





Supplementary figure S1 Separation of FCGR3 copy number alleles

Pulsed-field gel electrophoresis of four *Sbfl*-digested DNA samples of different *FCGR3* total copy number, showing separation of 3 copy sample into constituent 2 and 1 copy alleles, and 5 copy sample into 3 and 2 copy alleles. Note that the size marker used was concatenated lambda phage DNA (New England Biolabs), which is known to migrate slightly faster than digested human genomic DNA. The absolute sizes of the bands are therefore overestimates of the real size, but the difference between 1 copy, 2 copy and 3 copy bands is consistent with length differences of 82kb, representing a gain of a full repeat unit.



Supplementary figure S2a Segregation analysis of CEPH families

Families 1418 is shown, the order of *FCGR3B*-HNA1a and *FCGR3B*-HNA1b on *FCGR3B* duplicated chromosomes is not known.

Human Mutation

Supplementary figure S2b Segregation analysis of CEPH families

Families 1347 is shown, the order of FCGR3B-HNA1a and FCGR3B-HNA1b

FCGR3B-HNA1a FCGR3B-HNA1b

on FCGR3B duplicated chromosomes is not known.

FCGR3A



Supplementary figure S2c Segregation analysis of CEPH families

Families 1340 is shown, the order of *FCGR3B*-HNA1a and *FCGR3B*-HNA1b on *FCGR3B* duplicated chromosomes is not known.



Supplementary figure S3 Association of SNPs with FCGR3 CNV

Between 342 and 391 SNPs from Phase II HapMap project in the 200kb either side of the *FCGR3* CNV region were correllated with *FCGR3* CNV status, specifically *FCGR3A* copy number, *FCGR3B* copy number and HNA1 genotype, in different populations. Results are plotted using the Genome Graphs function of the UCSC Genome Browser March 2006 (build 36.1, hg18), with colours corresponding to population: blue = Yoruba from Ibadan, Nigeria, purple = Chinese from Beijing, black = Japanese from Tokyo, red = CEPH Europeans from Utah (mostly of Scandanavian or English origin). The y-axis for each graph is from r²=0 to r²=0.5. Regions identified as segmental duplications are omitted from the analysis. Beneath

these graphs RefSeq genes are shown, followed by SNP-SNP pairwise linkage disequilibrium, where darker red indicates strong linkage disequilibrium as measured by r^2 .