

PrivaSeq2 Figures

--Attacker Strikes Back--

Fig 1a

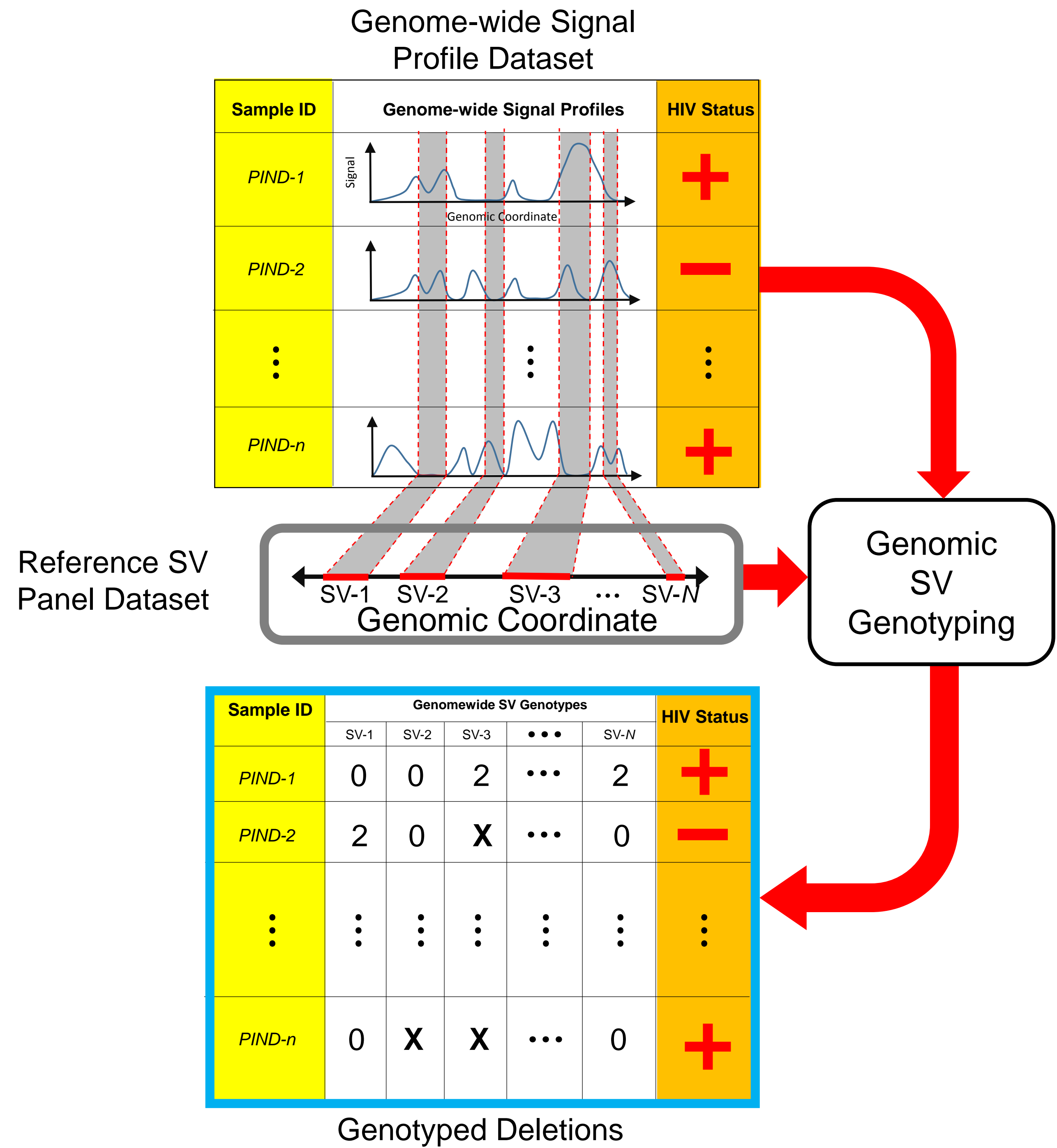


Fig 1b

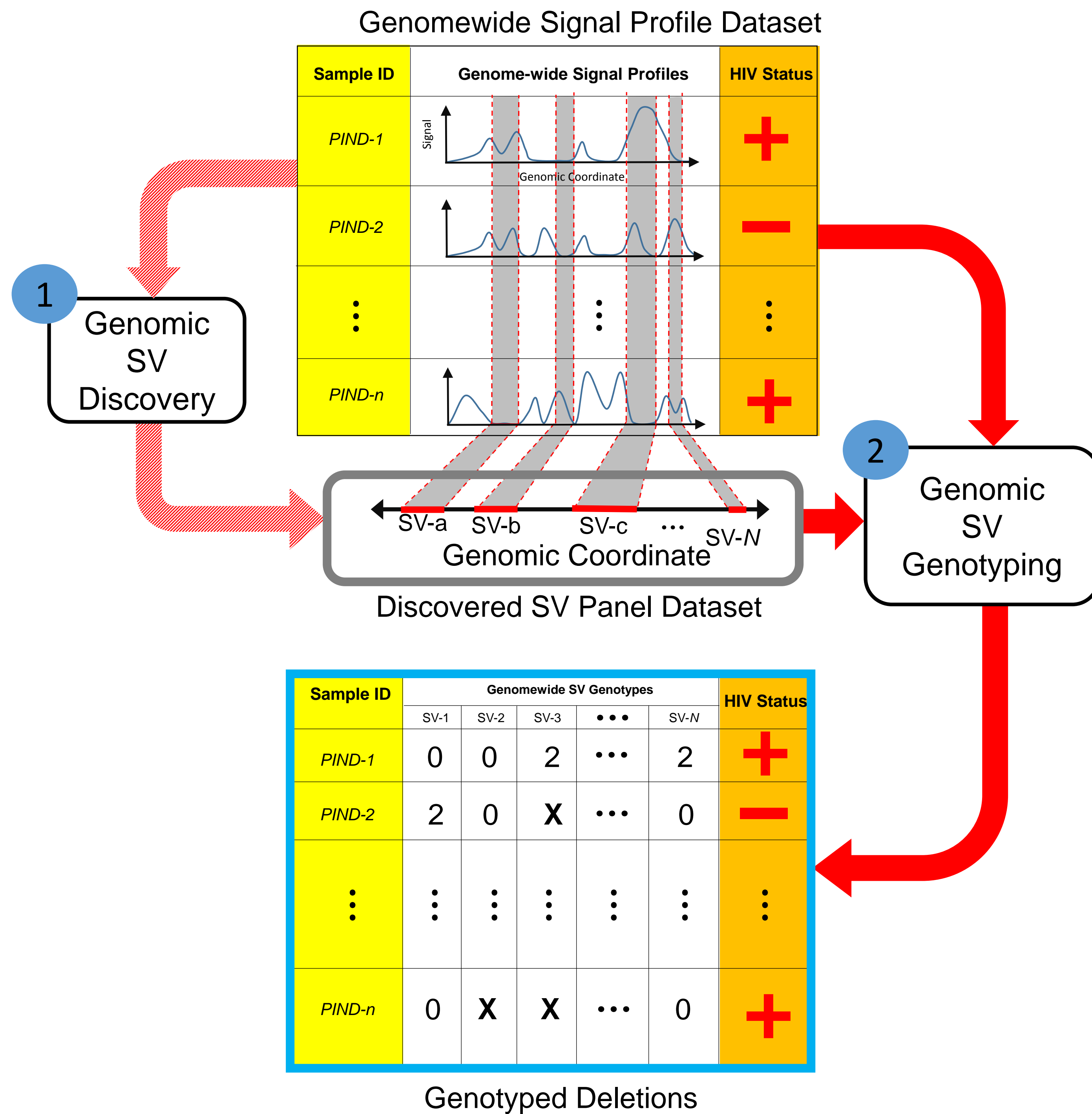


Fig 1c

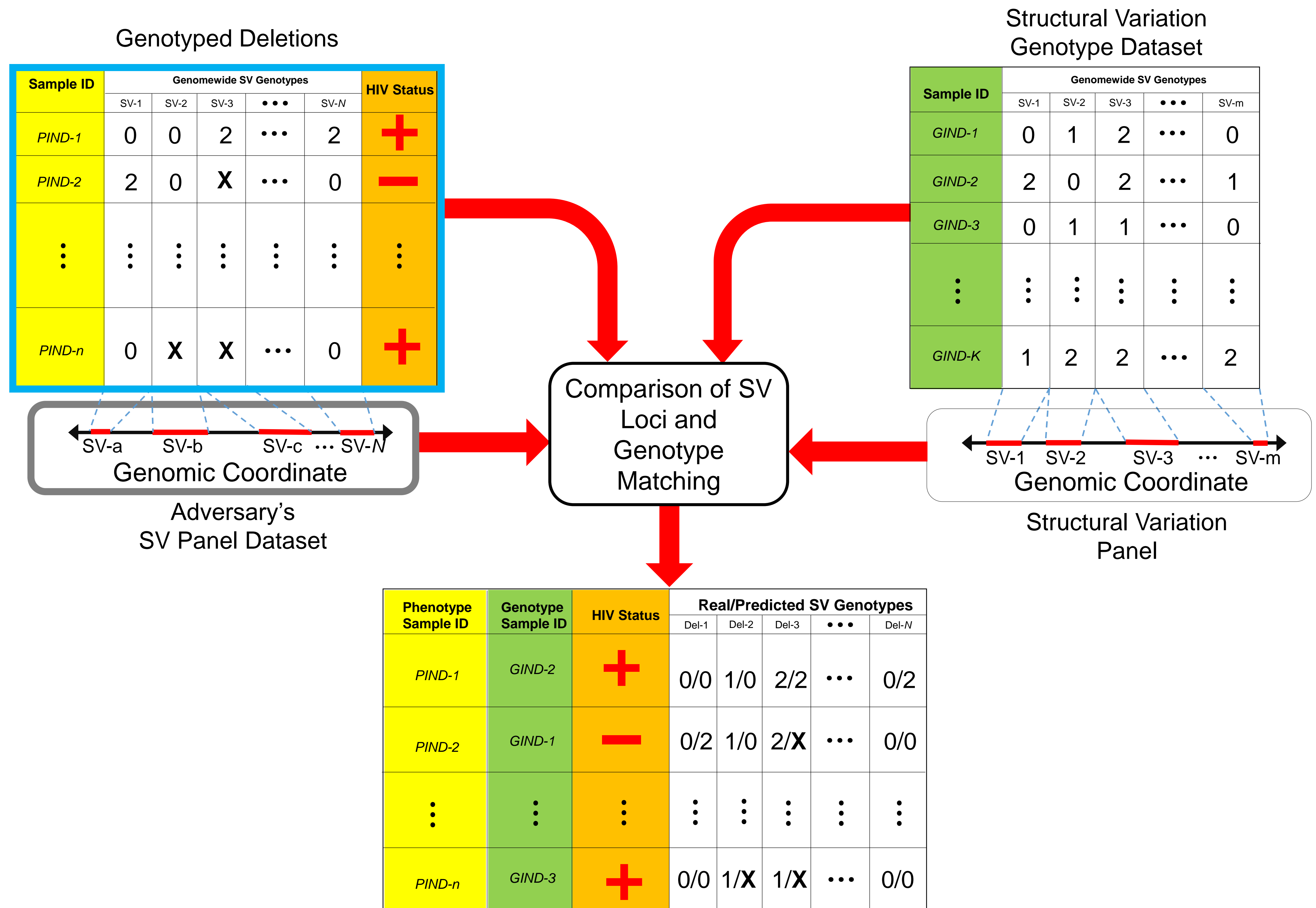


Fig 1 *[[ADD RNA-Seq TRACK]]*

c) NA12878 large deletion

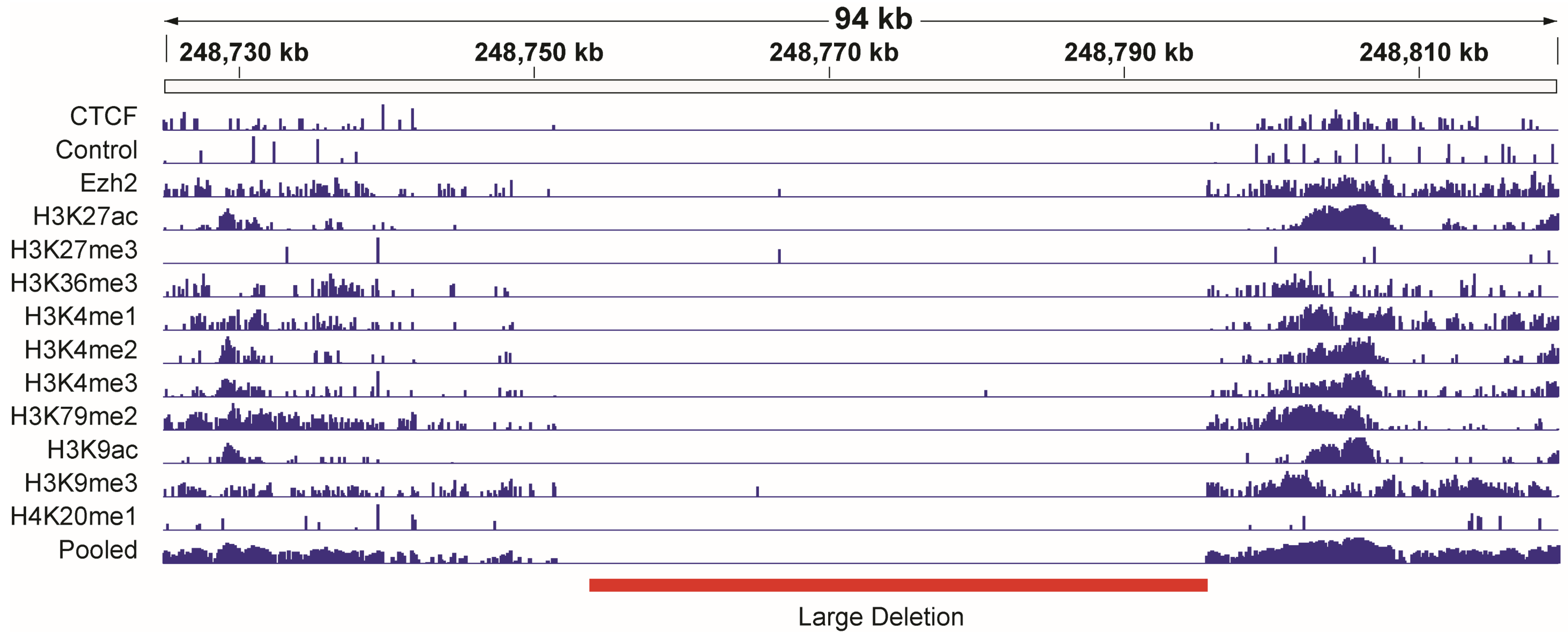


Fig 1

b) Detection and Genotyping of small and large deletions from signal profiles

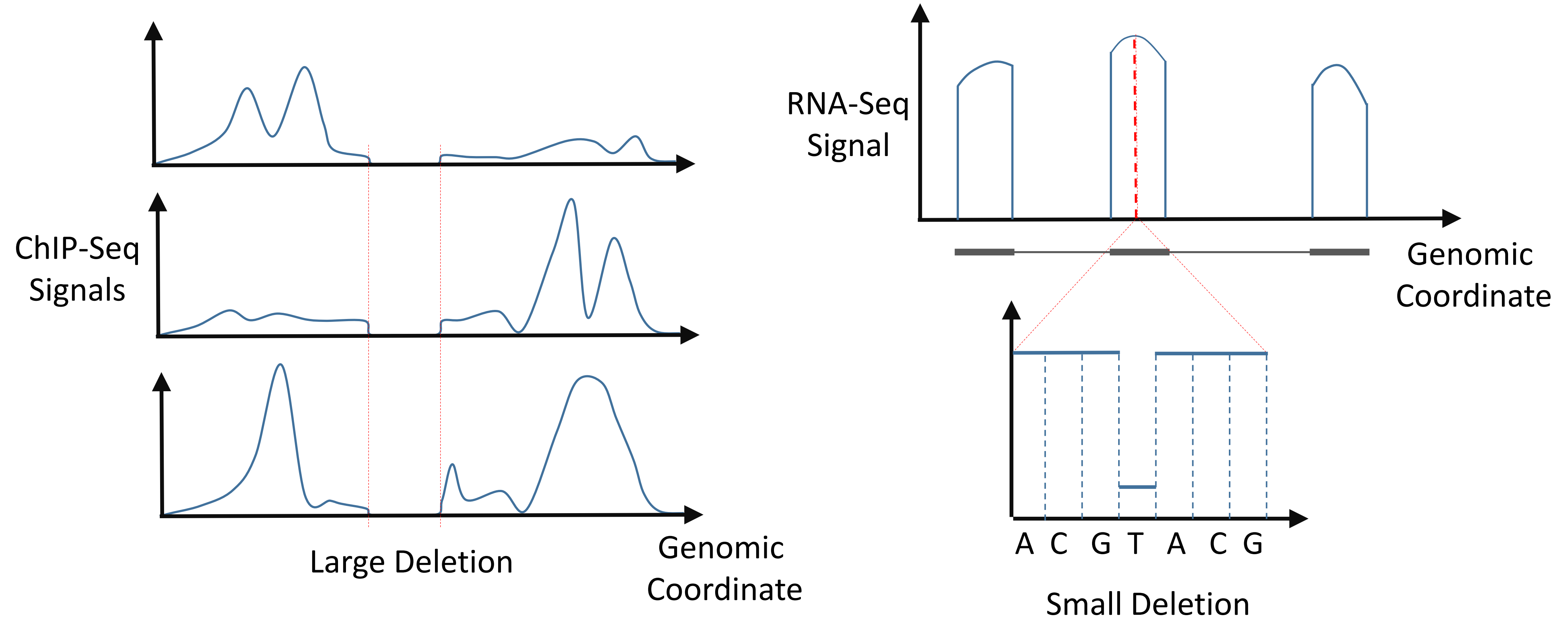
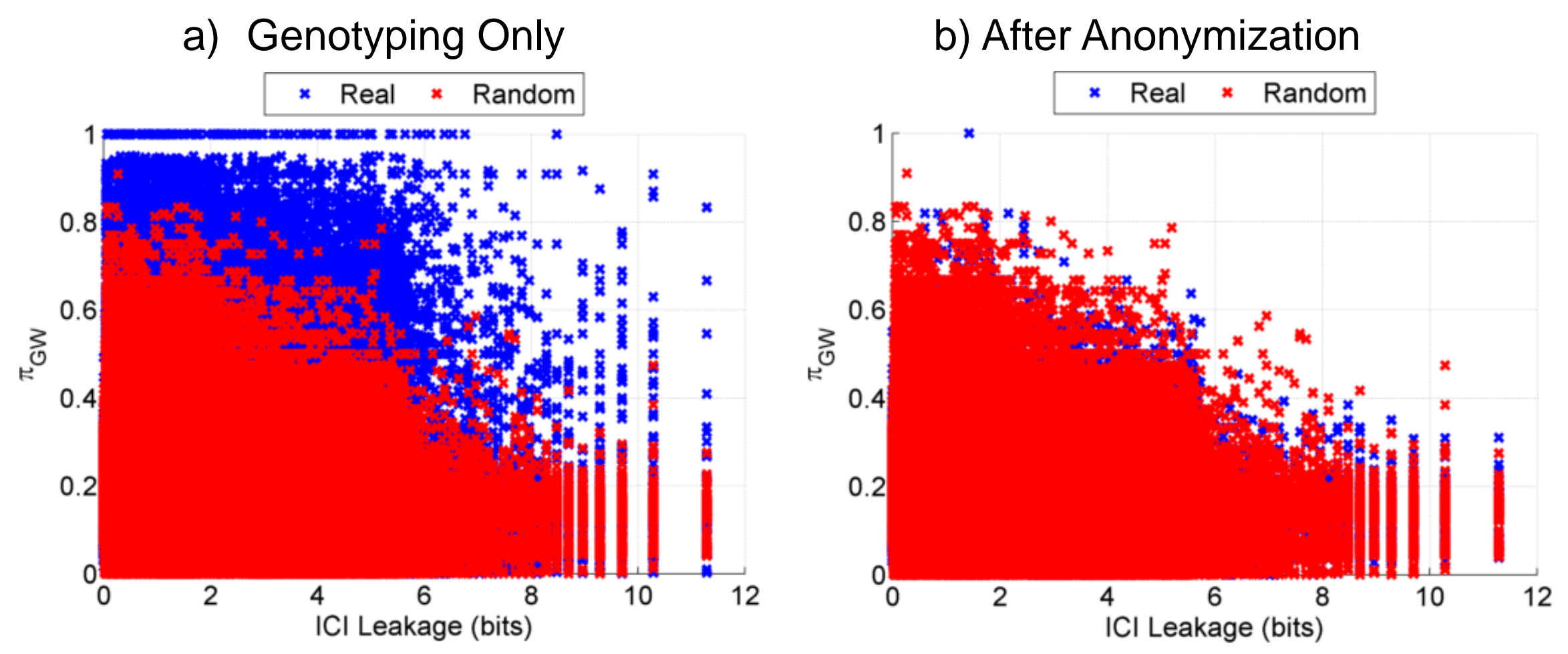
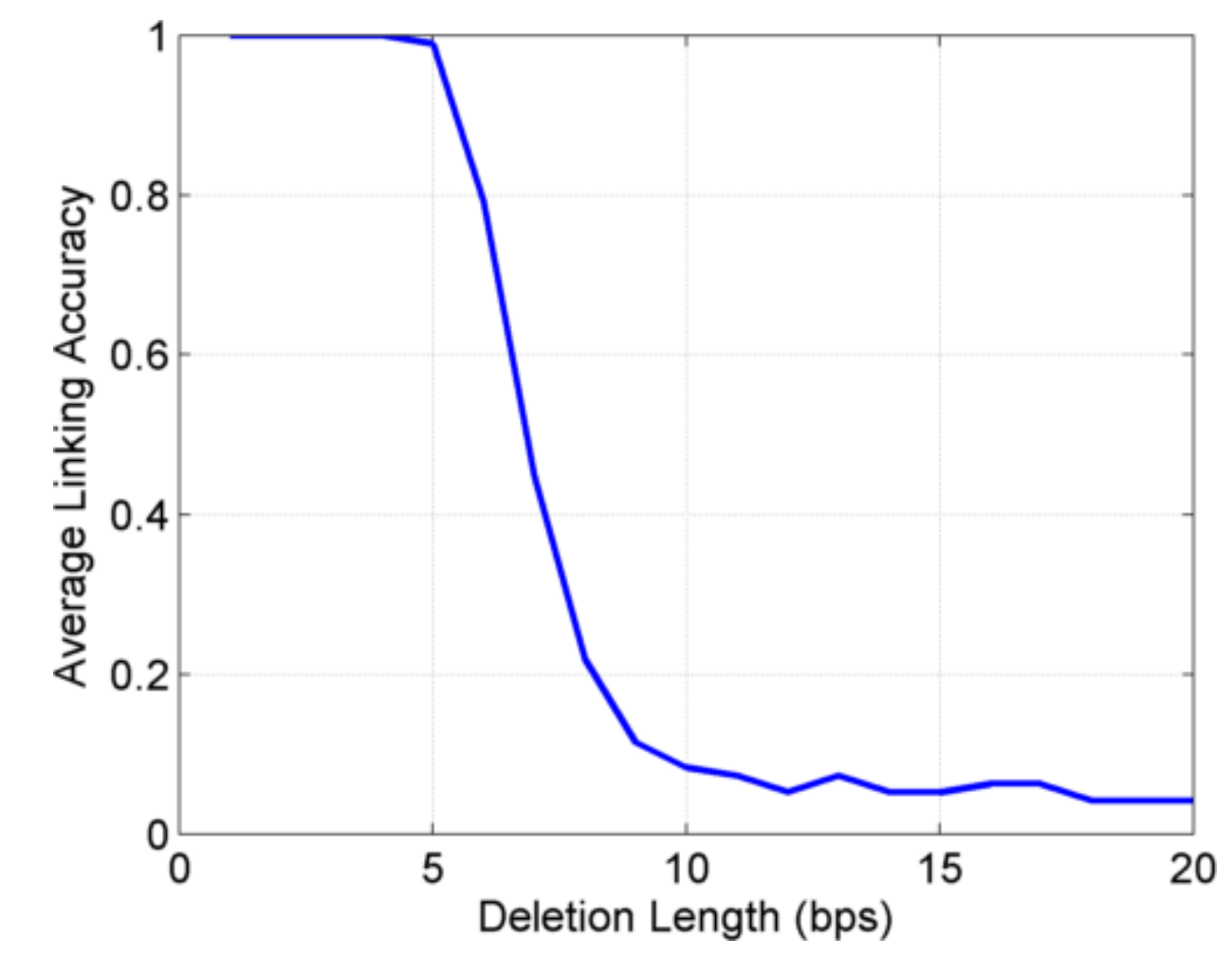


Fig 2

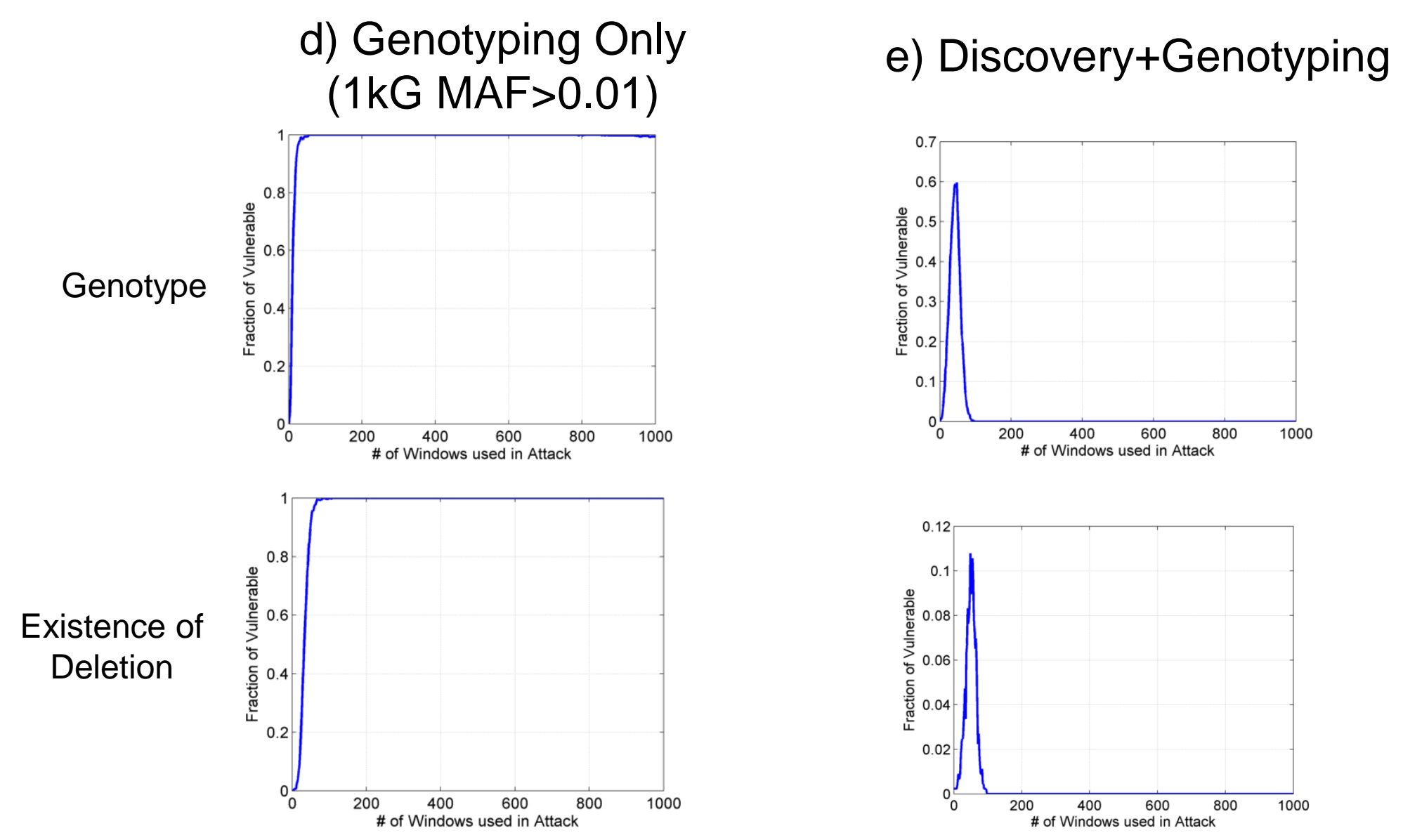
a) GEUVADIS ICI-\Pi_GW estimates:



e) Indel length vs vulnerability statistics



c, d) GEUVADIS extremity attack: c) Genotyping only
d) Joint Discovery + Genotyping



f,g) Min # of identifying variants

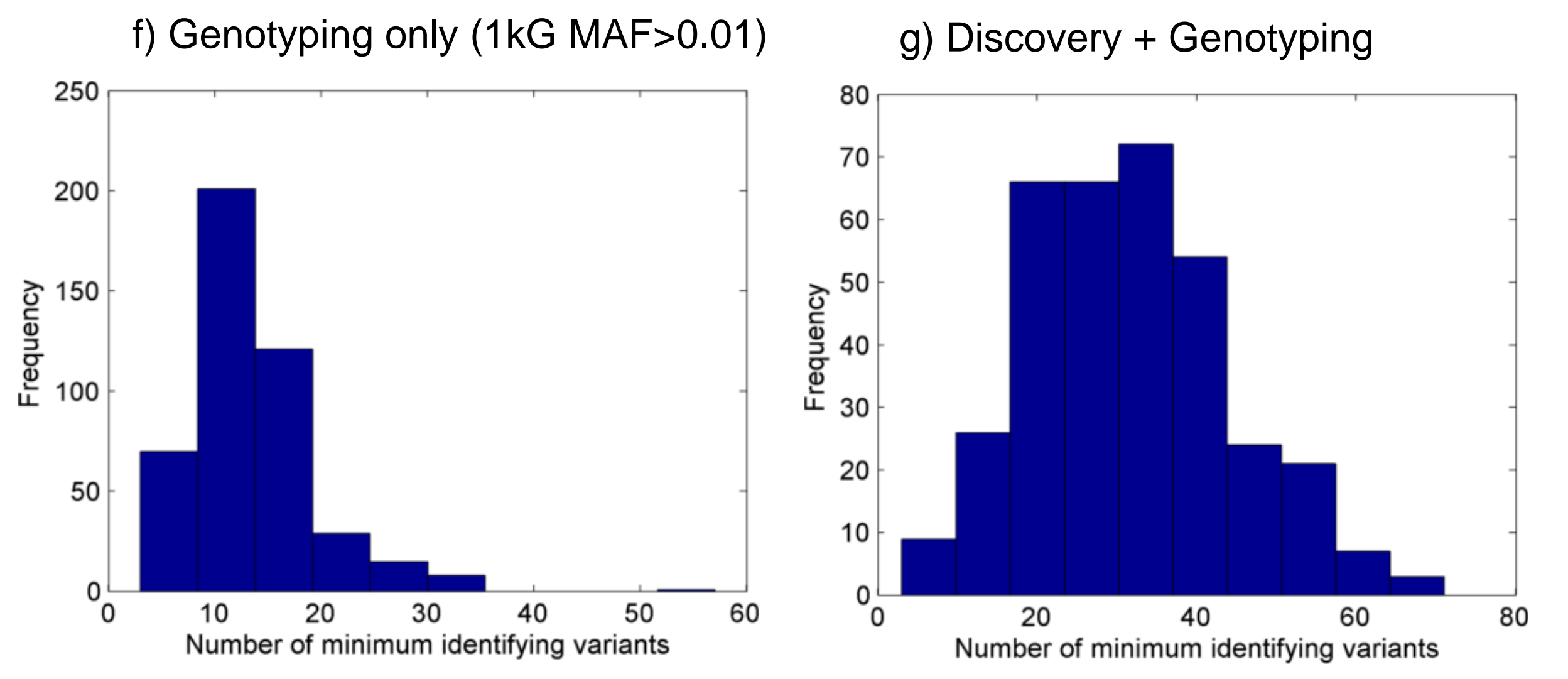
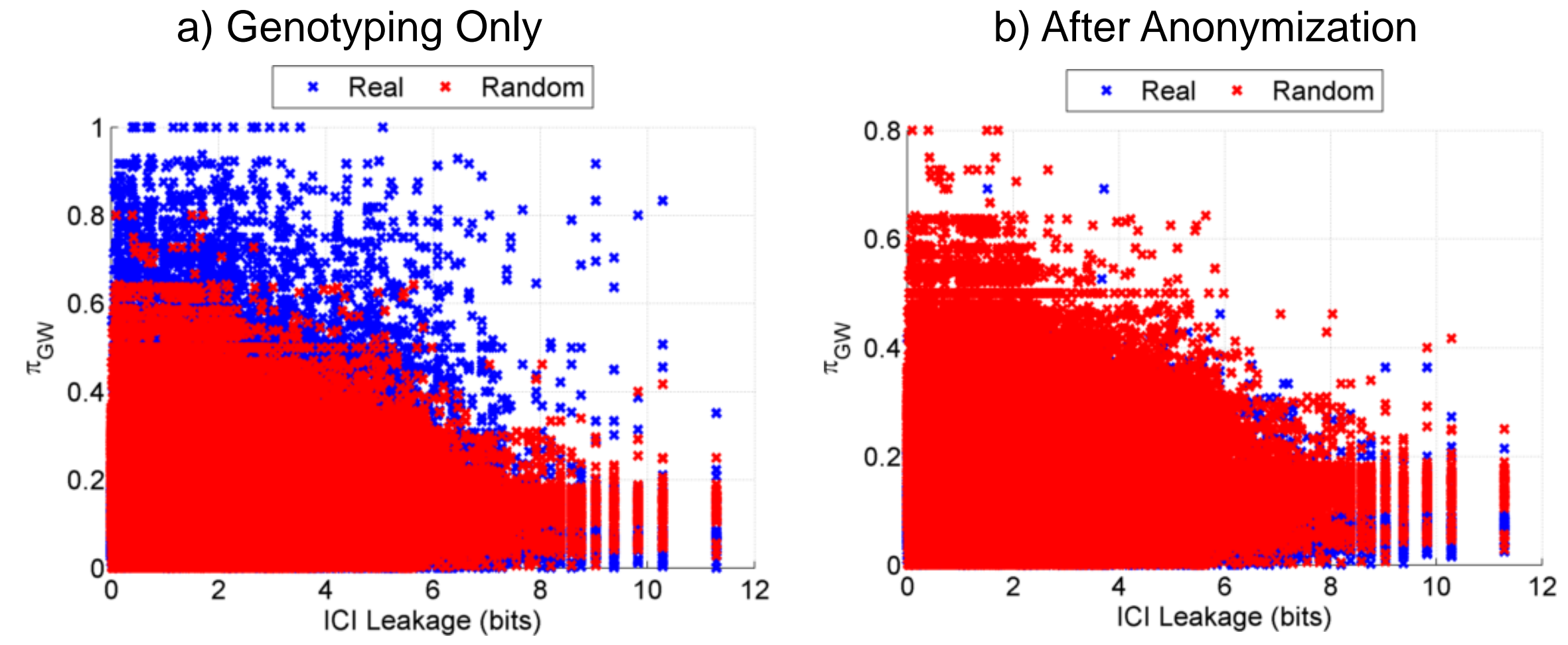


Fig 3

a, b) Gtex ICI-\Pi_GW estimates: a) known indels, b) De-novo indels (1kG indels, GTeX mapped reads)



Gtex extremity attack: known indels (1kG MAF>0.01)
c) Genotyping d) d_1,2 vs Vulnerability

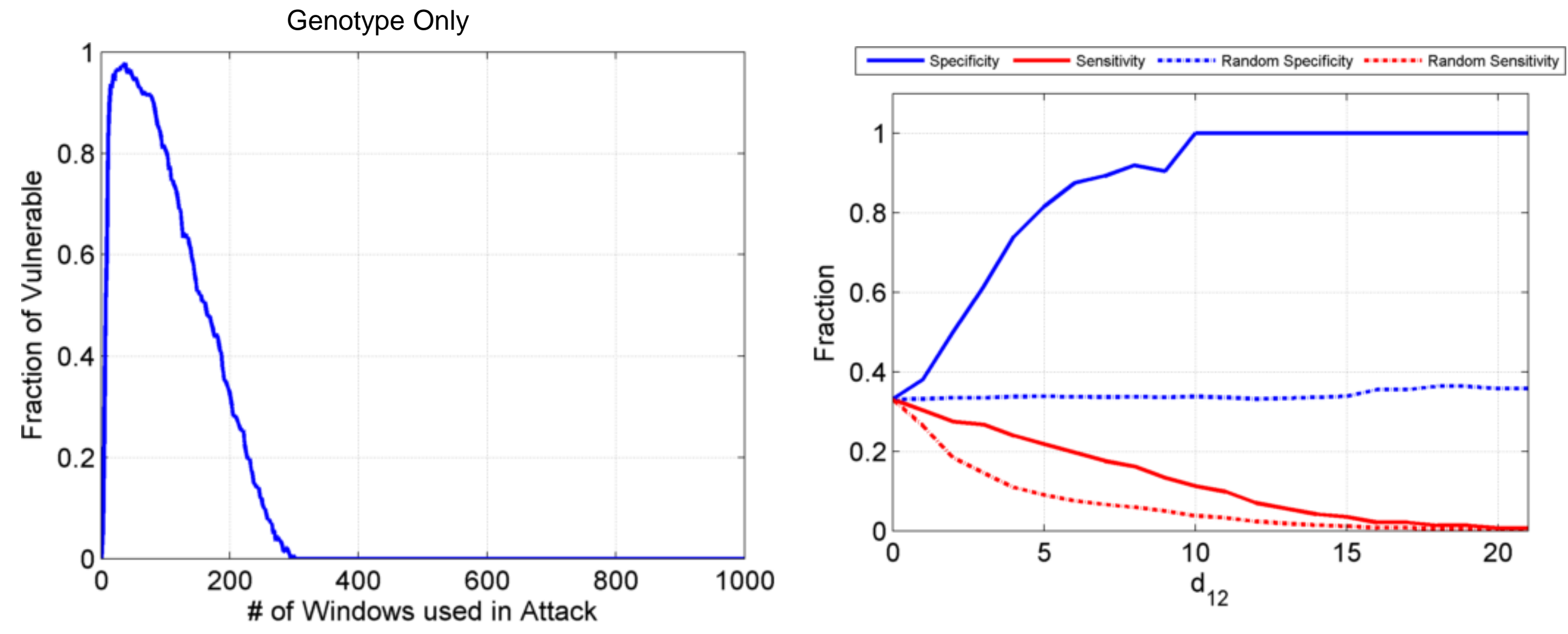
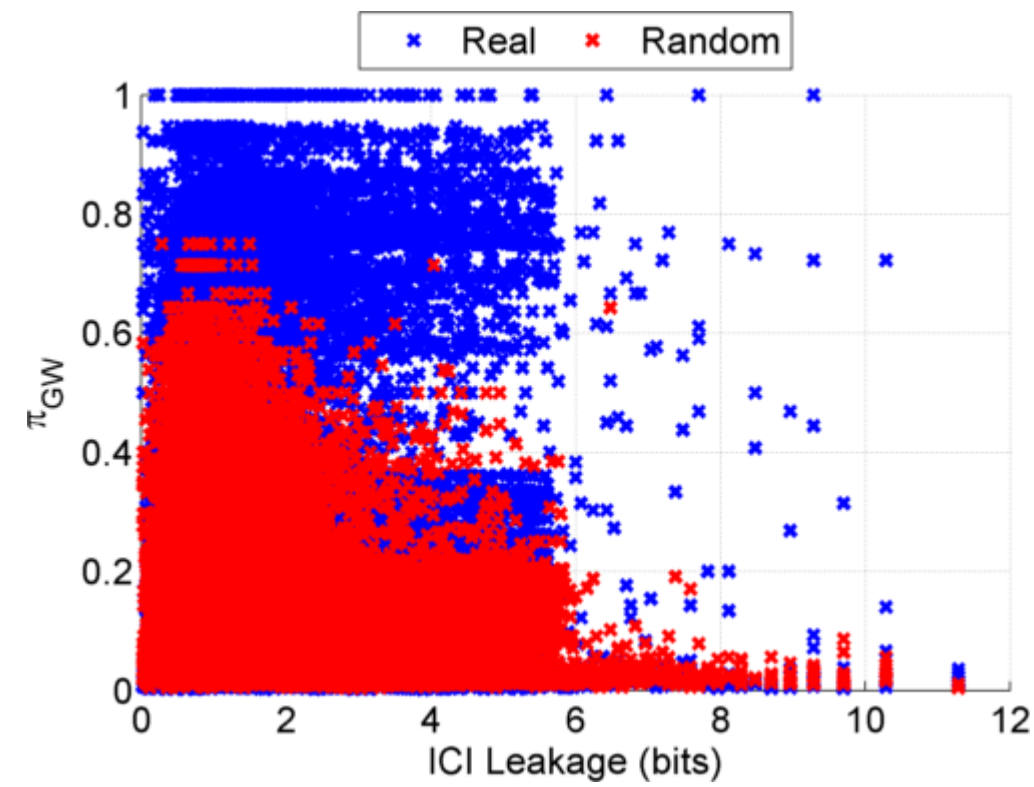


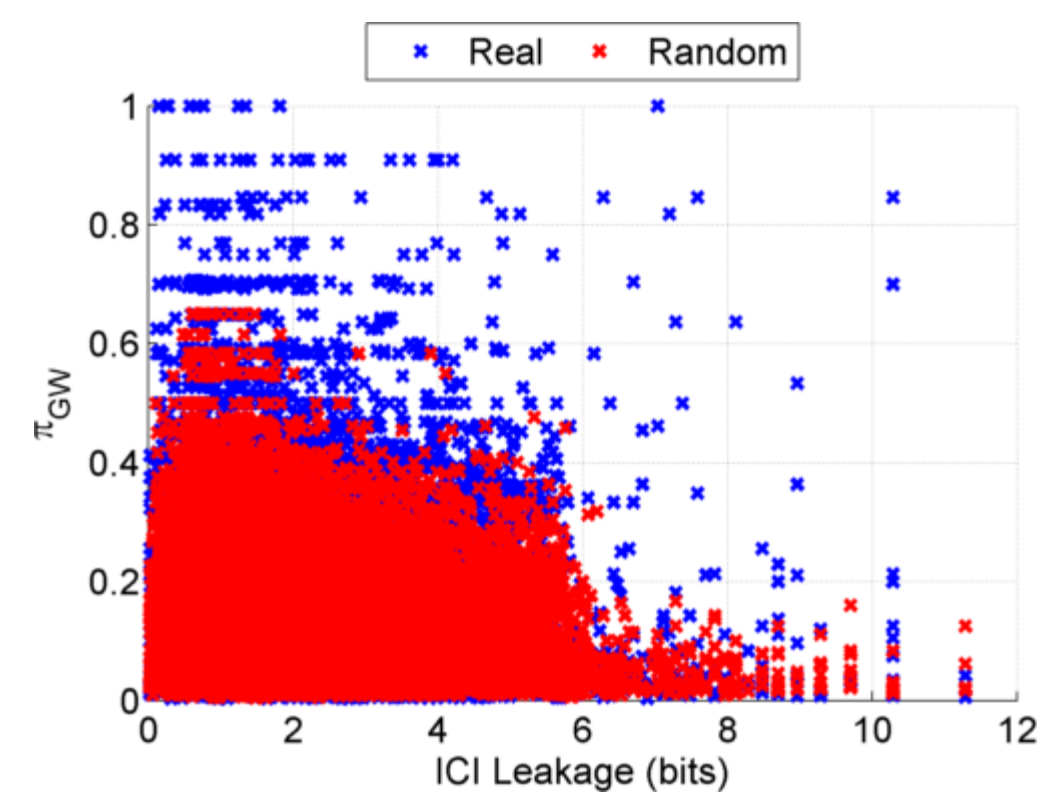
Fig 4

a) Genotyping large deletions

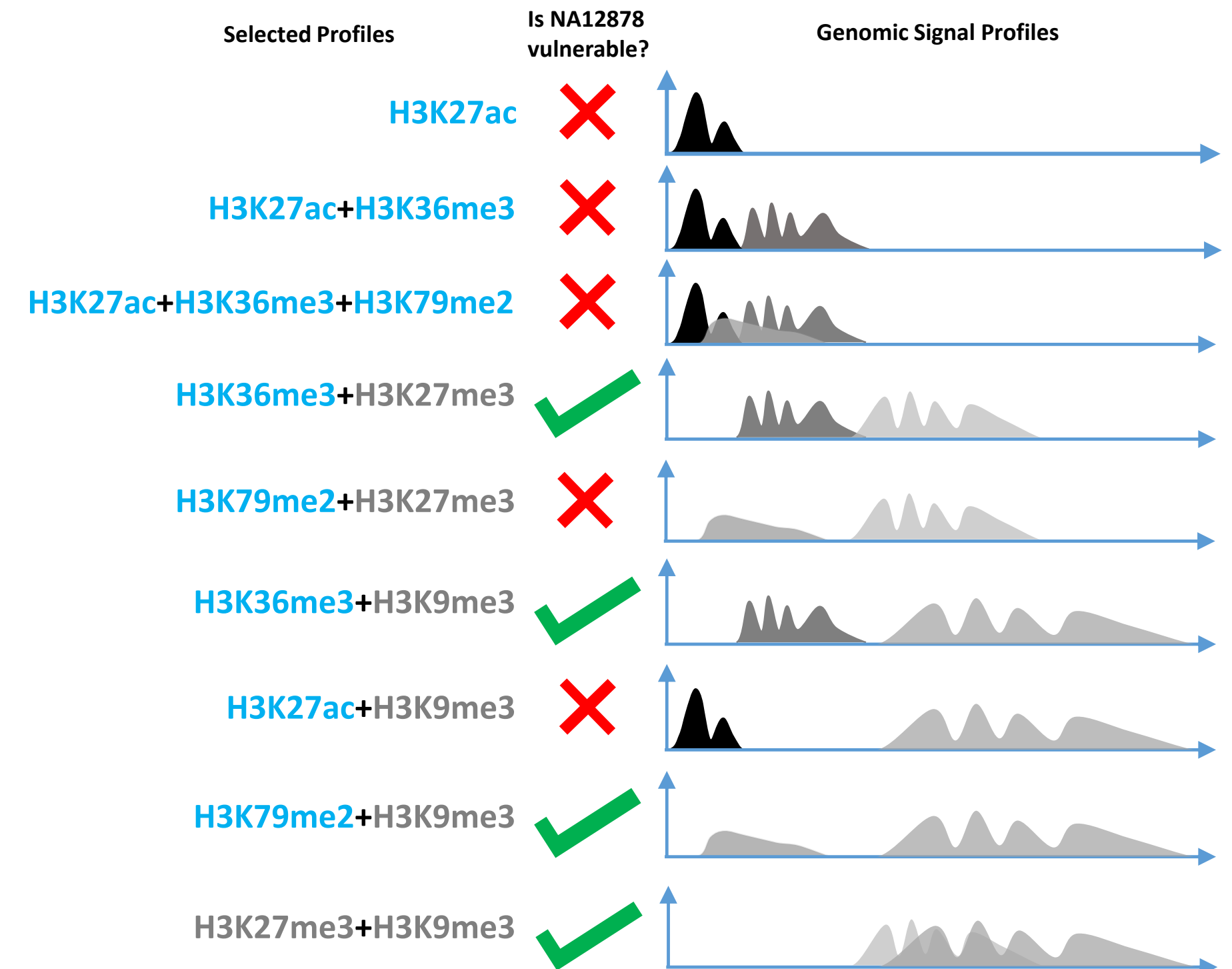
Kasowski



Kilpinen



e) Combinations of datasets imply different vulnerability levels (known variants, variant existence)



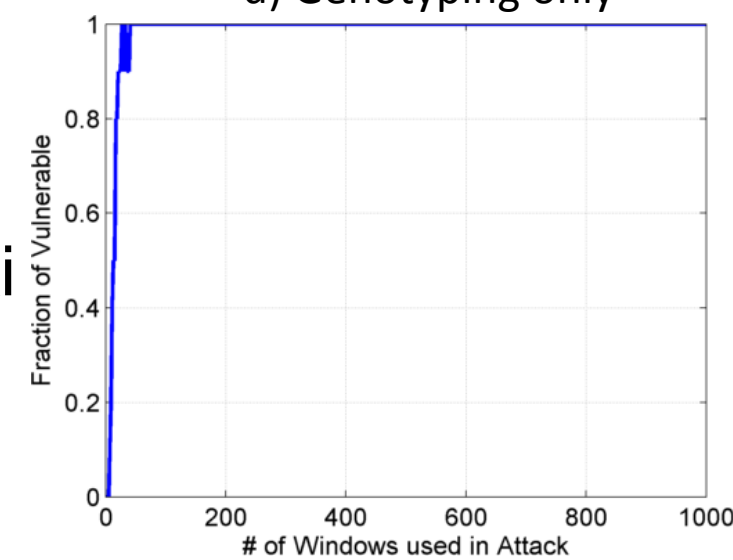
c,d) Extremity Attack Accuracy:

c) Genotyping Only

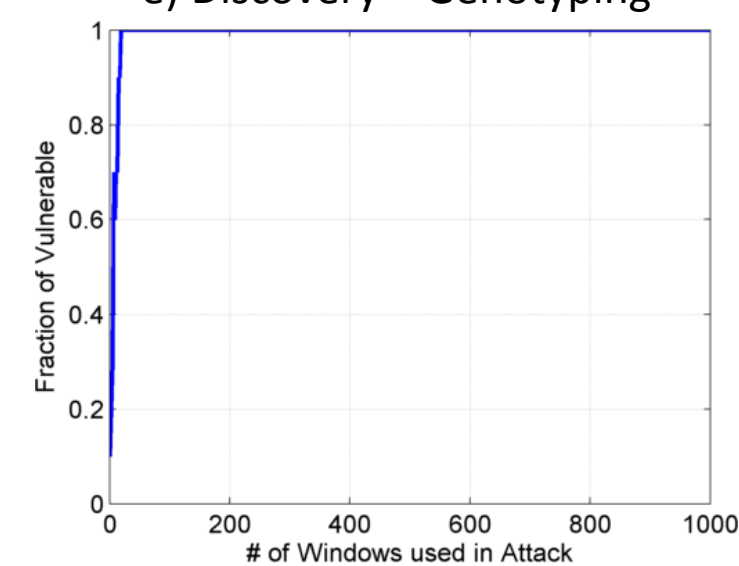
d) Joint Discovery and Genotyping

d) Genotyping only

Kasowski



e) Discovery + Genotyping



Kilpinen

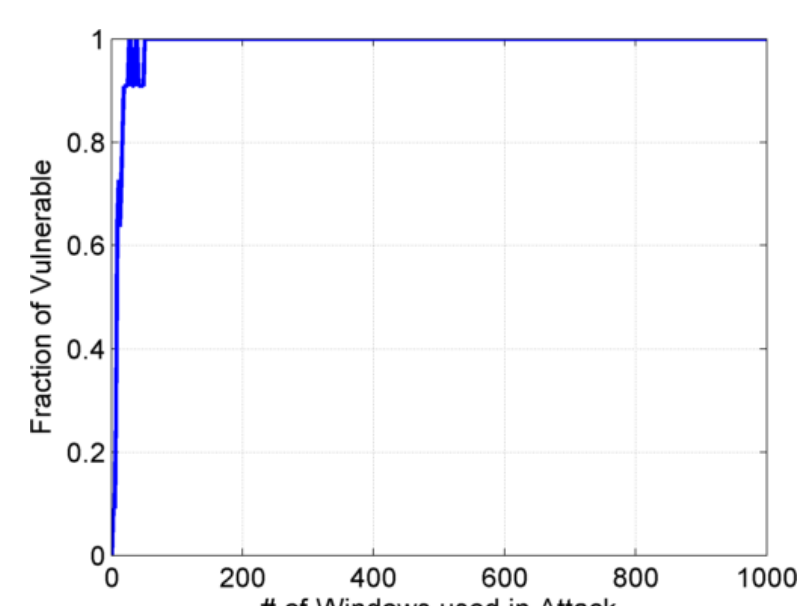
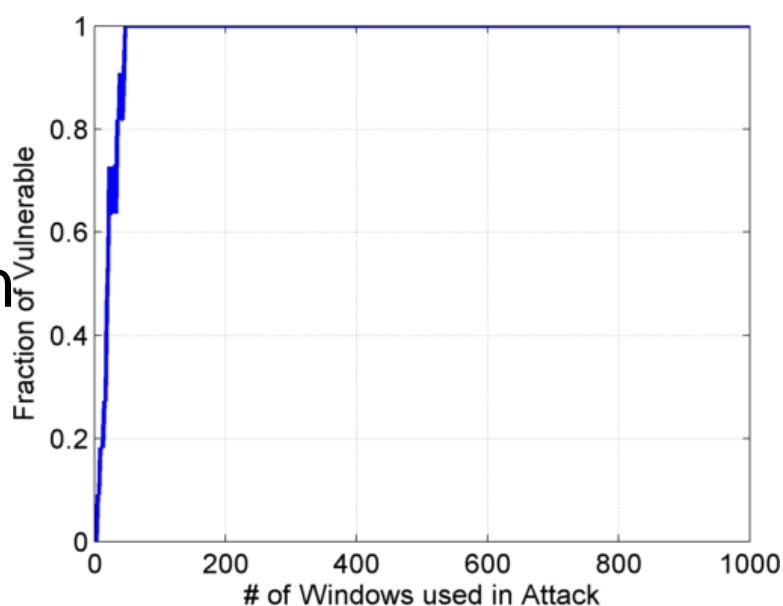
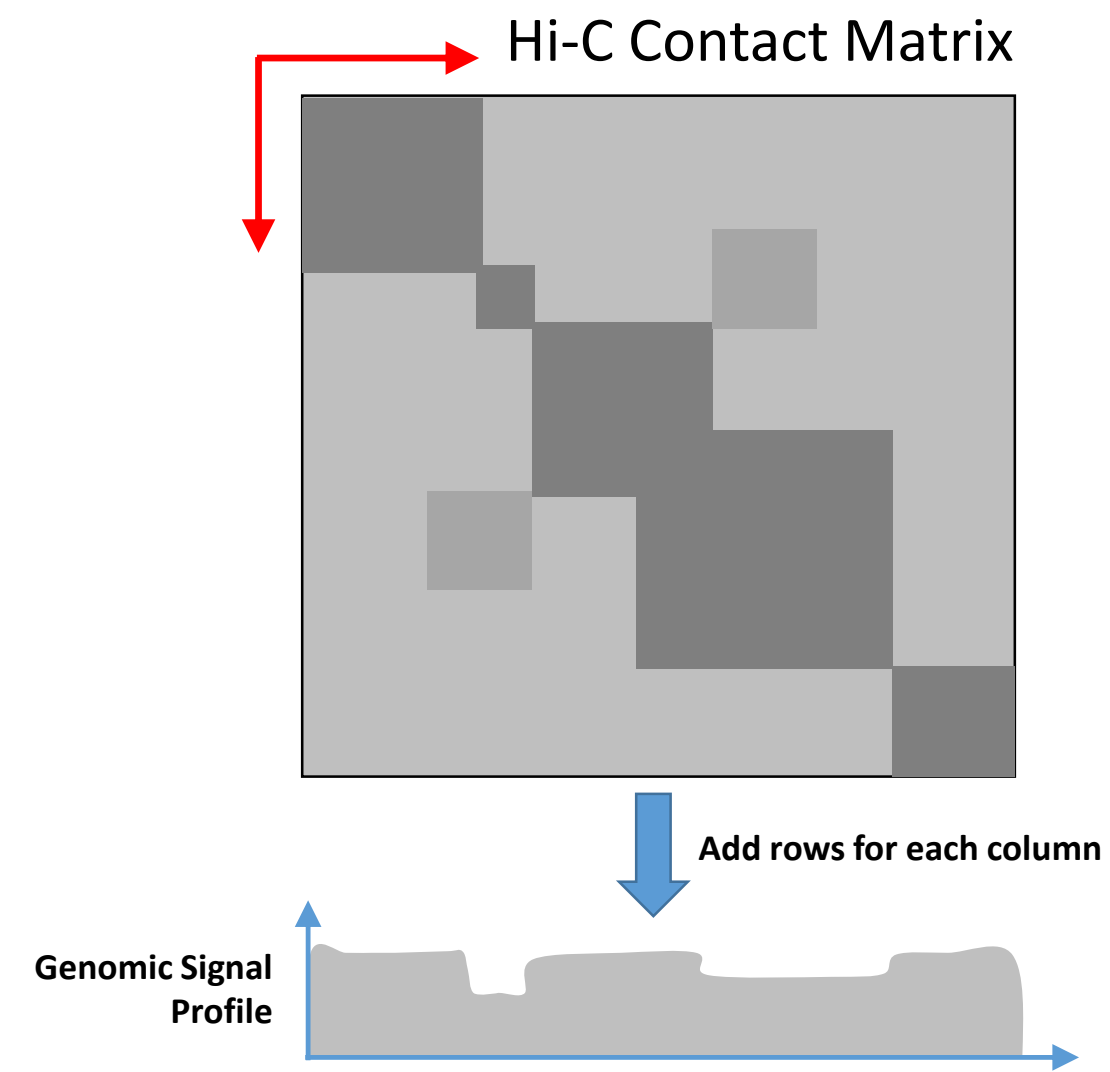


Fig 5

a) Hi-C analysis b) Per SV ICI-\Pi_GW estimates: NA12878 Hi-C data (different resolutions)



Resolution (kbs)	Is NA12878 vulnerable among 1kG samples?
1	✓
5	✓
10	✓
25	✗
50	✗

Supplementary Figures

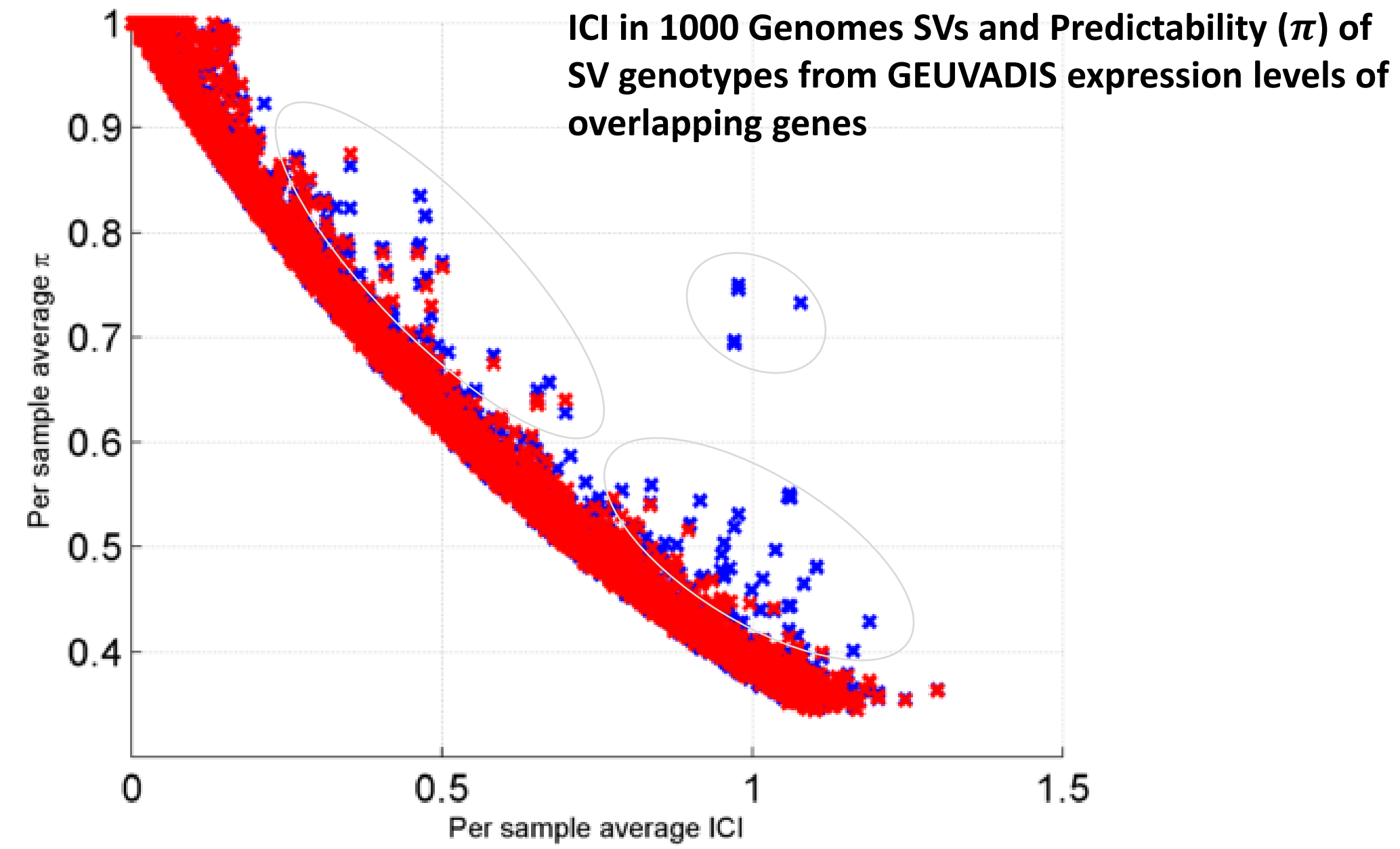
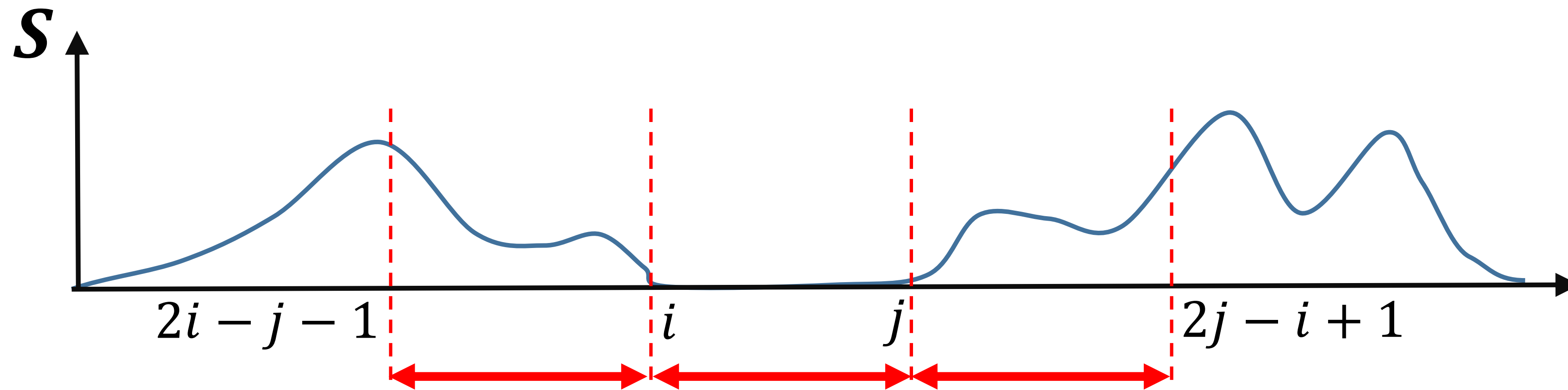
b) π_{SW} vs ICI for 1kG Structural Variants

Fig S2 Features for genotyping deletions



Left
Neighbor

Deletion

Right
Neighbor

$$\bar{S}[2i-j-1, i-1]$$

$$\bar{S}[i, j]$$

$$\bar{S}[j+1, 2j-i+1]$$

$\eta_{[i,j]}$: How balanced are the
neighbor signal levels?

$\rho_{[i,j]}$: How deep is the dip in the
signal?

$\tau_{[i,j]}$: How high are the neighbor
signal levels?

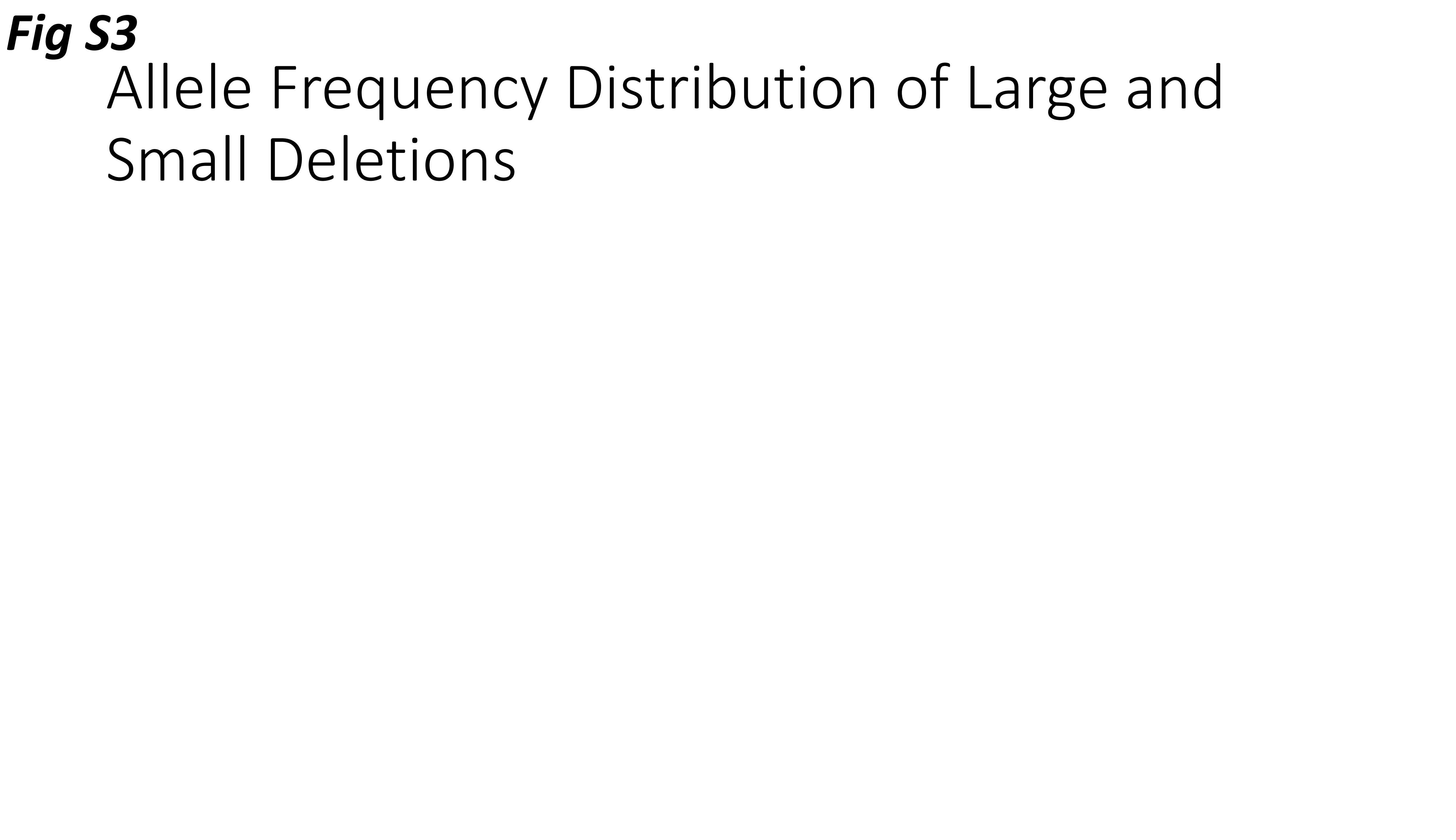


Fig S3

Allele Frequency Distribution of Large and Small Deletions

Fig S4

a)

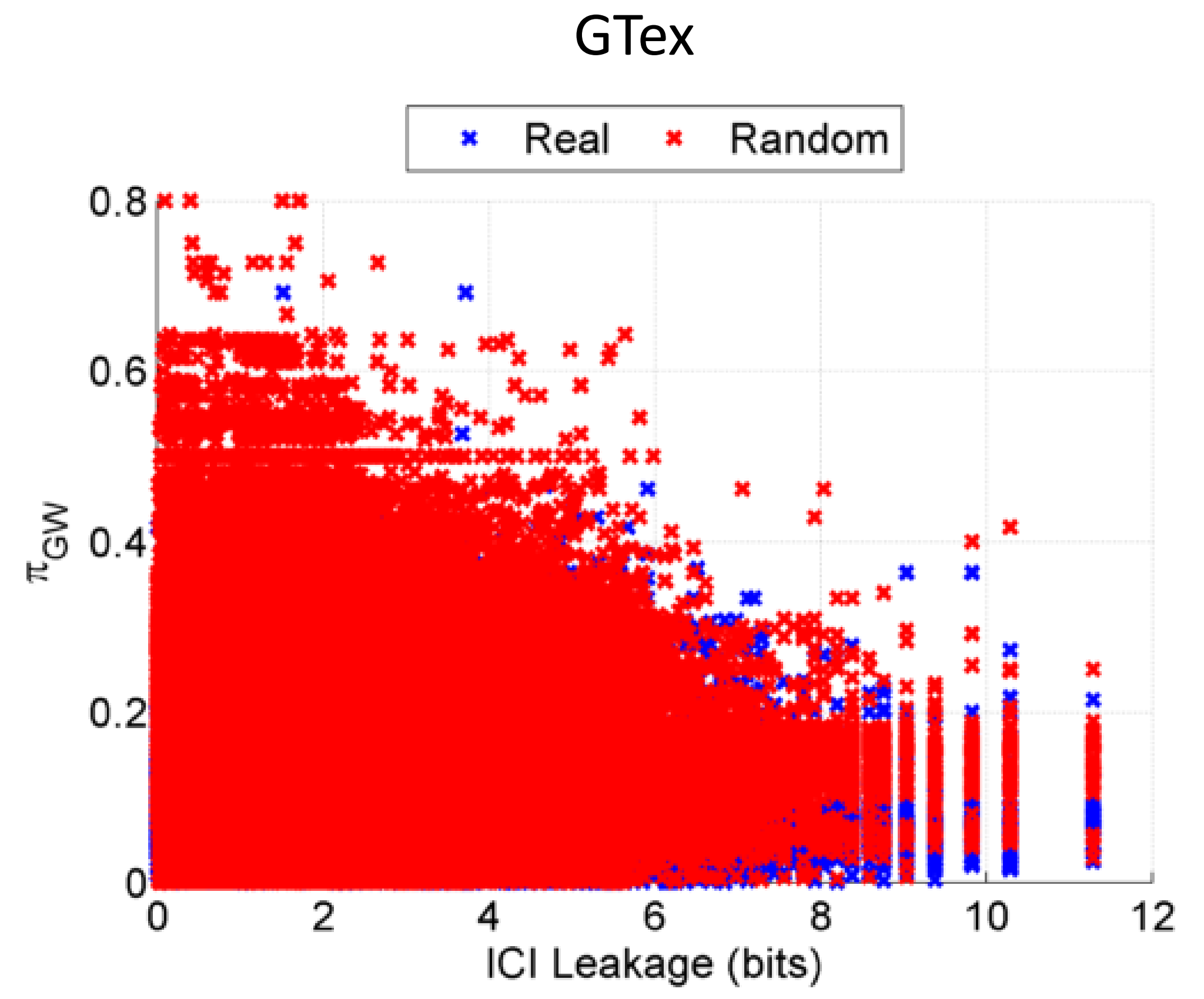
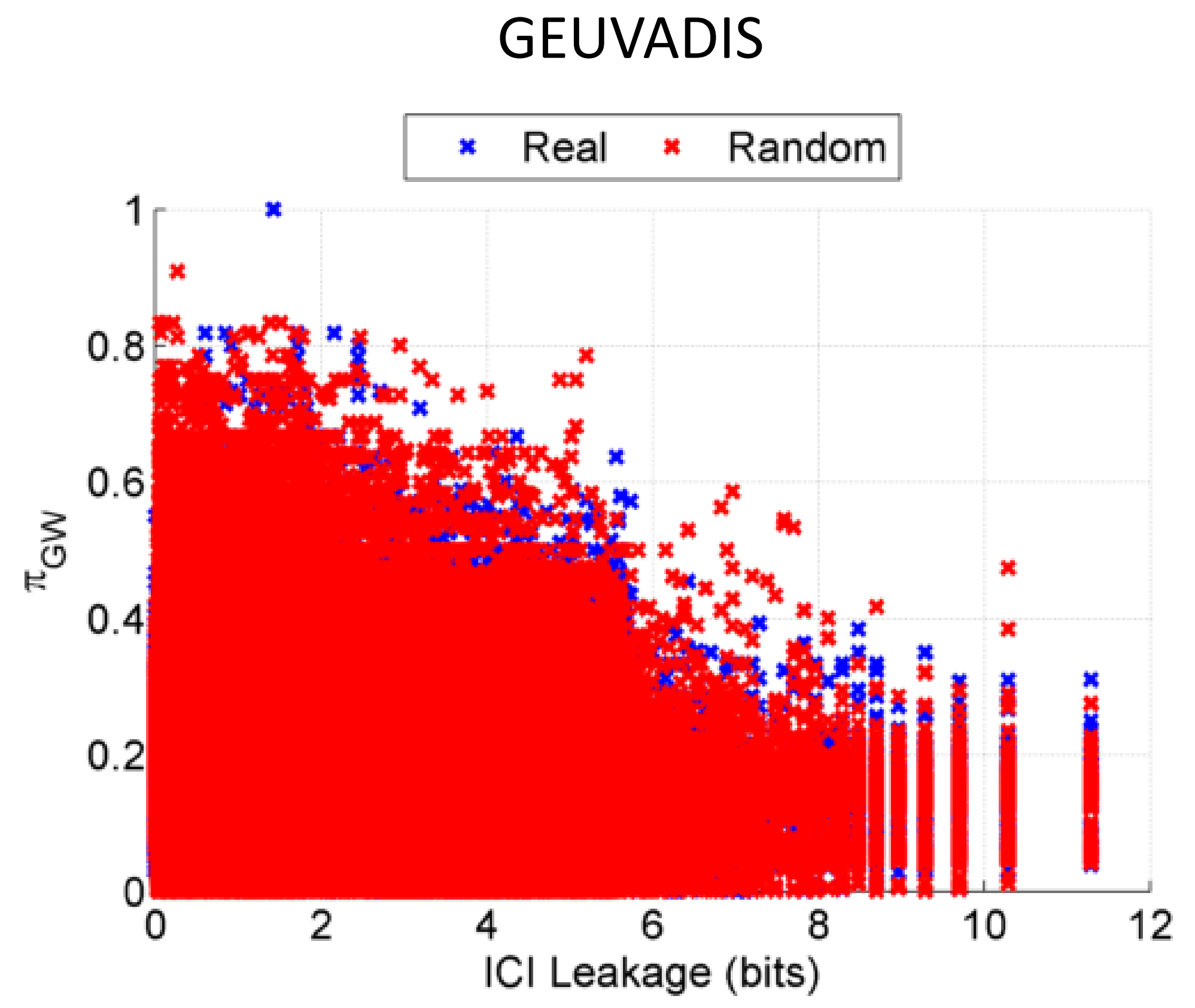


Fig 1

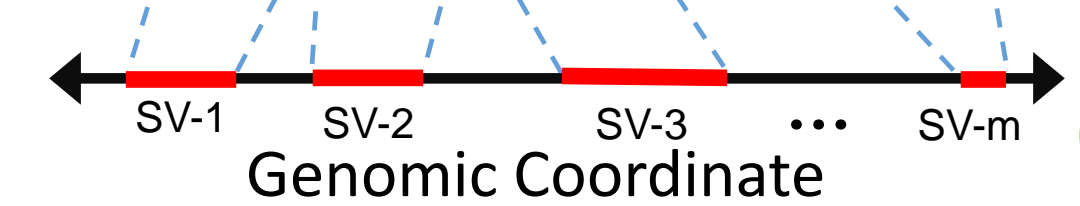
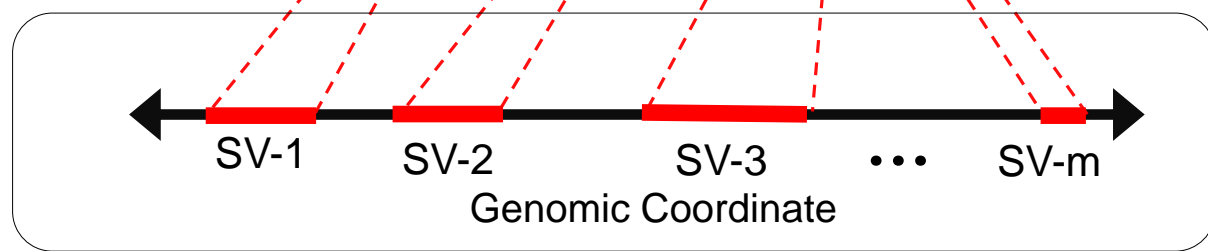
1 Genomewide Signal Profile Dataset

Sample ID	Genome-wide Signal Profiles	HIV Status
<i>PIND-1</i>		+
<i>PIND-2</i>		-
⋮	⋮	⋮
<i>PIND-n</i>		+

2 Structural Variation Genotype Dataset

Sample ID	Genomewide SV Genotypes				
	SV-1	SV-2	SV-3	⋯	SV-m
<i>GIND-1</i>	0	1	2	⋯	0
<i>GIND-2</i>	2	0	2	⋯	1
<i>GIND-3</i>	0	1	1	⋯	0
⋮	⋮	⋮	⋮	⋮	⋮
<i>GIND-K</i>	1	2	2	⋯	2

3 Genomic Deletion Panel Dataset



1 Genomic Deletion Genotyping

Sample ID	Genomewide SV Genotypes					HIV Status
	SV-1	SV-2	SV-3	⋯	SV-m	
<i>PIND-1</i>	0	0	2	⋯	2	+
<i>PIND-2</i>	2	0	X	⋯	0	-
⋮	⋮	⋮	⋮	⋮	⋮	⋮
<i>PIND-n</i>	0	X	X	⋯	0	+

2 Genotype Matching

Phenotype Sample ID	Genotype Sample ID	HIV Status	Real/Predicted SV Genotypes				
			SV-1	SV-2	SV-3	⋯	SV-m
<i>PIND-1</i>	<i>GIND-2</i>	+	0/0	1/0	2/2	⋯	0/2
<i>PIND-2</i>	<i>GIND-1</i>	-	0/2	1/0	2/X	⋯	0/0
⋮	⋮	⋮	⋮	⋮	⋮	⋮	⋮
<i>PIND-n</i>	<i>GIND-3</i>	+	0/0	1/X	1/X	⋯	0/0

Genotyped Deletions

Fig 1

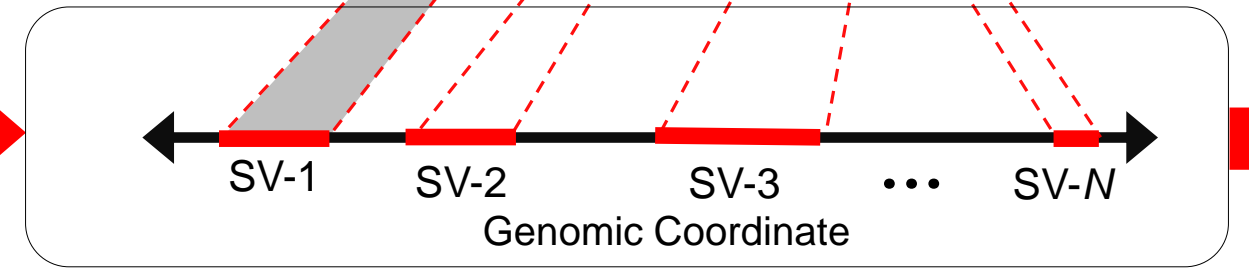
1 Genomewide Signal Profile Dataset

Sample ID	Genome-wide Signal Profiles	HIV Status
<i>PIND-1</i>		+
<i>PIND-2</i>		-
⋮	⋮	⋮
<i>PIND-n</i>		+

2 Structural Variation Genotype Dataset

Sample ID	Genomewide SV Genotypes				
	SV-1	SV-2	SV-3	⋯	SV-m
<i>GIND-1</i>	0	1	2	⋯	0
<i>GIND-2</i>	2	0	2	⋯	1
<i>GIND-3</i>	0	1	1	⋯	0
⋮	⋮	⋮	⋮	⋮	⋮
<i>GIND-K</i>	1	2	2	⋯	2

1 Genomic Deletion Discovery



Discovered Deletion Panel Dataset

Sample ID	Genomewide SV Genotypes					HIV Status
	SV-1	SV-2	SV-3	⋯	SV-N	
<i>PIND-1</i>	0	0	2	⋯	2	+
<i>PIND-2</i>	2	0	X	⋯	0	-
⋮	⋮	⋮	⋮	⋮	⋮	⋮
<i>PIND-n</i>	0	X	X	⋯	0	+

Genotyped Deletions

2 Genomic Deletion Genotyping

3 Comparison of Deletion Loci and Genotype Matching

Phenotype Sample ID	Genotype Sample ID	HIV Status	Real/Predicted SV Genotypes				
			Del-1	Del-2	Del-3	⋯	Del-N
<i>PIND-1</i>	<i>GIND-2</i>	+	0/0	1/0	2/2	⋯	0/2
<i>PIND-2</i>	<i>GIND-1</i>	-	0/2	1/0	2/X	⋯	0/0
⋮	⋮	⋮	⋮	⋮	⋮	⋮	⋮
<i>PIND-n</i>	<i>GIND-3</i>	+	0/0	1/X	1/X	⋯	0/0

