Varant:
An Open Source **Var**iant **An**notation **T**ool

Kunal Kundu[1]

Uma Sunderam[1], Steven E. Brenner[2], Rajgopal Srinivasan[1]

[1]Tata Consultancy Services Ltd, Innovation Labs, Hyderabad, India.
[2]Department of Plant and Microbial Biology, University of California, Berkeley

Why we developed this tool ?

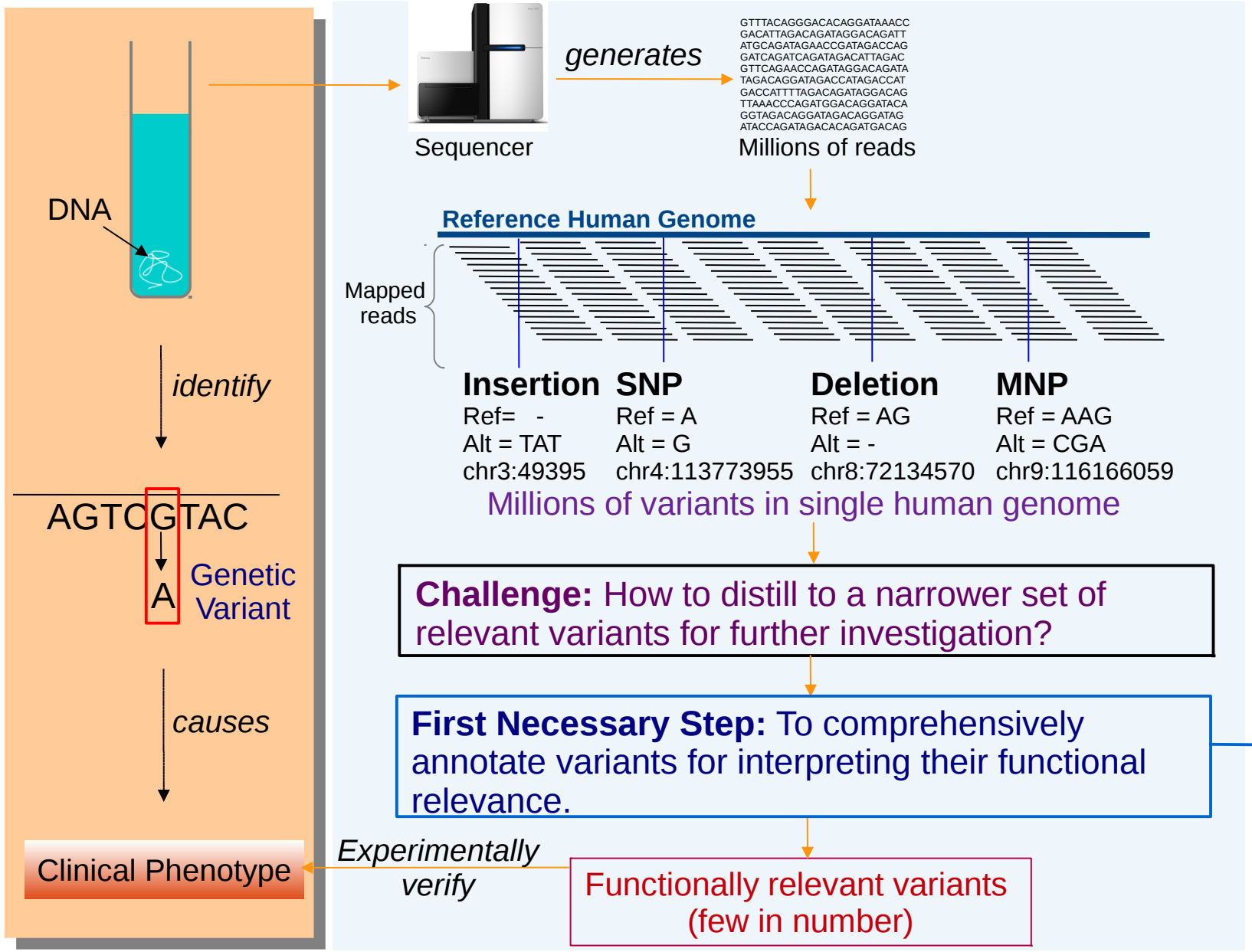
How does this tool work ?

What is the accurracy of this tool ?

Motivation

Why we developed this tool ?

AIM in clinical study

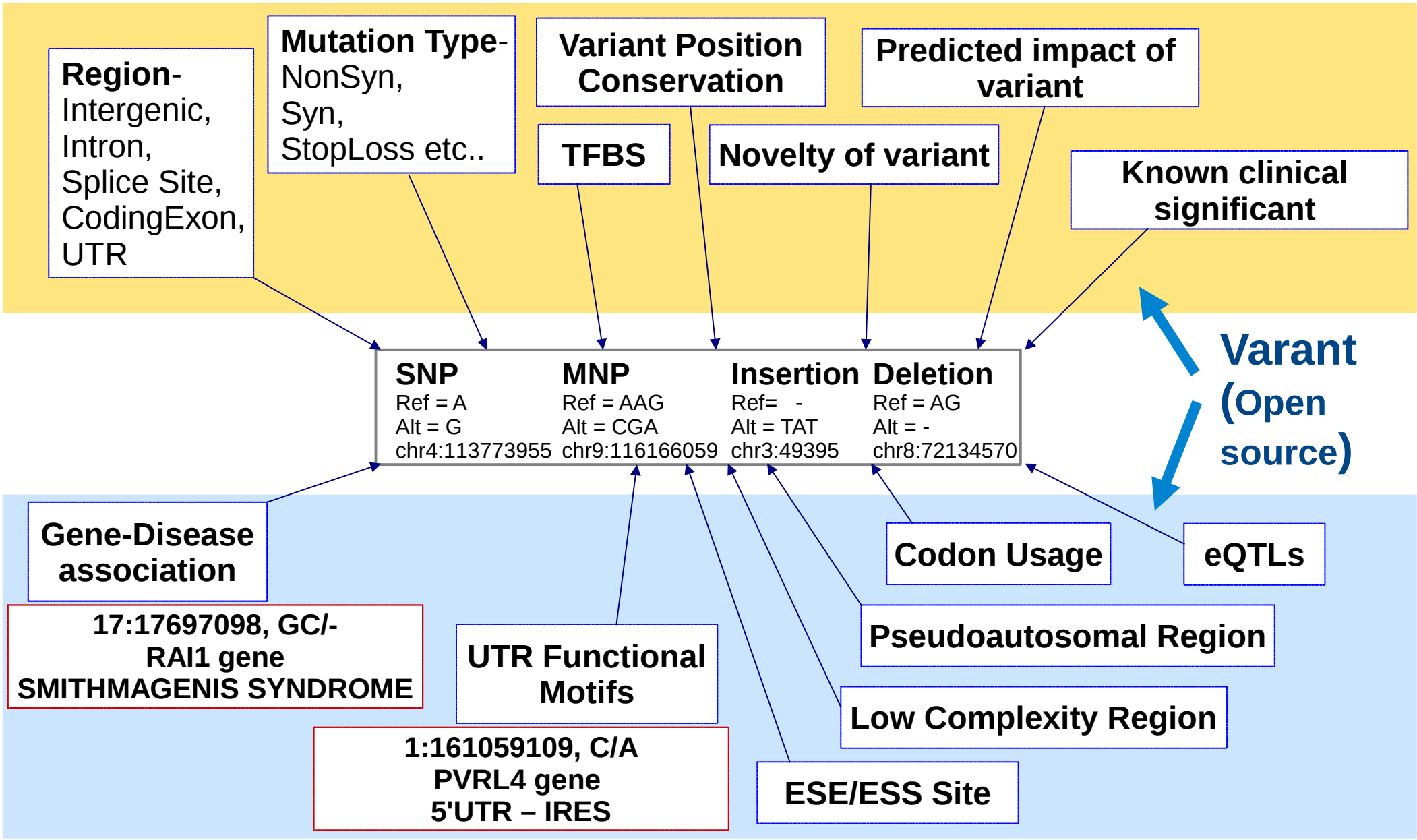


With this outlook we have developed a **comprehensive** and **extensible** **open source** tool for human genome variation annotation called **Varant.**

Why we developed this tool ?

Annotation Types

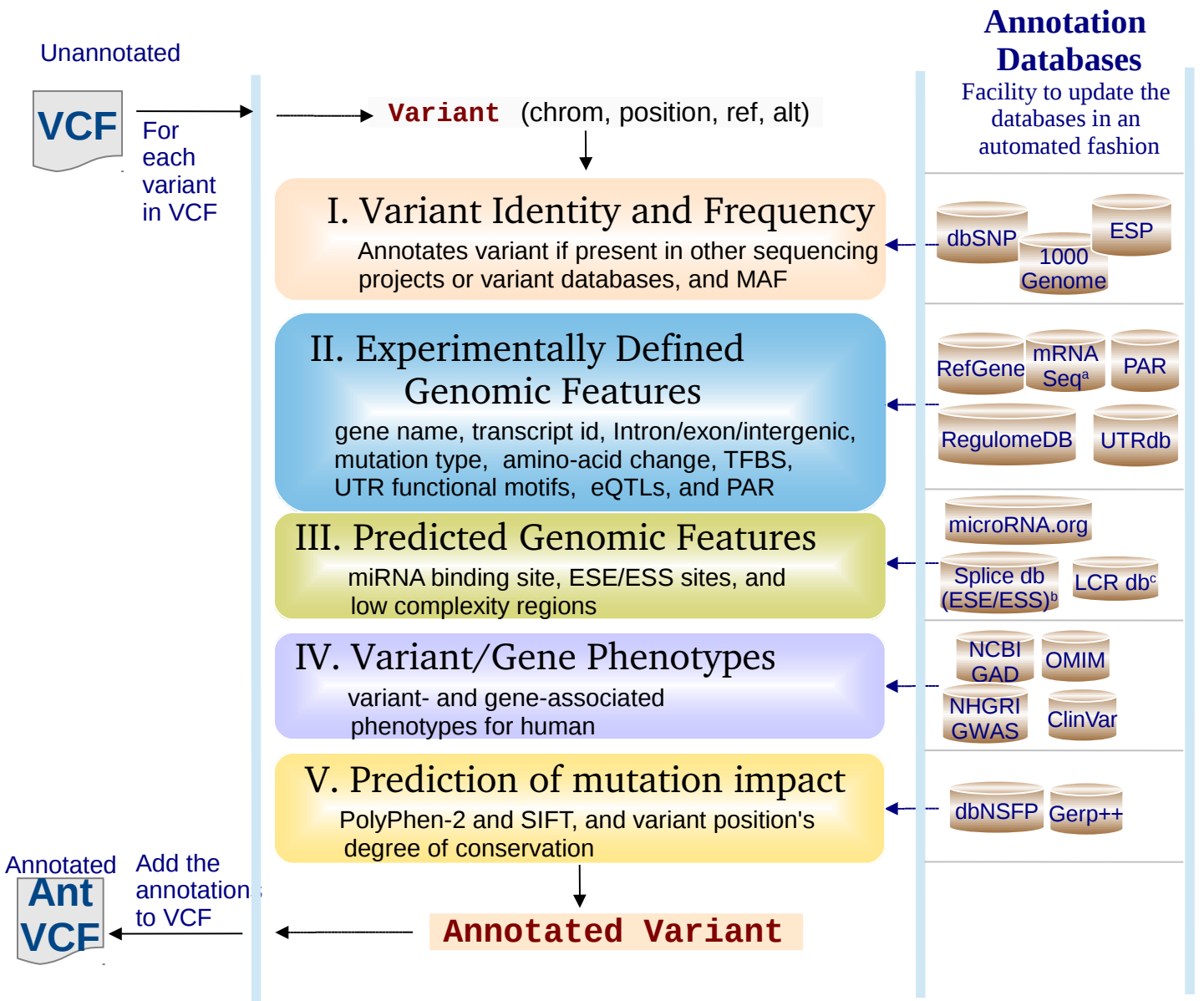
Annovar (Commercial), SnpEff (Open source)



Installation and Working

How does this tool work ?

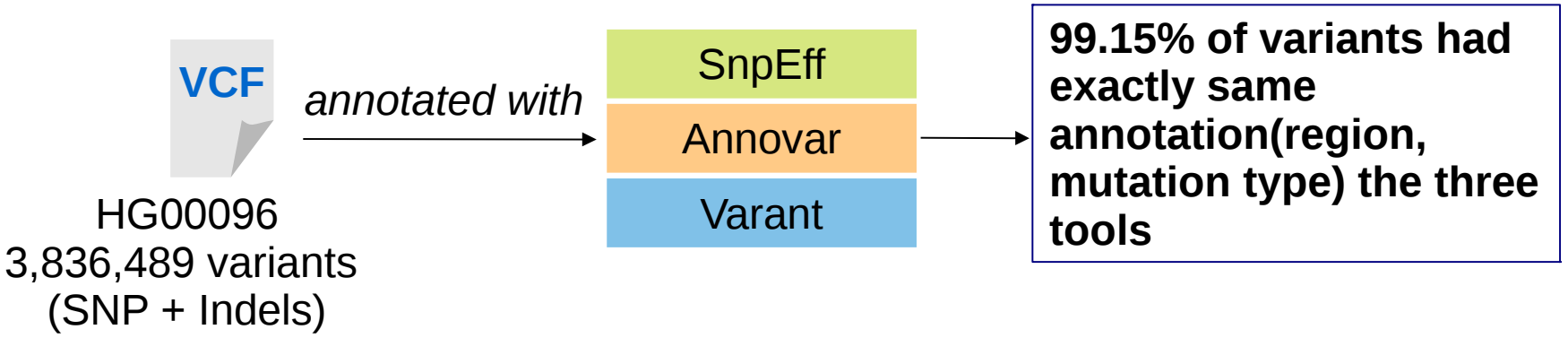
1. Download data sources (Varant depends upon 17 data sources) → **2.** Create SQLite databases → **3.** Set path to locations of data sources and their SQLite databases



[a]Computed from refgene definition and using reference genome.
[b]Computed using Burge et al. hexamer data on ESS/ESE.
[c]Computed using the mdust Program (http://bit.ly/mdust-LC)

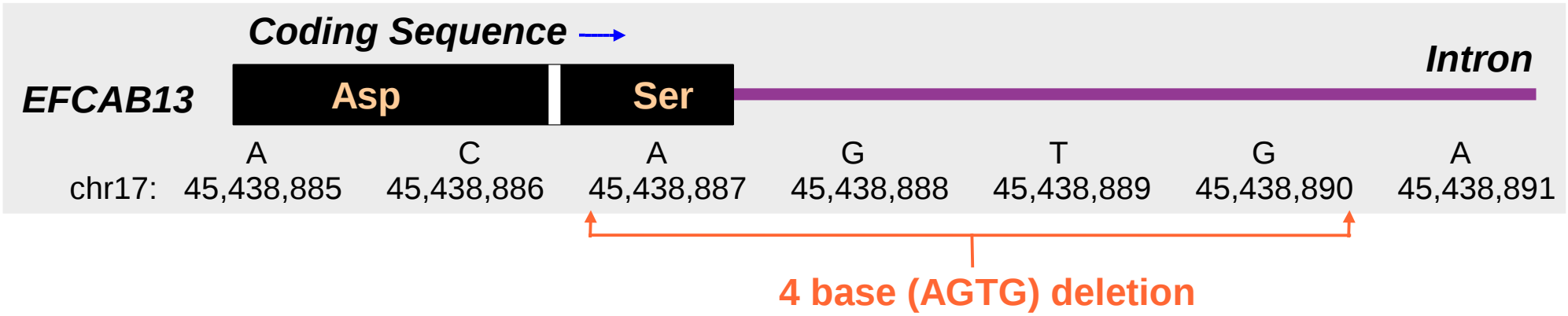
Annotation Accuracy

What is the accurracy of this tool ?



When Varant disagrees with other methods, its predictions are superior

Annotation of a deletion variant that spans intron exon boundary



SnpEff – splicing
Annovar – exonic;frameshift
Varant – intron-exon boundary;splicing (*better precision*)

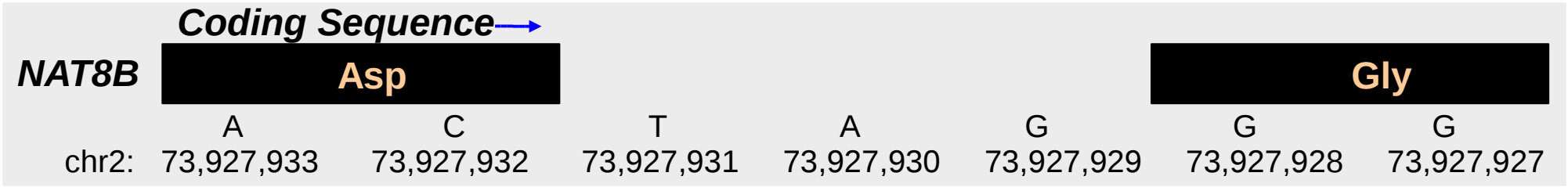
Annotation Accuracy

What is the accurracy of this tool ?

Annotation of Indels that do not alter coding sequence

SnpEff – frameshift (incorrect)
Annovar – frameshift (incorrect)
Varant – NoCDSChange (correct)

Annotation of variants that alters stop codon in CDS



SnpEff – StopLoss
Annovar – exonic (mutation type not computed)
Varant – Ambiguous_StopLoss (better precision)

Summary

- Varant provides a broad range of annotations for interpreting the relevance of genetic variants.
- Varant is easily deployed.
- Varant is accurate: annotations are equal or better than other commonly used tools.
- Varant is open source.

Acknowledge

Rajgopal Srinivasan, PhD
Head, TCS Applications Research Council,
TCS Innovation Labs
Hyderabad, India

Steven E. Brenner, PhD
Professor,
Department of Plant and Microbial Biology,
UC, Berkeley

Thank You...