SCISCO GENETICS®
Genotyping the Immune System

ScisGo-KIR-v3

Killer Ig-Like Receptor (KIR) haplotype and allele typing with ScisGo-KIR-v3

ScisGo-KIR-v3 is a system that allows for the determination of gene copy number and haplotype as well as targeted KIR gene allele determination. Gene copy number is determined by comparisons of amplicon-derived read numbers between distinct KIR exon/loci. Haplotypes are deduced from gene copy number according to established reference haplotypes. Allele determination is made by mapping of individual exon sequences as illustrated in Fig. 1 to the IMGT/KIR reference database supplemented with the Scisco Genetics 1000 genomes reference data resource (Fig. 2).

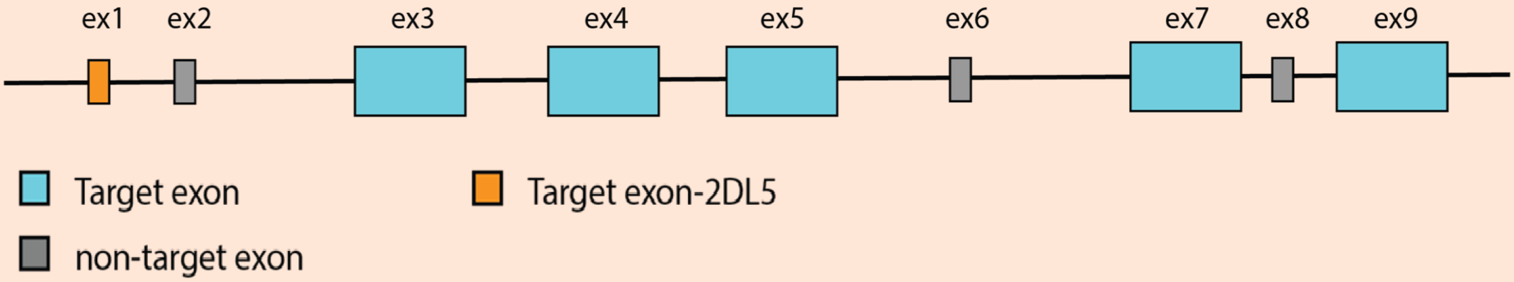


Figure 1. Targeted short-read sequencing of KIR loci. Exons are targeted in multiplex amplicon reactions as indicated in the color key using inter-gene amplicons that span exons from multiple genes.

ScisGo-KIR-v3 was applied to obtain KIR haplotype and partial resolution allele types on 2,542 of the 1000 genomes samples representing 25 ancestral populations. This level of sequencing leaves a level of ambiguity in the allele data due to polymorphism in unsampled exons that can be used as is or if needed solved through long-read genomic sequencing. Haplotypes were deduced from exon sequence reads (relative frequencies). The most significant finding from the 1000 genomes data is the variation of haplotype frequency among different populations (Fig. 2), including a total of 16 rare haplotypes that our system predicts have an as yet undetermined structure.

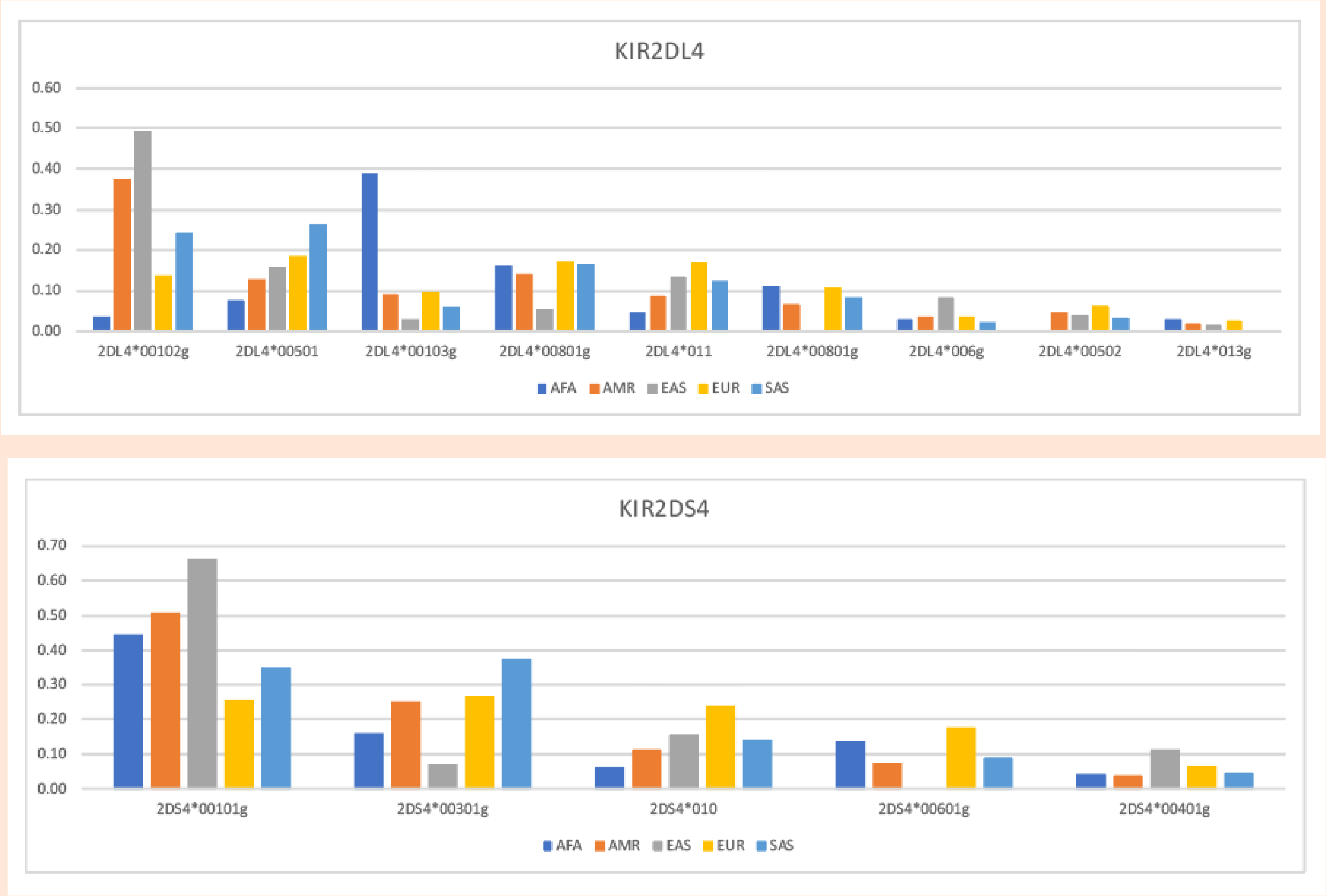


Figure 2a. Allele frequencies for KIRDS4 and KIR2DL4 summarized for the population groups. KIR allele typing is available for most KIR genes.

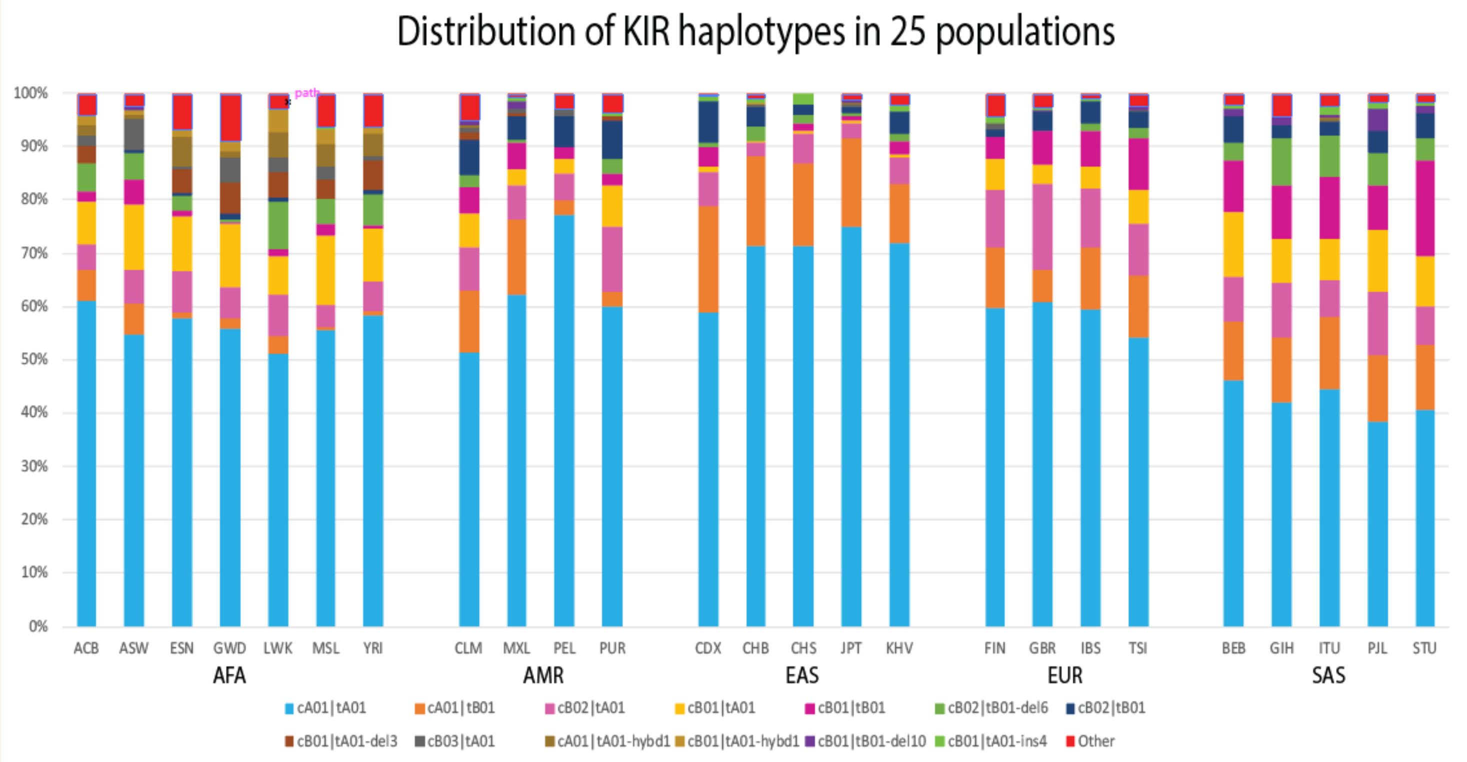
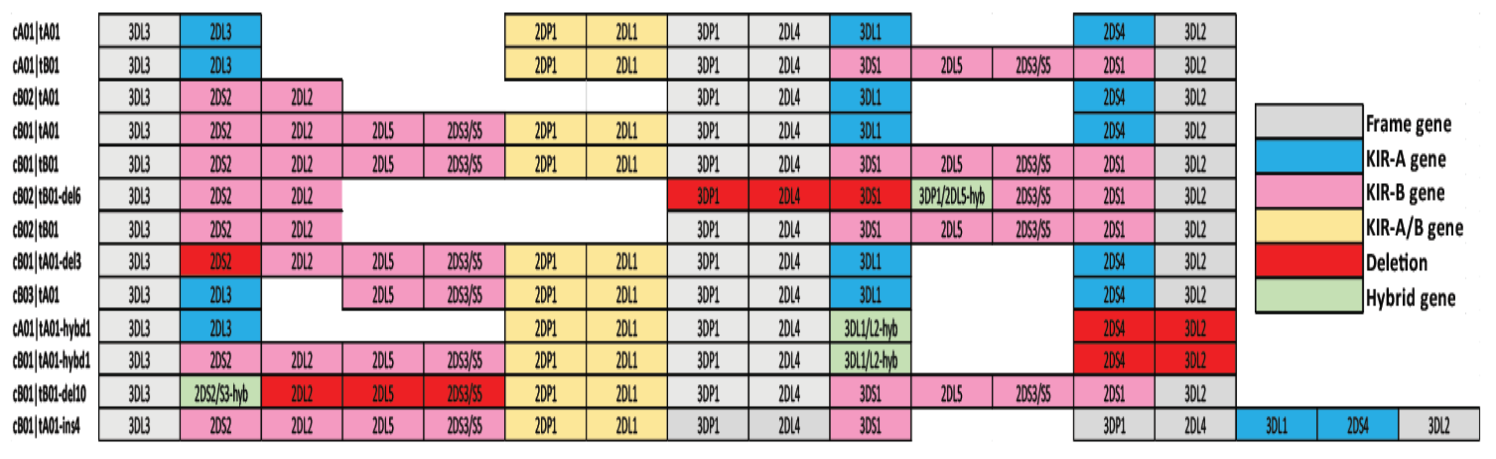


Figure 2b. KIR haplotype and allele frequencies in the twenty-five 1000 genomes populations. (*top*) gene content diagrams of 13 common KIR haplotypes measured in the bar graphs



cA01|tA01	3DL3	2DL3				2DP1	2DL1	3DP1	2DL4	3DL1			2DS4	3DL2			
cA01|tB01	3DL3	2DL3				2DP1	2DL1	3DP1	2DL4	3DS1	2DL5	2DS3/S5	2DS1	3DL2			
cB02|tA01	3DL3	2DS2	2DL2					3DP1	2DL4	3DL1			2DS4	3DL2			
cB01|tA01	3DL3	2DS2	2DL2	2DL5	2DS3/S5	2DP1	2DL1	3DP1	2DL4	3DL1			2DS4	3DL2			
cB01|tB01	3DL3	2DS2	2DL2	2DL5	2DS3/S5	2DP1	2DL1	3DP1	2DL4	3DS1	2DL5	2DS3/S5	2DS1	3DL2			
cB02|tB01-del6	3DL3	2DS2	2DL2					3DP1	2DL4	3DS1	3DP1/2DL5-hyb	2DS3/S5	2DS1	3DL2			
cB02|tB01	3DL3	2DS2	2DL2					3DP1	2DL4	3DS1	2DL5	2DS3/S5	2DS1	3DL2			
cB01|tA01-del3	3DL3	2DS2	2DL2	2DL5	2DS3/S5	2DP1	2DL1	3DP1	2DL4	3DL1			2DS4	3DL2			
cB03|tA01	3DL3	2DL3		2DL5	2DS3/S5	2DP1	2DL1	3DP1	2DL4	3DL1			2DS4	3DL2			
cA01|tA01-hybd1	3DL3	2DL3				2DP1	2DL1	3DP1	2DL4	3DL1/L2-hyb			2DS4	3DL2			
cB01|tA01-hybd1	3DL3	2DS2	2DL2	2DL5	2DS3/S5	2DP1	2DL1	3DP1	2DL4	3DL1/L2-hyb			2DS4	3DL2			
cB01|tB01-del10	3DL3	2DS2/S3-hyb	2DL2	2DL5	2DS3/S5	2DP1	2DL1	3DP1	2DL4	3DS1	2DL5	2DS3/S5	2DS1	3DL2			
cB01|tA01-ins4	3DL3	2DS2	2DL2	2DL5	2DS3/S5	2DP1	2DL1	3DP1	2DL4	3DS1			3DP1	2DL4	3DL1	2DS4	3DL2

Figure 2c. Showing the percentage of each haplotype structure within each of twenty-five 1000 genomes populations according to the color coding immediately beneath the graphs. Haplotype designation ‘other’ includes 68 rare haplotypes including 16 of unknown structure.