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 (\pm SE) over pairs of SNPs are 0.58 \pm 0.10 and 0.25 \pm 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).

Pair of SNPs	Distance (kb)	<i>p</i>	ĝ	$\hat{D}^{'}$	ŕ	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

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

^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.