



Article

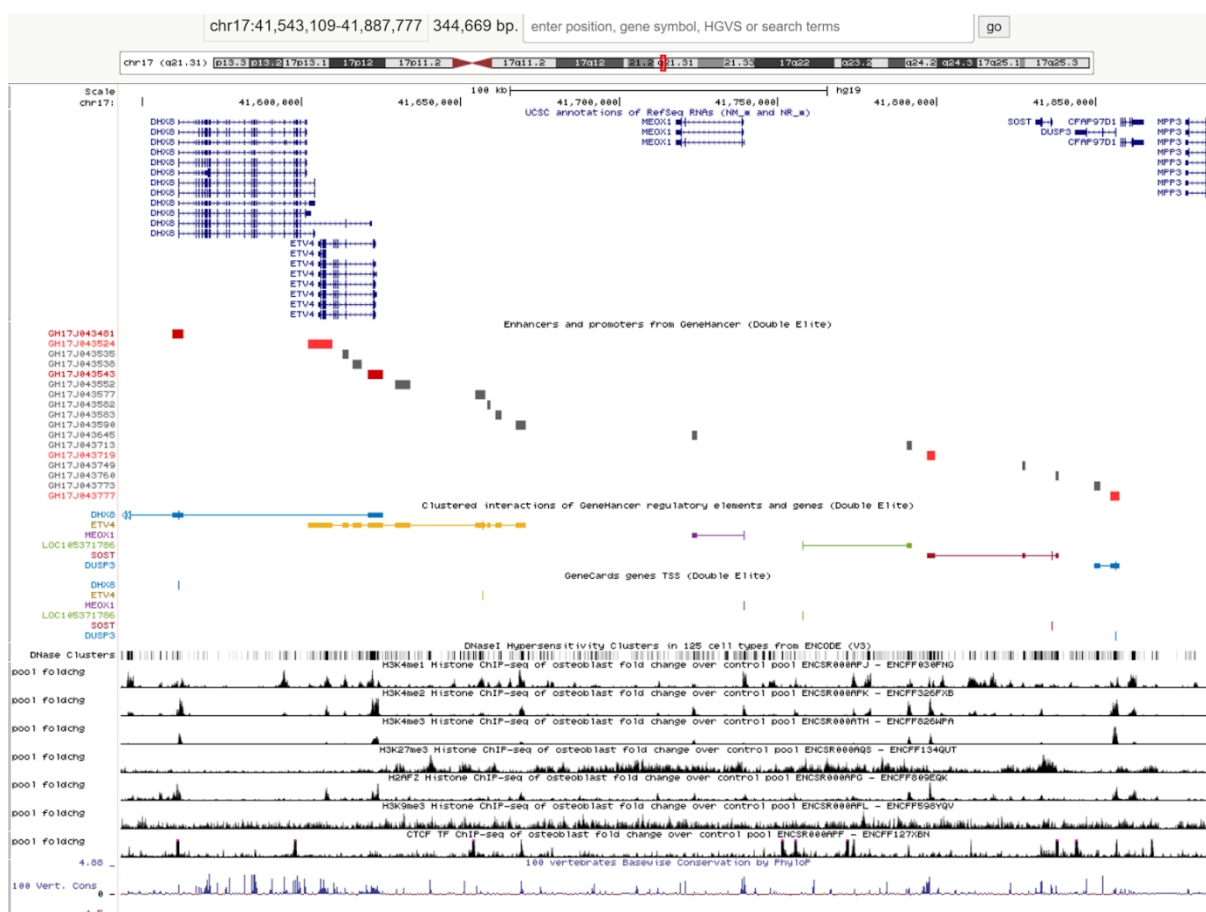
Genetics and Genomics of *SOST*: Functional Analysis of Variants and Genomic Regulation in Osteoblasts

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Supplementary Figure S1. Schema of *SOST* genomic region from the UCSC genome browser with the following tracks (downwards): In red, the promoter regions and in grey the enhancer regions according to GeneHancer track. In different colors, the interaction regions according to GeneHancer track and the TSS for each gene. In black, osteoblast ChIP-seq profiles of H3K4me1, H3K4me3, H3K4me2, CTCF, H3K27ac, H3K9me3 and H2A.Z from ENCODE. In blue, vertebrate conservation.



Supplementary Figure S2. Study of the rs552004150 variant. (A) HBM16 family pedigree, in which 2 individuals (HBM16 and HBM16a; filled symbols) present with the HBM phenotype. The SNP genotype is indicated below the symbols; family members with no information on the bone density phenotype are displayed in white. (B) TRANSFAC result of the ECR5 region. The position of the variant is indicated with a vertical arrow.

Supplementary Table S1. GTEx eQTL data for all variants found in the resequencing of *SOST*.

SNP	Gene	Tissue
rs1237278	<i>SOST</i>	artery-tibial, aorta, coronary; lung; heart-atrial appendage; brain-cortex
	<i>MPP3</i>	thyroid
	<i>NAG3</i>	skin-sun exposed
rs851058	<i>SOST</i>	artery-tibial, aorta, coronary; lung; heart-atrial appendage; thyroid
	<i>C17orf105</i>	brain-cerebellum
	<i>CD300LG</i>	muscle-skeletal
	<i>DUSP3</i>	cells-cultured fibroblasts; whole blood; esophagus-mucosa
	<i>MPP2</i>	skin-sun and not sun exposed
	<i>MPP3</i>	cells-cultured fibroblasts
	<i>NAGS</i>	lung; skin sun exposed
rs10534024	<i>SOST</i>	artery-tibial, aorta, coronary; lung; heart-atrial appendage; brain-cortex, nerve-tibial
	<i>MPP3</i>	thyroid; testis
rs17882143	<i>DBF4B</i>	brain-caudate; thyroid
	<i>RPL27</i>	artery-aorta
rs17886183	<i>DUSP3</i>	whole blood
rs17881550	<i>SOST</i>	artery-tibial, aorta, coronary; heart-atrial appendage; lung; thyroid; nerve-tibial
	<i>AOC2</i>	lung
	<i>C17orf105</i>	brain-cerebellum

<i>CD300LG</i>	muscle-skeletal
<i>DUSP3</i>	esophagus-mucosa
<i>MPP2</i>	skin-sun and not sun exposed
<i>NAGS</i>	skin-sun exposed; lung

Supplementary Table S2. In silico predictions for the 3 SNPs in *SOST*.

SNP	Prediction	Allele	
rs17885799	ESE	C	Two new Exonic Splicing Enhancer (ESE)
-	-	-	miRNA
rs17883310	miRNA	C	hsa-miR-1915-3p hsa-miR-3141 hsa-miR-4685
rs17886183	miRNA	A	hsa-miR-5583-3p

ESE: New Exonic Splicing Enhancer predicted. miRNA: Differences in the miRNA binding site depending on the allele.