

Supplemental Fig. S4. Genetic architecture of genome-wide susceptibility loci significantly associated with serum 25-hydroxyvitamin D concentration-related traits. (A) The lead single nucleotide polymorphisms (SNPs) are red or blue diamonds. All other SNPs show which of the lead SNPs they are in highest linkage disequilibrium (LD) with, with the color matching the lead SNP. The extent of LD with the lead SNP is shown by a gradient of color. Regional plots for rs11723621 SNP (chr4:71749645, group-specific component [GC]) are indicated by red dots and those for rs7041 (chr4:71752617, GC) are indicated by blue dots. (B, C, D) The lead SNPs are shown as purple diamonds. Each SNP is plotted as a circle along the chromosomal position, and LD between the lead SNP and the other SNPs is colored as a scale from low (blue) to high (red) or is colored gray if LD information was not available in the 1,000 genome phase 3 East Asian samples. Regional plots for rs11023332 (phosphodiesterase 3B [PDE3B]), for rs12803256 (actin epsilon 1, pseudogene [ACTE1P]), and for rs3831470 (glutaminedependent NAD(+) synthetase [NADSYNI]). Color coding of the other plotted SNPs in the region represents LD (r2) with the top associated SNP that was calculated using 1,000 Genomes from East Asians samples. The recombination rate for the 1,000 Genomes from East Asians samples is indicated by the blue line and the right-hand y-axis. The x-axis shows the chromosomal position of the SNPs and genes based on NCBI Human Genome Build 38 coordinates. ALG1L9P, asparagine-linked glycosylation 1-like 9, pseudogene; CALCA, calcitonin-related polypeptide alpha; CALCB, calcitonin-related polypeptide beta; COPB1, coat complex subunit beta 1; CYP2R1, cytochrome P450 2R1; DHCR7, 7-dehydrocholesterol reductase; KRTAP5-7, keratin-associated protein 5-7; KRTAP5-8, keratin-associated protein 5-8; KRTAP5-9, keratin-associated protein 5-9; KRTAP5-10, keratin-associated protein 5-10; KRTAP5-11, keratin-associated protein 5-11; GC, group-specific component; INSC, inscuteable spindle orientation adaptor protein; FAM86C1, family with sequence similarity 86 member C1, pseudogene; FLJ42102, uncharacterized LOC399923; MIR6754, microRNA 6754; NPFFR2, neuropeptide FF receptor 2; PSMA1, proteasome 20S subunit alpha 1; RRAS2, RAS related protein 2; SHANK2, SH3 and multiple ankyrin repeat domains protein 2; SLC4A4, solute carrier family 4 member 4; ZNF705E, zinc finger protein 705E.