Transmission distortion analysis of only those hcSNPs found in ultraconserved elements (100% conservation) using trio data from FHS. Highly constrained SNPs found in elements conserved at 100% showed a nominally significant under-transmission (N = 33; proportion of derived allele transmission = 48.1%; average difference = -6.57, $P = 0.048$) when compared to those found in elements conserved at 99% and 98% (N = 177 and 229; proportion of derived allele transmission = 49.7% and 50.2%; average difference = -0.91 and 0.18, $P = 0.34$ and 0.54, respectively; data not shown). The less frequent transmission of the derived alleles at hcSNPs in 100% elements is unlikely to be explained by a stronger evolutionary constraint among SNPs in the 100% conserved elements, since we focused on the hcSNPs for this analysis and the distribution of the conservation scores for hcSNPs in the 100% conserved elements did not significantly differ from the distribution of conservation scores in the 98% and 99% conserved elements (mean SNP conservation score = 0.981 vs. 0.974, $P = 0.166$ by two-tailed Mann-Whitney test).

(Left) Distortion in the transmission of the derived alleles of hcSNPs from heterozygous parents to offspring was measured by subtracting the ancestral allele transmissions from the derived allele transmission for each SNP. (Right) The average proportion of derived allele transmission when SNPs were stratified into deciles by DAF. Error bars reflect the standard error in the proportion of derived allele transmissions within the decile.