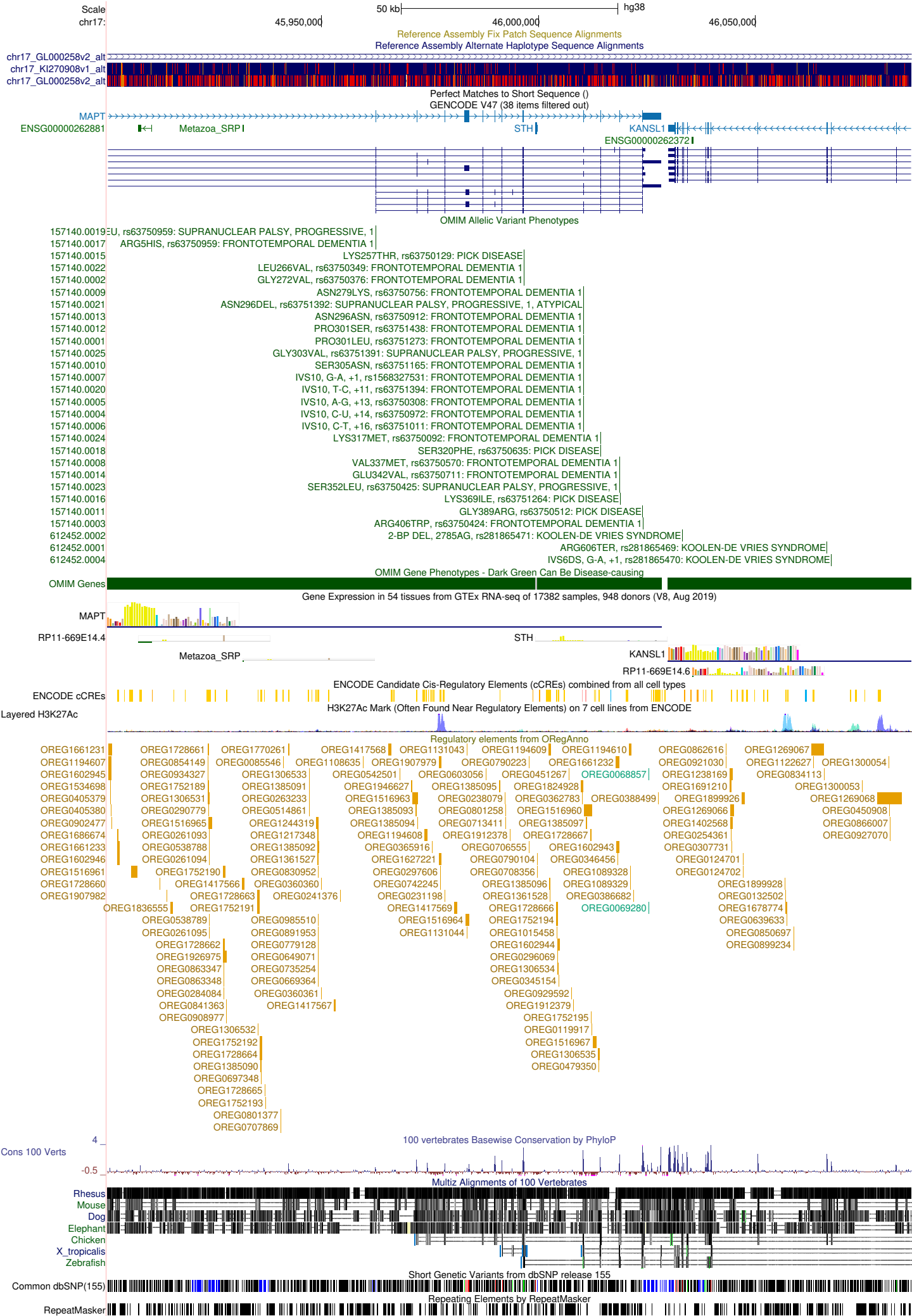
Scale
50 kb
hg38
chr17:
45,950,000
46,000,000
46,050,000
Reference Assembly Fix Patch Sequence Alignments
Reference Assembly Alternate Haplotype Sequence Alignments
chr17_GL000258v2_alt
chr17_KI270908v1_alt
chr17_GL000258v2_alt
Perfect Matches to Short Sequence ()
GENCODE V47 (38 items filtered out)
MAPT
ENSG00000262881
Metazoa_SRP
STH
KANSL1
ENSG00000262372
OMIM Allelic Variant Phenotypes
157140.0019 EU, rs63750959: SUPRANUCLEAR PALSY, PROGRESSIVE, 1
157140.0017 ARG5HIS, rs63750959: FRONTOTEMPORAL DEMENTIA 1
157140.0015 LYS257THR, rs63750129: PICK DISEASE
157140.0022 LEU266VAL, rs63750349: FRONTOTEMPORAL DEMENTIA 1
157140.0002 GLY272VAL, rs63750376: FRONTOTEMPORAL DEMENTIA 1
157140.0009 ASN279LYS, rs63750756: FRONTOTEMPORAL DEMENTIA 1
157140.0021 ASN296DEL, rs63751392: SUPRANUCLEAR PALSY, PROGRESSIVE, 1, ATYPICAL
157140.0013 ASN296ASN, rs63750912: FRONTOTEMPORAL DEMENTIA 1
157140.0012 PRO301SER, rs63751438: FRONTOTEMPORAL DEMENTIA 1
157140.0001 PRO301LEU, rs63751273: FRONTOTEMPORAL DEMENTIA 1
157140.0025 GLY303VAL, rs63751391: SUPRANUCLEAR PALSY, PROGRESSIVE, 1
157140.0010 SER305ASN, rs63751165: FRONTOTEMPORAL DEMENTIA 1
157140.0007 IVS10, G-A, +1, rs1568327531: FRONTOTEMPORAL DEMENTIA 1
157140.0020 IVS10, T-C, +11, rs63751394: FRONTOTEMPORAL DEMENTIA 1
157140.0005 IVS10, A-G, +13, rs63750308: FRONTOTEMPORAL DEMENTIA 1
157140.0004 IVS10, C-U, +14, rs63750972: FRONTOTEMPORAL DEMENTIA 1
157140.0006 IVS10, C-T, +16, rs63751011: FRONTOTEMPORAL DEMENTIA 1
157140.0024 LYS317MET, rs63750092: FRONTOTEMPORAL DEMENTIA 1
157140.0018 SER320PHE, rs63750635: PICK DISEASE
157140.0008 VAL337MET, rs63750570: FRONTOTEMPORAL DEMENTIA 1
157140.0014 GLU342VAL, rs63750711: FRONTOTEMPORAL DEMENTIA 1
157140.0023 SER352LEU, rs63750425: SUPRANUCLEAR PALSY, PROGRESSIVE, 1
157140.0016 LYS369ILE, rs63751264: PICK DISEASE
157140.0011 GLY389ARG, rs63750512: PICK DISEASE
157140.0003 ARG406TRP, rs63750424: FRONTOTEMPORAL DEMENTIA 1
612452.0002 2-BP DEL, 2785AG, rs281865471: KOOLEN-DE VRIES SYNDROME
612452.0001 ARG606TER, rs281865469: KOOLEN-DE VRIES SYNDROME
612452.0004 IVS6DS, G-A, +1, rs281865470: KOOLEN-DE VRIES SYNDROME
OMIM Gene Phenotypes - Dark Green Can Be Disease-causing
OMIM Genes
Gene Expression in 54 tissues from GTEx RNA-seq of 17382 samples, 948 donors (V8, Aug 2019)
MAPT
RP11-669E14.4
Metazoa_SRP
STH
KANSL1
RP11-669E14.6
ENCODE Candidate Cis-Regulatory Elements (cCREs) combined from all cell types
ENCODE cCREs
H3K27Ac Mark (Often Found Near Regulatory Elements) on 7 cell lines from ENCODE
Layered H3K27Ac
Regulatory elements from ORegAnno
OREG1661231
OREG1728661
OREG1770261
OREG1417568
OREG1131043
OREG1194609
OREG1194610
OREG0862616
OREG1269067
OREG1194607
OREG0854149
OREG0085546
OREG1108635
OREG1907979
OREG0790223
OREG1661232
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OREG1300054
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OREG1306533
OREG0542501
OREG0603056
OREG0451267
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OREG1238169
OREG0834113
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OREG1946627
OREG1385095
OREG1824928
OREG1691210
OREG1300053
OREG0405379
OREG1306531
OREG0263233
OREG1516963
OREG0238079
OREG0362783
OREG0388499
OREG1899926
OREG1269068
OREG0405380
OREG0290779
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OREG0801377
OREG0707869
4
-0.5
100 vertebrates Basewise Conservation by PhyloP
Cons 100 Verts
Multiz Alignments of 100 Vertebrates
Rhesus
Mouse
Dog
Elephant
Chicken
X_tropicalis
Zebrafish
Short Genetic Variants from dbSNP release 155
Common dbSNP(155)
Repeating Elements by RepeatMasker
RepeatMasker