Example of the advantages of D' over r as a general measure of the strength of LD

Values of $\hat{D}'$ and $\hat{r}$ for nine pairs of SNPs on the short arm of human chromosome 11 from a sample on 45 Japanese individuals in Tokyo, Japan (JPT), are shown in suppl. table 7. It can be seen that $\hat{D}'$ and $\hat{r}$ provide a different perspective on the amount of LD between SNPs and its relationship with physical distance along this region of human chromosome 11. Thus, $\hat{D}'$-values are always higher than those of $\hat{r}$ and their mean values (±SE) over pairs of SNPs are 0.58 ± 0.10 and 0.25 ± 0.07, respectively, which is consistent with most previous studies describing patterns of LD in populations [9-14]. This research shows that the relative efficiency of the two LD measures is very similar when their differences in range are considered. Therefore, the estimates of D' and r are apparently different because both LD measures also operate on different scales. For example, values of $\hat{D}'$ and $\hat{r}$ for the SNP pair rs3813727 and rs3813726 (top of suppl. table 7) seem to be very different (+1.0 and +0.16, respectively) but they are the maximum possible values of their respective ranges of positive sign. Overall, D' has the advantage of providing a more direct and reliable information on the strength of LD than r because it always ranges from -1.0 to +1.0. On the other hand, estimates of D' (in absolute values) reveal a strong negative relationship of the strength of LD between pairs of SNPs with physical distance (r_s= -0.90, P= 0.001, one-sided test), a consistent pattern with the prediction of higher LD when there is lower physical distance. In contrast, $\hat{r}$-values for pairs of SNPs are not significantly correlated with physical distance (r_s= -0.12, P= 0.38, one-sided test). This example illustrates the advantage of D' over r to identify gradients of LD along chromosomes, which is very useful for gene mapping of quantitative characters and human diseases (see [1, 2] and references therein).

Supplementary Table 7. LD between SNPs located on the short arm of human chromosome 11 in a sample of Japanese individuals[a]

Pair of SNPs	Distance (kb)	$\hat{p}$	$\hat{q}$	$\hat{D}'$	$\hat{r}$	Range of $\hat{r}$ (min, max)[b]
rs3813727 x rs3813726	0.08	0.69	0.99	+1.0	+0.16	-0.07, +0.16
rs3813726 x rs7948416	0.44	0.99	0.86	-1.0	-0.05	-0.05, +0.26
rs7927539 x rs6578561	0.45	0.91	0.93	+0.81	+0.69	-0.09, +0.86
rs4320977 x rs11036405	0.84	0.71	0.92	-0.76	-0.15	-0.19, +0.46
rs6578565 x rs6578568	2.51	0.91	0.78	+0.63	+0.37	-0.17, +0.59
rs3752382 x rs3759074	3.84	0.97	0.86	+0.54	+0.25	-0.08, +0.45
rs2187610 x rs7936823	4.76	0.80	0.64	+0.48	+0.34	-0.37, +0.66
rs7927539 x rs7934373	7.09	0.91	0.73	+0.20	+0.11	-0.19, +0.51
rs7934373 x rs11036227	17.73	0.73	0.68	+0.13	+0.12	-0.42, +0.89

[a] Source of data: HapMap Project database (http://hapmap.ncbi.nlm.nih.gov/; HapMap Genome Browser release#24; Phase 1 & 2 – full dataset); [b] min and max are the minimum and maximum values of the range of $\hat{r}$, respectively, which were computed as described in 'Methods' section.