Infinium® GSAv2 Clinical Research Coverage Summary
(this content will be on AoU array also)

ClinVar Known and/or Actionable   55,385

- > 13,000 markers in ACMG-59 genes
- > 40,000 clinically interesting variants from ClinVar for optimal RoR performance
- > 25,000 markers in TruSight Inherited Disease Panel genes
- Updated as of April 2018

Pharmacogenomics   17,220

- All annotated variants in PharmGKB as April 2018
- Optimized ADME and CPIC**-17 coverage
- 145 drug associations
- Unpublished content provided by PGx-KOLs

Common Disease Variants from GWAS   10,574

- Common Disease Common Variant Associations
- Non-Clinical Associations for Lifestyle and Wellness
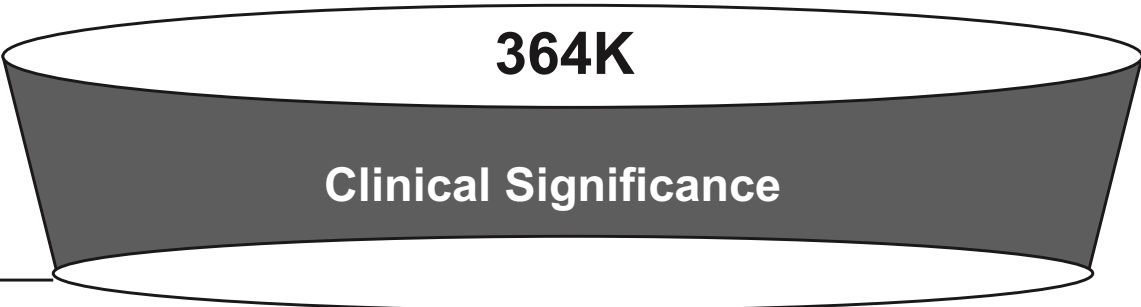
HLA and MHC   8,577

- Expert-selected HLA SNPs to enable low resolution HLA calling by imputation across diverse population ethnicities
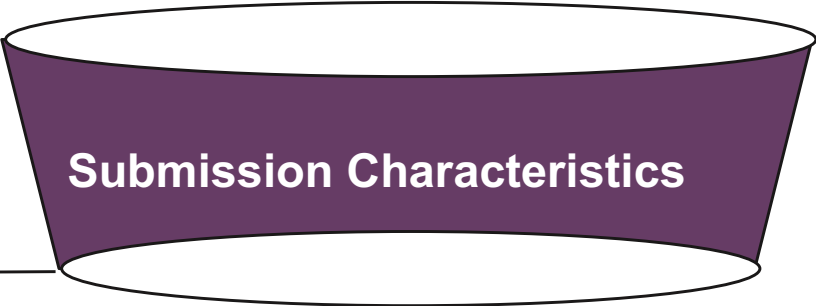
gnomAD Exome   67,371

- Comprehensive exome coverage from a diverse panel of > 123,136 exomes
- gnomAD version 2.0 updated October 2017

**CPIC – Clinical Pharmacogenetics Implementation Consortium
*counts represent unique variants in each category however the sum in each category does not represent the number of unique variants, as variants are likely annotated in multiple sources.
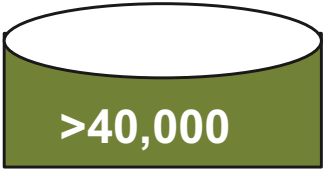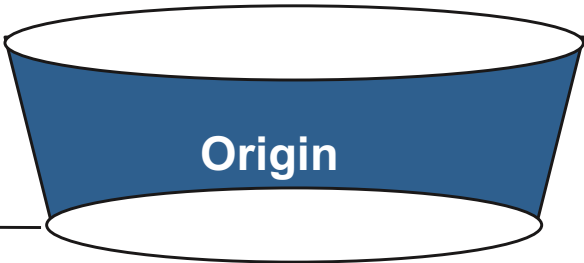
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ClinVar Selection Strategy

- **Pathogenic and Likely Pathogenic**
- **ACMG variants were included regardless of annotation**
- **Removed variants missing clinical annotations**

364K

Clinical Significance

- **Exclude missing review status**
- **Must have multiple submitters**
- **Medical Genetics Review score of 4 or 5 stars**

Submission Characteristics

- **Exclude somatic mutations**

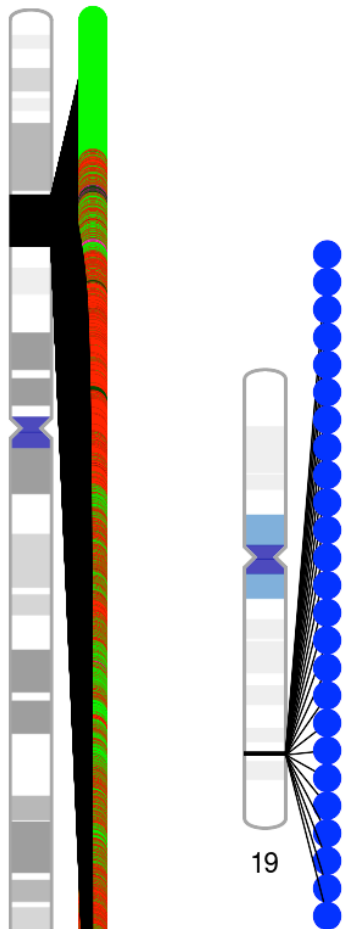**Origin**

>40,000

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*counts represent unique variants in each category however the sum in each category does not represent the number of unique variants, as variants are likely annotated in multiple sources.

Infinium® GSAv2 :Human Leukocyte Antigen (HLA) Complex

HLA content optimized for imputation and association discovery

- **MHC region occurs on Chr6 from 6p22.1 to 6p21.3 and contains 224 genes spanning 3.6 megabase pairs**

- **8,577 markers for imputation for discovery**

  - HLA collaborators report > 0.80 imputation accuracy for > 96% variants in Europeans

- **26 markers in KIR**

HLA Imputation Accuracy		
	Europeans	Asians
HLA-A	99.1%	98.1%
HLA-B	96.8%	65.6%
HLA-C	99.1%	68.8%
HLA-DQA1	98.5%	96.3%
HLA-DQB1	99.1%	96.5%
HLA-DRB1	96.9%	92.3%
All loci	98.3%	86.4%

19

6

- KIR-Gene
- MHC extended coverage
- HLA-Imputation/MHC extended coverage

- HLA-Imputation
- HLA-Gene/MHC extended coverage
- HLA-Imputation/HLA-Gene/MHC extended coverage

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