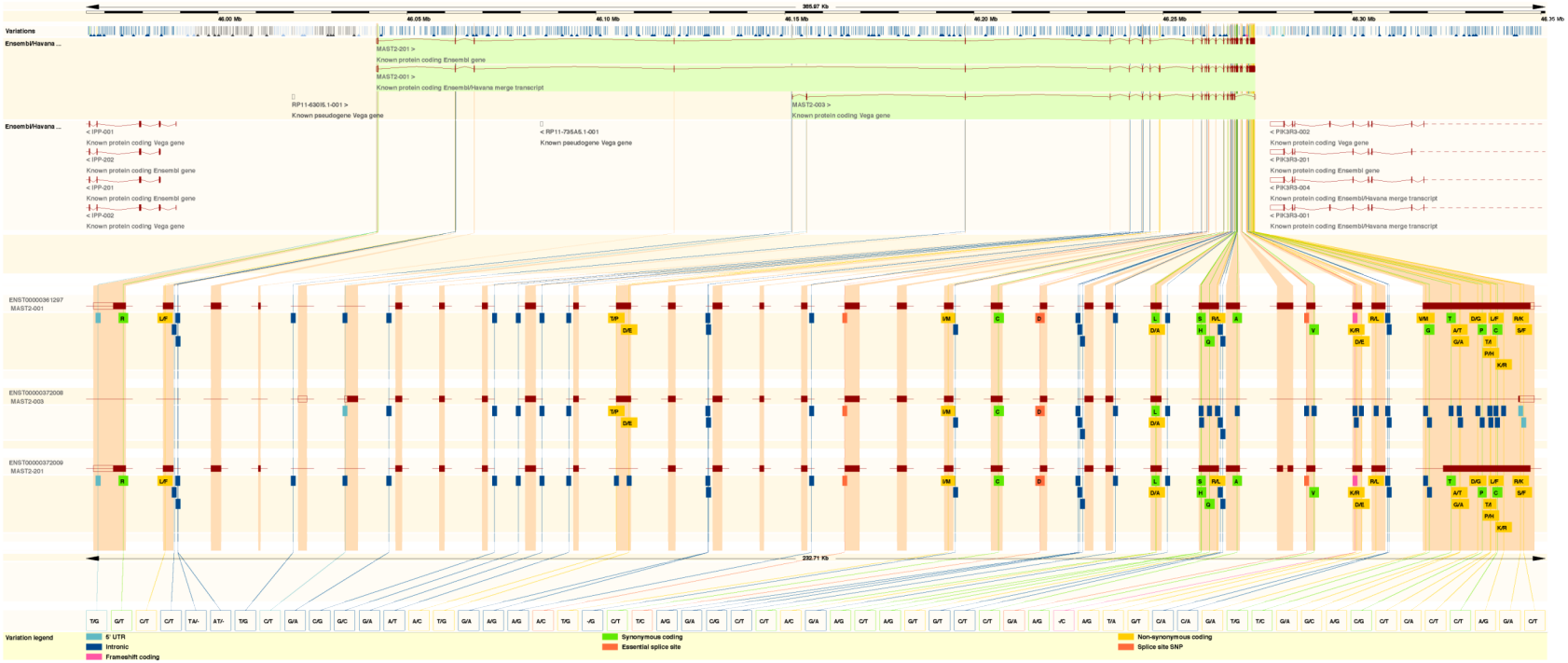
MAST2



UCSC genome browser



Ensembl genome browser



Comments

No 1000 Genomes VCF files on Amazon S3

TREAT: A Bioinformatics Tool for Variant Annotations and Visualizations in Targeted and Exome Sequencing Data

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Abstract

Summary: TREAT (Targeted RE-sequencing Annotation Tool) is a tool for facile navigation and mining of the variants from both targeted re-sequencing and whole exome sequencing. It provides a rich integration of publicly available as well as in-house developed annotations and visualizations for variants, variant-hosting genes, and host-gene pathways.

Implementation and Availability: TREAT is freely available to non-commercial users as either a stand-alone annotation and visualization tool, or as a comprehensive workflow integrating sequencing alignment and variant calling. The executables, instructions, and the Amazon Cloud Images of TREAT can be downloaded at the website: http://ndc.mayo.edu/mayo/research/biostat/stand-alone-packages.cfm