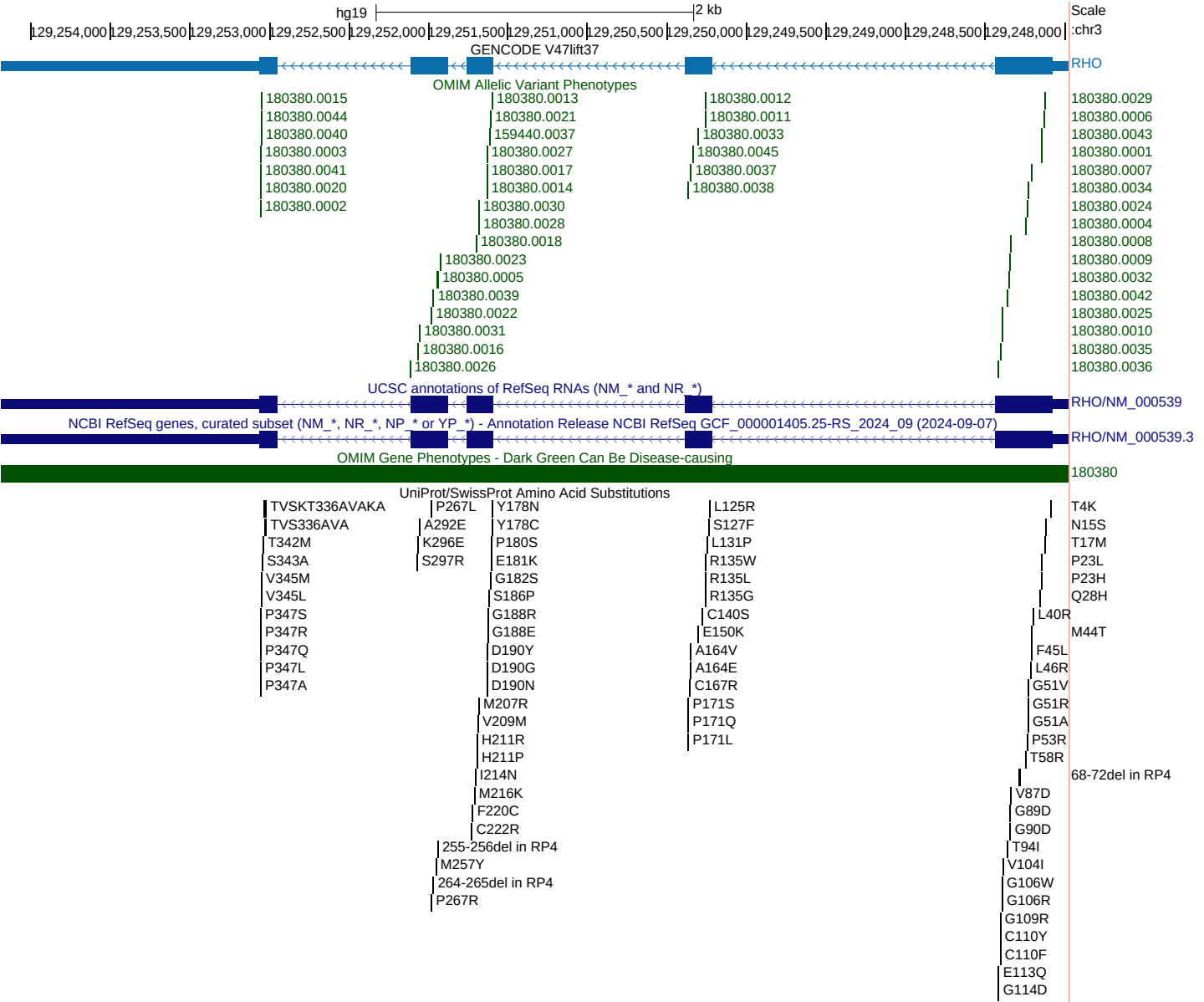
hg19
2 kb
Scale
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:chr3
GENCODE V47lift37
RHO
OMIM Allelic Variant Phenotypes
180380.0015
180380.0044
180380.0040
180380.0003
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180380.0023
180380.0005
180380.0039
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180380.0031
180380.0016
180380.0026
180380.0012
180380.0011
180380.0033
180380.0045
180380.0037
180380.0038
180380.0029
180380.0006
180380.0043
180380.0001
180380.0007
180380.0034
180380.0024
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180380.0008
180380.0009
180380.0032
180380.0042
180380.0025
180380.0010
180380.0035
180380.0036
UCSC annotations of RefSeq RNAs (NM_* and NR_*)
RHO/NM_000539
NCBI RefSeq genes, curated subset (NM_*, NR_*, NP_* or YP_*) - Annotation Release NCBI RefSeq GCF_000001405.25-RS_2024_09 (2024-09-07)
RHO/NM_000539.3
OMIM Gene Phenotypes - Dark Green Can Be Disease-causing
180380
UniProt/SwissProt Amino Acid Substitutions
TVSKT336AVAKA
TVS336AVA
T342M
S343A
V345M
V345L
P347S
P347R
P347Q
P347L
P347A
P267L
A292E
K296E
S297R
Y178N
Y178C
P180S
E181K
G182S
S186P
G188R
G188E
D190Y
D190G
D190N
M207R
V209M
H211R
H211P
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M216K
F220C
C222R
255-256del in RP4
M257Y
264-265del in RP4
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L125R
S127F
L131P
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R135L
R135G
C140S
E150K
A164V
A164E
C167R
P171S
P171Q
P171L
T4K
N15S
T17M
P23L
P23H
Q28H
L40R
M44T
F45L
L46R
G51V
G51R
G51A
P53R
T58R
68-72del in RP4
V87D
G89D
G90D
T94I
V104I
G106W
G106R
G109R
C110Y
C110F
E113Q
G114D