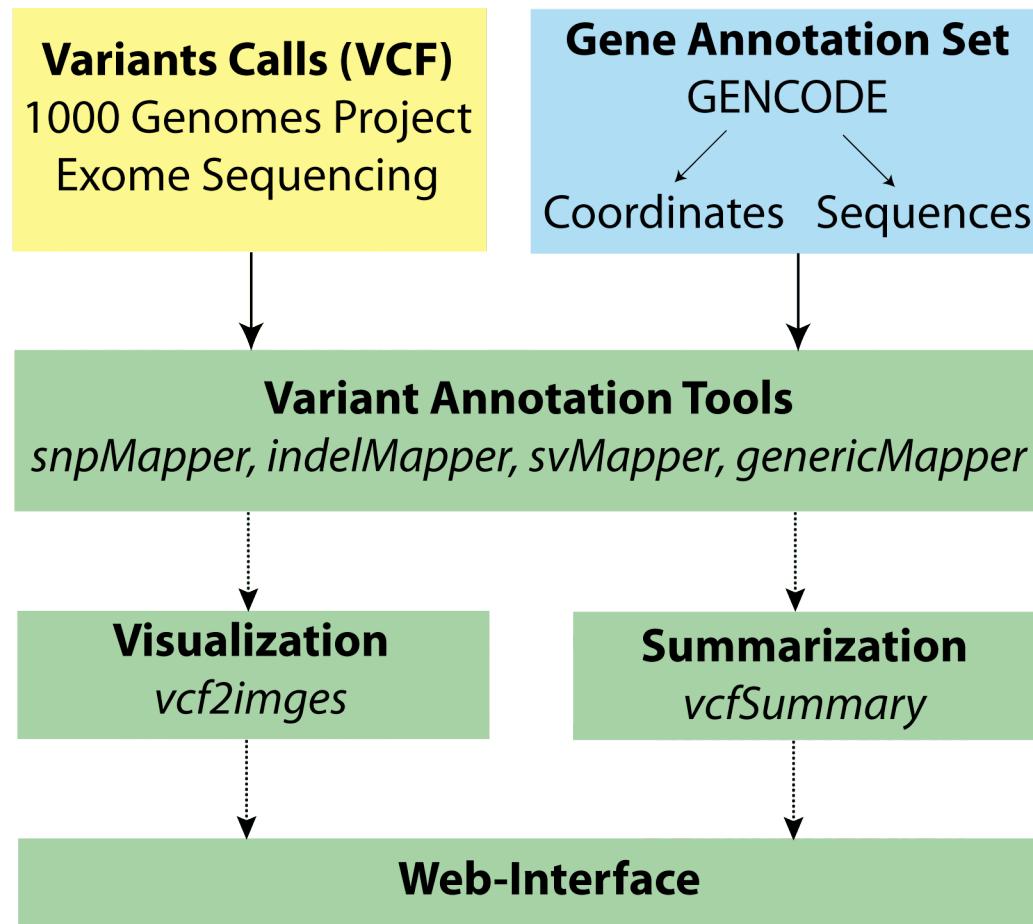


VAT paper update

Paper Outline

- Abstract
- Introduction
- Main
 - Rationale
 - Description of modules (workflow; Fig 1)
 - Case study (application of VAT to 1000 genomes pilot data)
 - Comparison between methods (Table)
 - Features
 - Performance
 - Discussion and Conclusion

VAT Workflow



Comparison between variant annotation tools

		VAT	ANNOVAR	Ensembl Variant Effect Predictor	PICMI	SeqAnt
Features						
Variant types	SNPs	y	y	y	y	y
	Indels	y	y	y	n	y
	SVs	y	n	n	n	n
Input formats		VCF, Complete Genomics	VCF, Complete Genomics, PileUp, others	VCF, PileUp, others	VCF, others	Multiple input files required; no VCF support
Output formats		VCF, tabular	tabular	tabular	tabular	tabular
Organisms		Mainly human; simple to add other organisms	Human; other genomes available	Human; many others supported	Human; other genomes available	Human; other genomes available
Annotation sets		GENCODE; simple to add other annotation sets	UCSC Genes, RefSeq, Ensembl, others	Ensembl	Ensembl	RefSeq
Transcript based annotations		y	y	y	y	n
Sample summaries		y	n	n	n	n
Gene summaries		y	n	n	n	n
Graphical visualization		y	n	n	n	n
Mode	Command-line	y	y	n	n	n
	Web-interface	y	n	y	y	y
Assessment of protein function	SIFT	n	y	y	n	n
	PolyPhen	n	y	y	n	n
Performance (command-line version)				N/A	N/A	N/A