Supplemental Data

Genome-wide Copy-Number-Variation Study Identified a Susceptibility Gene, *UGT2B17*, for Osteoporosis

Tie-Lin Yang, Xiang-Ding Chen, Yan Guo, Shu-Feng Lei, Jin-Tang Wang, Qi Zhou, Feng Pan, Yuan Chen, Zhi-Xin Zhang, Shan-Shan Dong, Xiang-Hong Xu, Han Yan, Xiaogang Liu, Chuan Qiu, Xue-Zhen Zhu, Teng Chen, Meng Li, Hong Zhang, Liang Zhang, Betty M. Drees, James J. Hamilton, Christopher J. Papasian, Robert R. Recker, Xiao-Ping Song, Jing Cheng, and Hong-Wen Deng

Figure S1. CNV Redefined for Association Analyses

There were five major types of CNVRs (illustrated as in Plots A-E) detected in the Chinese GWA sample. Representation of SNP and CNV is denoted in the end after Plot E.

A: All the individual CNVs in a CNVR had the same boundaries. In this case, the CNVR included only one kind of CNV for association analysis.

B: All the individual CNVs in the CNVR had, at most, one SNP difference between their boundaries and CNVR's boundaries at each side. This case was considered the same as A.

C: Some CNVs extended and were involved in regions of some other CNVs.

D: CNVs with regions partially overlapped.

E: CNVR with complex overlapping regions.

For C, D and E, CNVRs were divided into several sub-CNVRs with the same configuration as A or B, thus all the sub-CNVRs contained only one kind of CNV for association analyses.

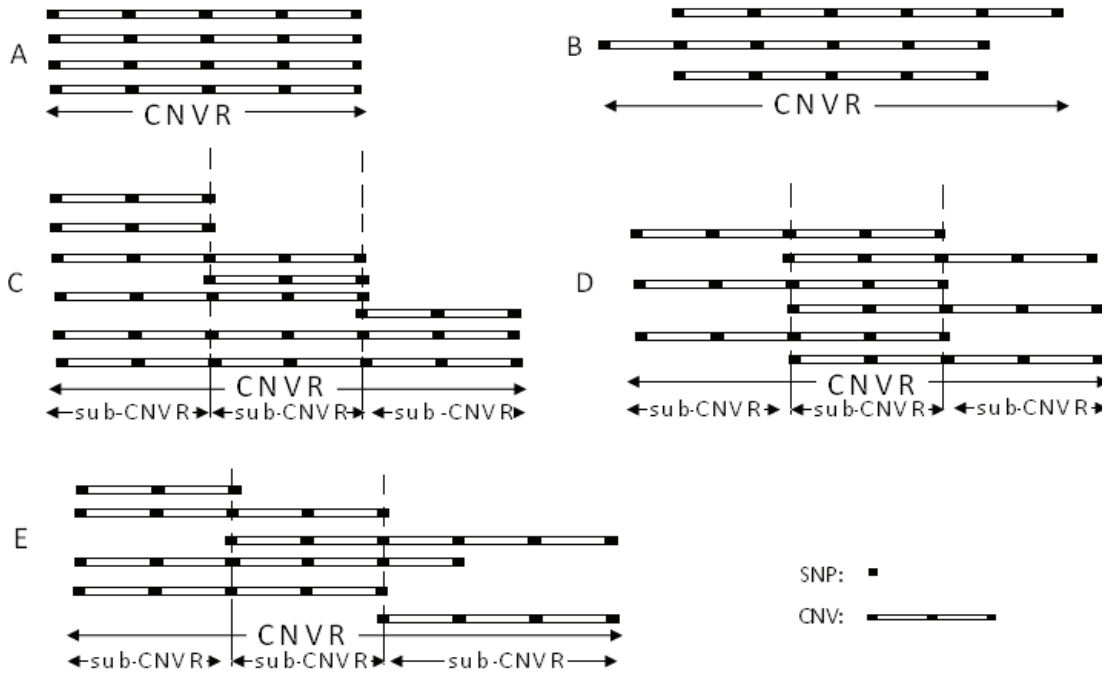

Figure S2. Gene-Specific Marker Locations for *UGT2B17*

UGT2B17 has six exons (1 to 6) indicated by black bars. Marker C is located within exon 1 of *UGT2B17*. Marker E is located within exon 6 of *UGT2B17*. Marker D is located upstream of the transcriptional start site of *UGT2B17*. The amplifications of marker C, D and E were used to assay for the presence of *UGT2B17*. *UGT2B17* deletion is shown above, which indicates the absence of ~150 kb genomic region covering the entire *UGT2B17* gene. Marker J is a deletion marker, whose amplification indicates the absence of *UGT2B17*. Dashed arrows show the locations of primers for deletion marker (J).

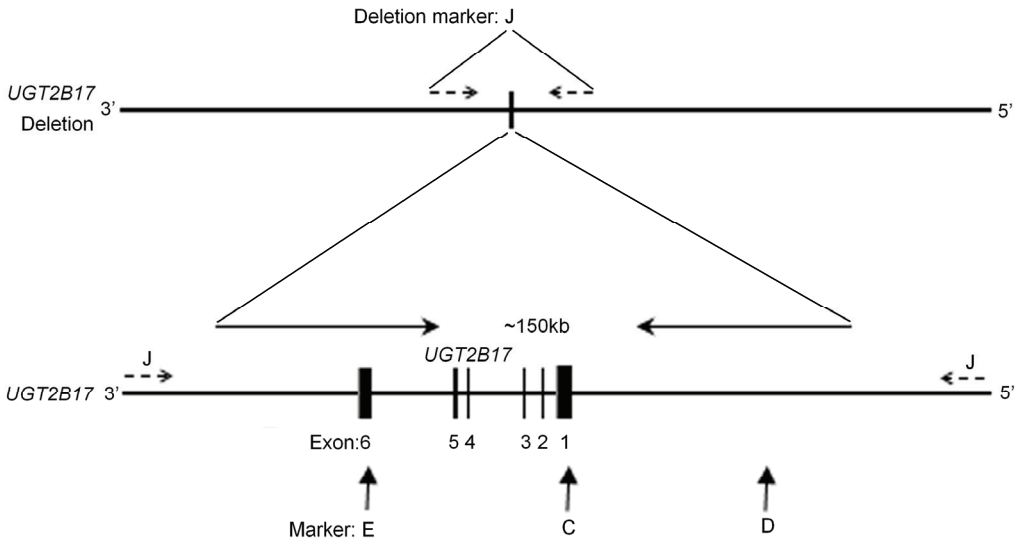

Table S1. Estimation of Gene Copy Number by Real-Time PCR

Genes	Primers Sequence (5' -> 3')	Amplicon (bp)	Gene deletion	*P* value
YTHDC1	F: CCACAGAAAGTAATAATGGGATG	215	No	0.213
	R: TAGGCTAGGAGAAACAGGTCAT			
TMPRSS11E	F: GCTTTCTAGCCCTGTTCCCTA	146	No	0.898
	R: TTGAGTTCCTCTTCCGATGC			
TMPRSS11E2	F: TCTGTCATTGTTGTCTGCTTTC	194	No	0.888
	R: GACCTGCTTCACGACTGTTC			
*UGT2B15*_marker G	F: GATAACAAGTGTTGGGAAGAGTG	329	No	0.572
	R: CTAGGAGTGGACTTGCTGAGAC			
*UGT2B17*_marker C	F: CCTGGAAGAGCTTGTTCAGA	316	Yes	<0.001
	R: CTGCATCTTCACAGAGCTTT			

Marker G: Within exon 1 of *UGT2B15*; marker C: Within exon 1 of *UGT2B17*;

F: Forward Primer; R: Reverse Primer.

P values for significance were estimated by *t*-test comparing average ΔCt values between subjects of different genotypes of CNV 4q13.2 predicted by the CNAT analyses.