Enhancing dbSNP and ALFA for Genomic Research

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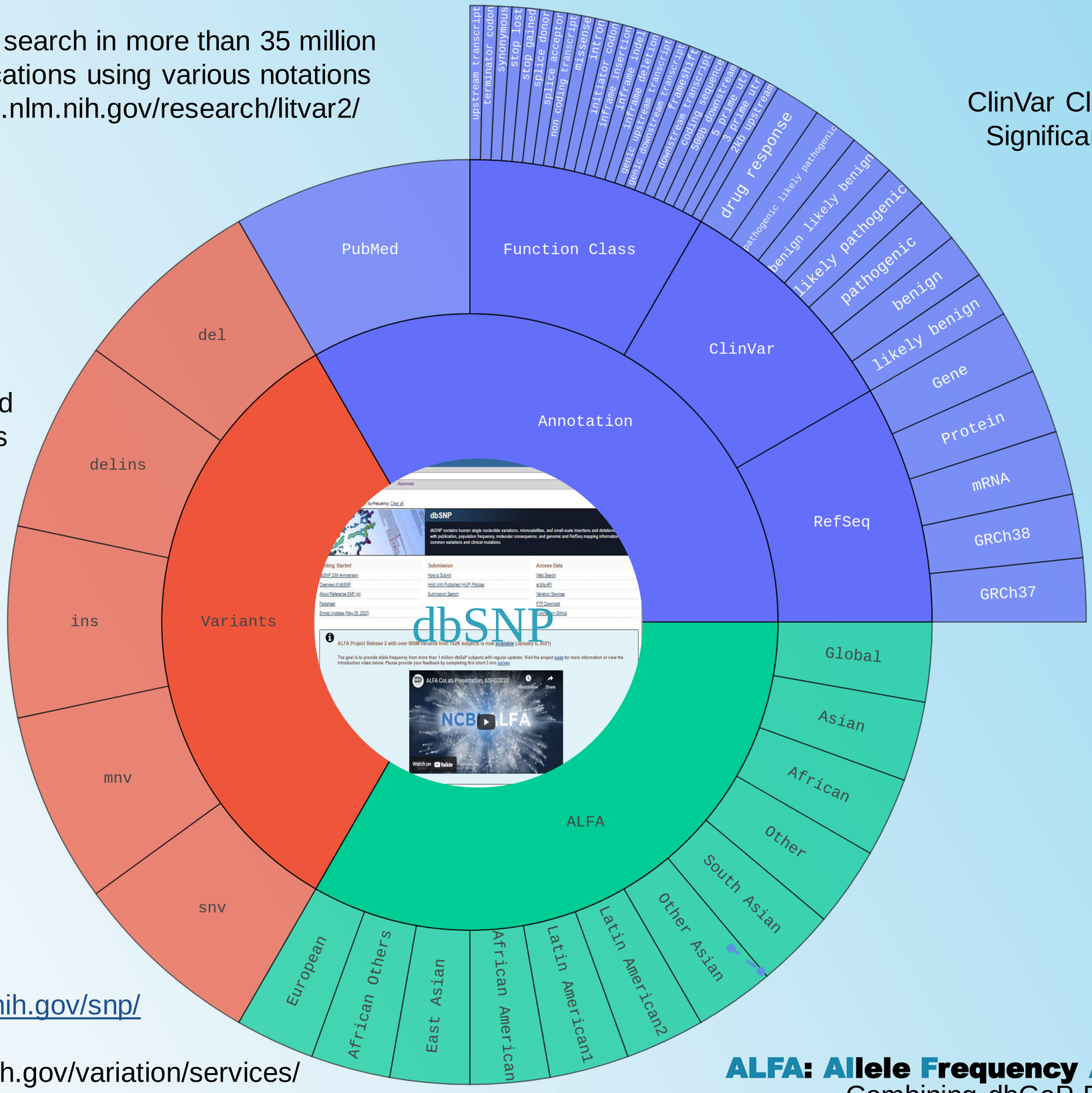
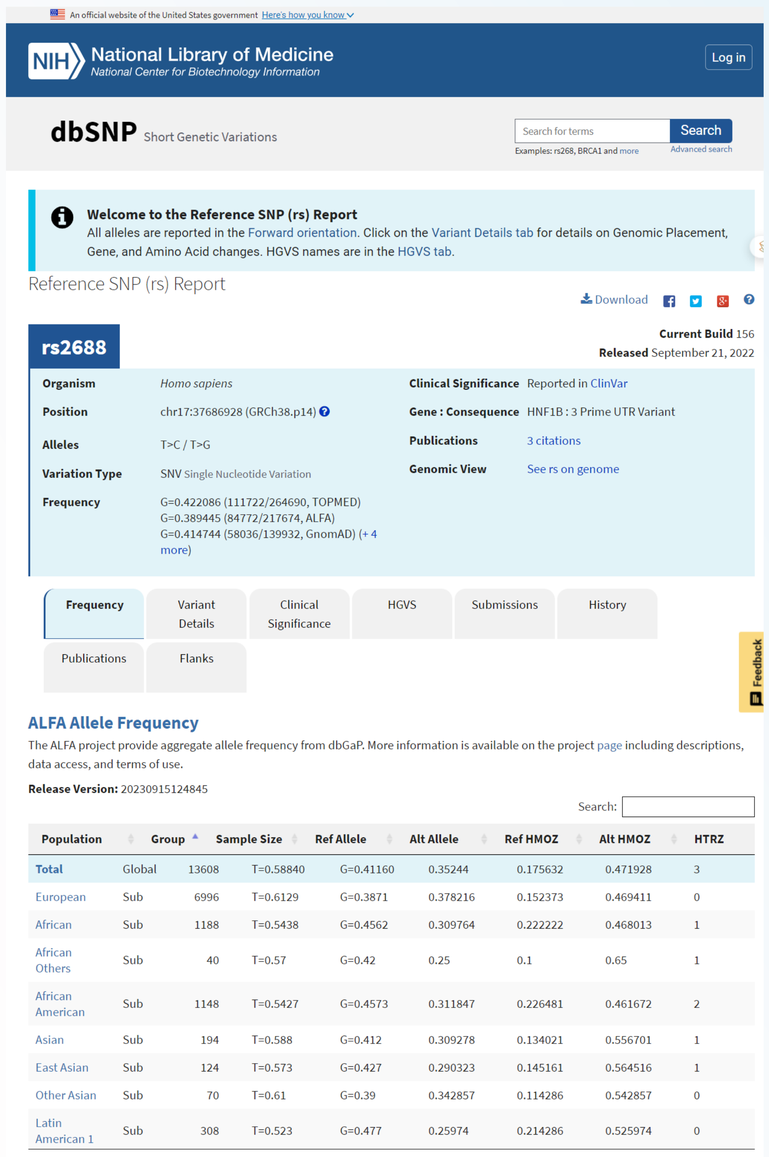
LitVar2 - variant search in more than 35 million biomedical publications using various notations
https://www.ncbi.nlm.nih.gov/research/litvar2/



- >3 billion variants aggregated from thousands of submitters world-wide
- 1.1 Billion unique reference variants (RefSNP)
- 95% RefSNP will population MAF
- Includes common and rare variants and ClinVar mutations

Data Access

Web: https://www.ncbi.nlm.nih.gov/snp/
FTP: ftp.ncbi.nih.gov/snp/
API: https://www.ncbi.nlm.nih.gov/variation/services/



Population	Group	Sample Size	Ref Allele	Alt Allele	Ref HMOZ	Alt HMOZ	HTRZ
Total	Global	13608	T=0.58840	G=0.41160	0.35244	0.175632	0.471928	3
European	Sub	6996	T=0.6129	G=0.3871	0.378216	0.152373	0.469411	0
African	Sub	1188	T=0.5438	G=0.4562	0.309764	0.222222	0.468013	1
African Others	Sub	40	T=0.57	G=0.42	0.25	0.1	0.65	1
African American	Sub	1148	T=0.5427	G=0.4573	0.311847	0.226481	0.461672	2
Asian	Sub	194	T=0.588	G=0.412	0.309278	0.134021	0.556701	1
East Asian	Sub	124	T=0.573	G=0.427	0.290323	0.145161	0.564516	1
Other Asian	Sub	70	T=0.61	G=0.39	0.342857	0.114286	0.542857	0
Latin American 1	Sub	308	T=0.523	G=0.477	0.25974	0.214286	0.525974	0

ALFA: Allele Frequency Aggregator

Combining dbGaP Data

dbGaP studies
Millions of subjects
Billions of variants
Trillions of genotypes

ALFA pipeline
Release 3:
Aggregated >5 Trillion genotypes from ~204K subjects
Computed frequency for >904 Million Variants
Minor Allele and Genotype Frequencies for 12 ancestry populations

Open-Access dbSNP
RefSNP
Variation Services API
Trackhubs and FTP Downloads

More coming soon!!!

Improved GrafPop for ALFA genetic ancestry inference