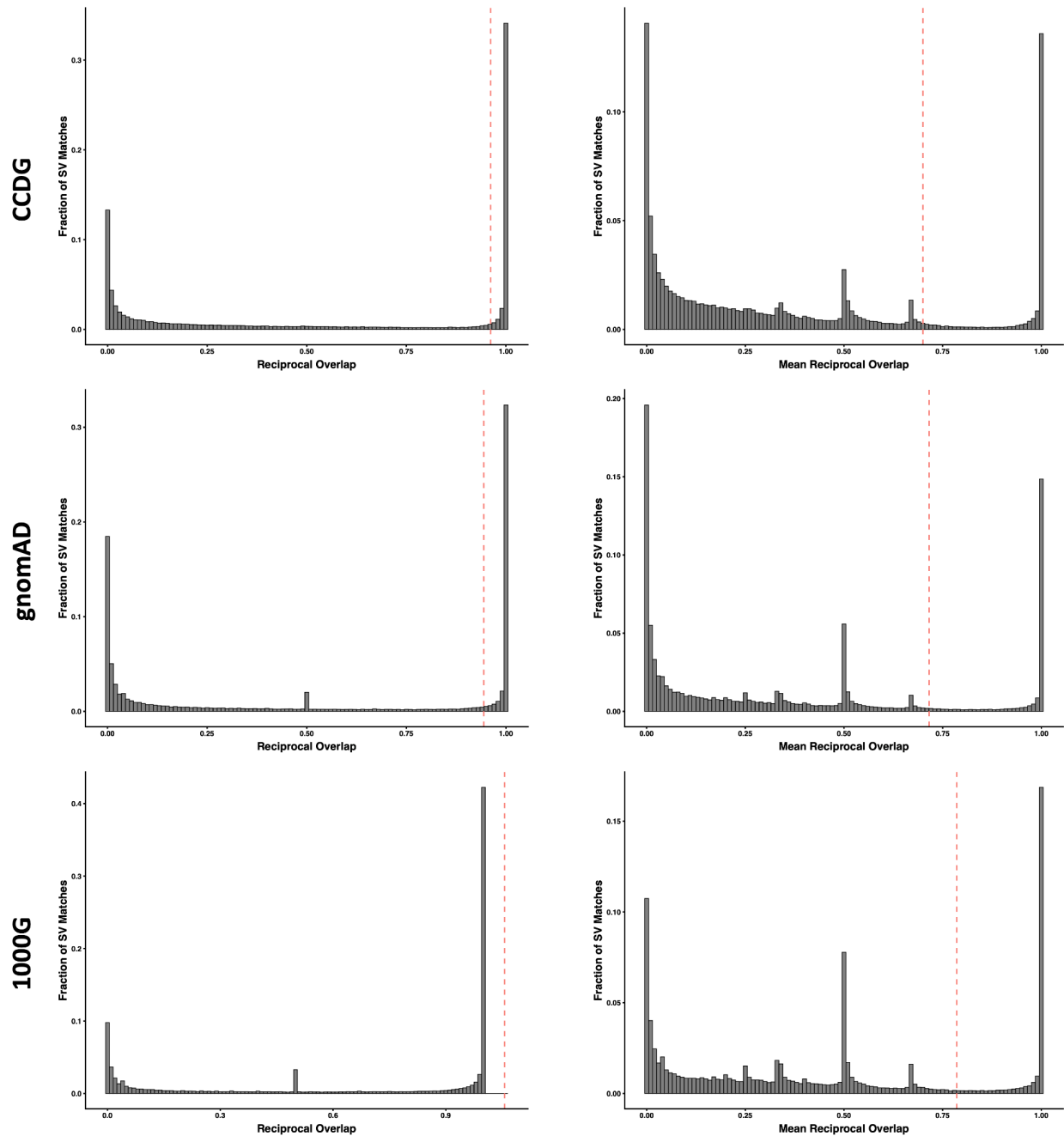


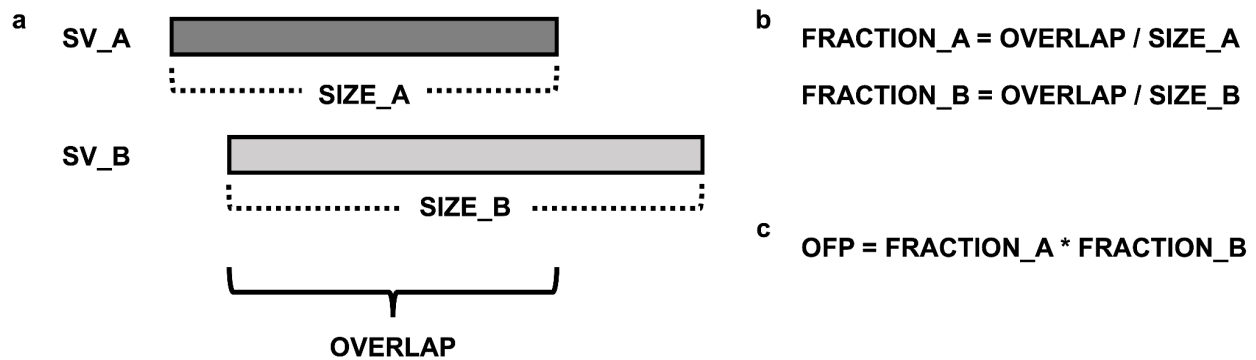
Supplementary Figure 1. Reported SV Rarities by CCDG, gnomAD, and 1000G

The fraction of SVs reported by CCDG, gnomAD, and 1000G found to be “Common” ($AF \geq 0.05$), “LowFreq” ($0.05 > AF \geq 0.01$), and “Rare” ($AF < 0.01$). Additionally, “Singletons” where allele count in genotypes is 1 ($AC = 1$), which are inherently a subset of the “Rare” SVs, are also plotted. **a.** Features all SVs reported by each dataset where on average, approximately 8% are “Common”, 6% are “LowFreq”, and 85% are “Rare” with 36% of all SVs being “Singletons”. **b.** Limits the SVs to SVTYPES that the datasets have in common (deletions, duplications, and inversions) where on average approximately 6% are “Common”, 4% are “LowFreq”, and 90% are “Rare” with 47% of all SVs being “Singletons”.



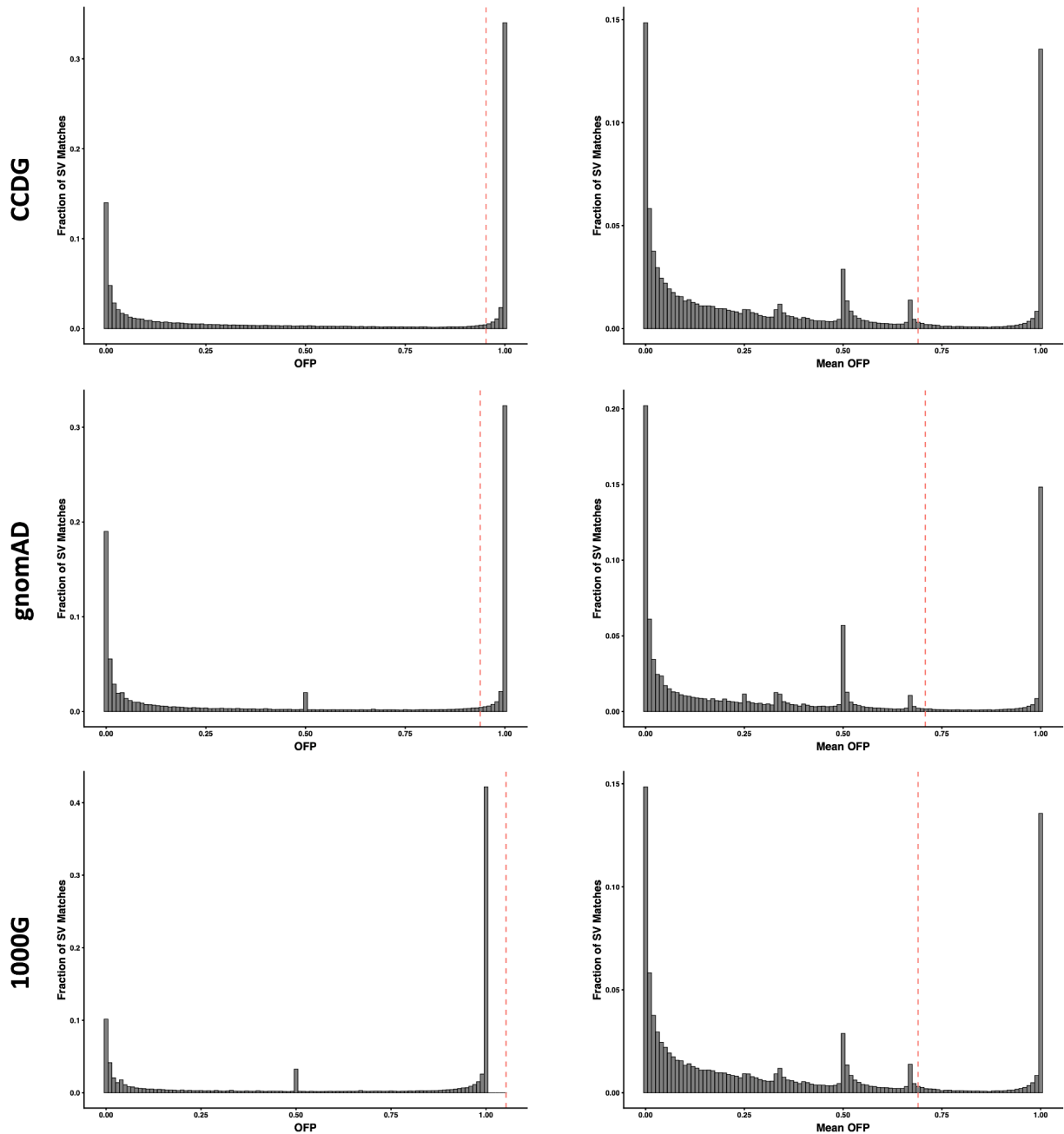
Supplementary Figure 2. Distributions of the Reciprocal Overlap Identified Across Matches from Dataset Comparisons

For the CCDG, gnomAD, and 1000G datasets, distributions of reciprocal overlaps observed between all possible matches between the datasets is plotted for each dataset with one standard deviation beyond the mean indicated as a red dashed line. On the left, distributions reflecting only the highest observed reciprocal overlap for each given SV in a dataset is plotted while on the right, distributions representing the mean observed reciprocal overlap for all matches to a given SV in a dataset is shown.



Supplementary Figure 3. Calculating the Overlap Fraction Product (OFP)

a. For each overlapping pair of SVs, the amount of overlap is determined. b. Using the overlap, the fraction of each SV that is found to overlap with the other is calculated. c. These fractions are then multiplied to create the Overlap Fraction Product (OFP).



Supplementary Figure 4. Distributions of OFPs Across Dataset Comparisons

For the CCDG, gnomAD, and 1000G datasets, distributions of OFPs observed between all possible matches between the datasets is plotted for each dataset with one standard deviation beyond the mean indicated as a red dashed line. On the left, distributions reflecting only the highest or best observed OFP for each given SV in a dataset is plotted while on the right distributions representing the mean OFP for all matches to a given SV in a dataset is shown.

Supplementary Table 1. Expected Columns for Input BED File

Expected Column	Description
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CHROM	Chromosome
START	Start position of SV
END	End position of SV
SVLEN	Length of SV
SVTYPE	Type of SV
SOURCE	Data source that reports the SV
SV_ID	Unique identifier for the SV
AF	Allele frequency
HomRef	Count of individuals with homozygous for the reference allele genotype
Het	Count of individuals with heterozygous genotype
HomAlt	Count of individuals with homozygous for the alternate allele genotype
Male_AF	Allele frequency of male samples
Male_HomRef	Count of male individuals with homozygous for the reference allele genotype
Male_Het	Count of male individuals with heterozygous genotype
Male_HomAlt	Count of male individuals with homozygous for the alternate allele genotype
Male_HemiAlt	Count of male individuals with hemizygous genotype
Male_HemiAF	Hemizygous allele frequency
Female_AF	Allele frequency of female samples
Female_HomRef	Count of female individuals with homozygous for the reference allele genotype
Female_Het	Count of female individuals with heterozygous genotype
Female_HomAlt	Count of female individuals with homozygous for the alternate allele genotype
AFR_AF	Allele frequency of AFR samples
AFR_HomRef	Count of AFR individuals with homozygous for the reference allele genotype
AFR_Het	Count of AFR individuals with heterozygous genotype
AFR_HomAlt	Count of AFR individuals with homozygous for the alternate allele genotype
AFR_Male_AF	Allele frequency of AFR male samples
AFR_Male_HomRef	Count of AFR male individuals with homozygous for the reference allele genotype
AFR_Male_Het	Count of AFR male individuals with heterozygous genotype
AFR_Male_HomAlt	Count of AFR male individuals with homozygous for the alternate allele genotype
AFR_Male_HemiAlt	Count of AFR male individuals with hemizygous genotype
AFR_Male_HemiAF	Hemizygous allele frequency of AFR samples
AFR_Female_AF	Allele frequency of AFR female samples
AFR_Female_HomRef	Count of AFR female individuals with homozygous for the reference allele genotype

AFR_Female_Het	Count of AFR female individuals with heterozygous genotype
AFR_Female_HomAlt	Count of AFR female individuals with homozygous for the alternate allele genotype
AMR_AF	Allele frequency of AMR samples
AMR_HomRef	Count of AMR individuals with homozygous for the reference allele genotype
AMR_Het	Count of AMR individuals with heterozygous genotype
AMR_HomAlt	Count of AMR individuals with homozygous for the alternate allele genotype
AMR_Male_AF	Allele frequency of AMR male samples
AMR_Male_HomRef	Count of AMR male individuals with homozygous for the reference allele genotype
AMR_Male_Het	Count of AMR male individuals with heterozygous genotype
AMR_Male_HomAlt	Count of AMR male individuals with homozygous for the alternate allele genotype
AMR_Male_HemiAlt	Count of AMR male individuals with hemizygous genotype
AMR_Male_HemiAF	Hemizgous allele frequency of AMR samples
AMR_Female_AF	Allele frequency of AMR female samples
AMR_Female_HomRef	Count of AMR female individuals with homozygous for the reference allele genotype
AMR_Female_Het	Count of AMR female individuals with heterozygous genotype
AMR_Female_HomAlt	Count of AMR female individuals with homozygous for the alternate allele genotype
EAS_AF	Allele frequency of EAS samples
EAS_HomRef	Count of EAS individuals with homozygous for the reference allele genotype
EAS_Het	Count of EAS individuals with heterozygous genotype
EAS_HomAlt	Count of EAS individuals with homozygous for the alternate allele genotype
EAS_Male_AF	Allele frequency of EAS male samples
EAS_Male_HomRef	Count of EAS male individuals with homozygous for the reference allele genotype
EAS_Male_Het	Count of EAS male individuals with heterozygous genotype
EAS_Male_HomAlt	Count of EAS male individuals with homozygous for the alternate allele genotype
EAS_Male_HemiAlt	Count of EAS male individuals with hemizygous genotype
EAS_Male_HemiAF	Hemizgous allele frequency of EAS samples
EAS_Female_AF	Allele frequency of EAS female samples
EAS_Female_HomRef	Count of EAS female individuals with homozygous for the reference allele genotype
EAS_Female_Het	Count of EAS female individuals with heterozygous genotype
EAS_Female_HomAlt	Count of EAS female individuals with homozygous for the alternate allele

	genotype
EUR_AF	Allele frequency of EUR samples
EUR_HomRef	Count of EUR individuals with homozygous for the reference allele genotype
EUR_Het	Count of EUR individuals with heterozygous genotype
EUR_HomAlt	Count of EUR individuals with homozygous for the alternate allele genotype
EUR_Male_AF	Allele frequency of EUR male samples
EUR_Male_HomRef	Count of EUR male individuals with homozygous for the reference allele genotype
EUR_Male_Het	Count of EUR male individuals with heterozygous genotype
EUR_Male_HomAlt	Count of EUR male individuals with homozygous for the alternate allele genotype
EUR_Male_HemiAlt	Count of EUR male individuals with hemizygous genotype
EUR_Male_HemiAF	Hemizgous allele frequency of EUR samples
EUR_Female_AF	Allele frequency of EUR female samples
EUR_Female_HomRef	Count of EUR female individuals with homozygous for the reference allele genotype
EUR_Female_Het	Count of EUR female individuals with heterozygous genotype
EUR_Female_HomAlt	Count of EUR female individuals with homozygous for the alternate allele genotype
OTH_AF	Allele frequency of OTH samples
OTH_HomRef	Count of OTH individuals with homozygous for the reference allele genotype
OTH_Het	Count of OTH individuals with heterozygous genotype
OTH_HomAlt	Count of OTH individuals with homozygous for the alternate allele genotype
OTH_Male_AF	Allele frequency of OTH male samples
OTH_Male_HomRef	Count of OTH male individuals with homozygous for the reference allele genotype
OTH_Male_Het	Count of OTH male individuals with heterozygous genotype
OTH_Male_HomAlt	Count of OTH male individuals with homozygous for the alternate allele genotype
OTH_Male_HemiAlt	Count of OTH male individuals with hemizygous genotype
OTH_Male_HemiAF	Hemizgous allele frequency of OTH samples
OTH_Female_AF	Allele frequency of OTH female samples
OTH_Female_HomRef	Count of OTH female individuals with homozygous for the reference allele genotype
OTH_Female_Het	Count of OTH female individuals with heterozygous genotype
OTH_Female_HomAlt	Count of OTH female individuals with homozygous for the alternate allele genotype
SAS_AF	Allele frequency of SAS samples

SAS_HomRef	Count of SAS individuals with homozygous for the reference allele genotype
SAS_Het	Count of SAS individuals with heterozygous genotype
SAS_HomAlt	Count of SAS individuals with homozygous for the alternate allele genotype
SAS_Male_AF	Allele frequency of SAS male samples
SAS_Male_HomRef	Count of SAS male individuals with homozygous for the reference allele genotype
SAS_Male_Het	Count of SAS male individuals with heterozygous genotype
SAS_Male_HomAlt	Count of SAS male individuals with homozygous for the alternate allele genotype
SAS_Male_HemiAlt	Count of SAS male individuals with hemizygous genotype
SAS_Male_HemiAF	Hemizygous allele frequency of SAS samples
SAS_Female_AF	Allele frequency of SAS female samples
SAS_Female_HomRef	Count of SAS female individuals with homozygous for the reference allele genotype
SAS_Female_Het	Count of SAS female individuals with heterozygous genotype
SAS_Female_HomAlt	Count of SAS female individuals with homozygous for the alternate allele genotype
PopMax_AF	The maximum AF across all populations
InPop	The number of populations that report the SV

Supplementary Table 2. SVAfotate Annotations

SVAFotate Parameter	Added Annotation	Description
default	Max_AF Max_Het Max_HomAlt Max_PopMax_AF	The maximum AF from all matching SVs The maximum count of heterozygote genotypes from all matching SVs The maximum count of homozygote alternate genotypes from all matching SVs The maximum PopMax_AF from all matching SVs
-a best	Best_[data_source]_ID Best_[data_source]_OFP Best_[data_source]_AF Best_[data_source]_Het Best_[data_source]_HomAlt Best_[data_source]_PopMax_AF	The SV ID of the best matching SV for that data source The OFP of the best matching SV for that data source The AF of the best matching SV for that data source The count of heterozygous genotypes for best matching SV for that data source The count of homozygous alternate genotypes for best matching SV for that data source The PopMax_AF of the best matching SV for that data source
-a mf	Max_Male_AF Max_Male_Het Max_Male_HomAlt Max_Female_AF Max_Female_Het Max_Female_HomAlt	The maximum male AF from all matching SVs The maximum male count of heterozygous genotypes from all matching SVs The maximum male count of homozygous alternate genotypes from all matching SVs The maximum female AF from all matching SVs The maximum female count of heterozygous genotypes from all matching SVs The maximum female count of homozygous alternate genotypes from all matching SVs
-a mis	[data_source]_Mismatches [data_source]_Mismatches_Count [data_source]_Mismatches_SVTYPEs Best_[data_source]_Mismatch_ID Best_[data_source]_Mismatch_OFP Best_[data_source]_Mismatch_SVTYPE Best_[data_source]_Mismatch_AF Best_[data_source]_Mismatch_Het Best_[data_source]_Mismatch_HomAlt	List of SV IDs from all overlapping SVs with a different SVTYPE for that data source The number of mismatches identified for that data source The other SVTYPEs identified from overlapping SVs with a different SVTYPE for that data source The SV ID of the best mismatching SV for that data source The OFP of the best mismatching SV for that data source The SVTYPE of the best mismatching SV for that data source The AF of the best mismatching SV for that data source The count of heterozygous genotypes for best mismatching SV for that data source The count of homozygous alternate genotypes for best mismatching SV for that data source
-a pops	Max_[population]_AF Max_[population]_Het Max_[population]_HomAlt	The maximum population specific AF from all matching SVs The maximum population specific count of heterozygote genotypes from all matching SVs The maximum population specific count of homozygote alternate genotypes from all matching SVs
-c	SV_Cov [data_source]_SV_Cov	The fraction of the SV that has been observed with the same SVTYPE across all data sources The fraction of the SV that has been observed with the same SVTYPE for that data source
-u	SV_Uniq	The number of unique regions found within the SV
-t	Target_Overlaps	The region identifier(s) for all overlaps between the SV and the regions of interest in the targets BED file