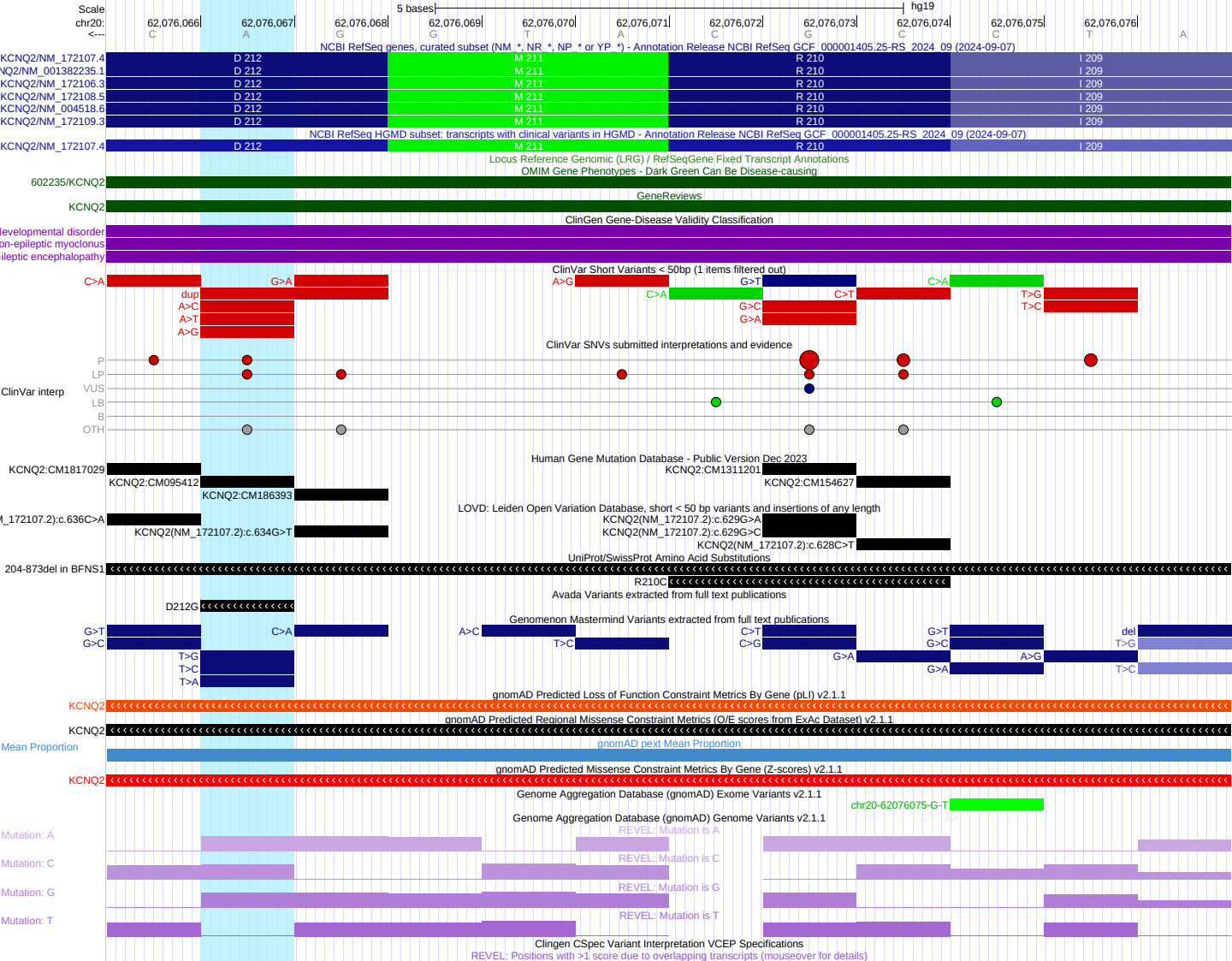
Scale
chr20:
<---
5 bases
hg19
62,076,066
62,076,067
62,076,068
62,076,069
62,076,070
62,076,071
62,076,072
62,076,073
62,076,074
62,076,075
62,076,076
C A G G T A C G C G T A
NCBI RefSeq genes, curated subset (NM_*, NR_*, NP_* or YP_*) - Annotation Release NCBI RefSeq GCF_000001405.25-RS_2024_09 (2024-09-07)
KCNQ2/NM_172107.4
NQ2/NM_001382235.1
KCNQ2/NM_172106.3
KCNQ2/NM_172108.5
KCNQ2/NM_004518.6
KCNQ2/NM_172109.3
D 212
M 211
R 210
I 209
NCBI RefSeq HGMD subset: transcripts with clinical variants in HGMD - Annotation Release NCBI RefSeq GCF_000001405.25-RS_2024_09 (2024-09-07)
Locus Reference Genomic (LRG) / RefSeqGene Fixed Transcript Annotations
OMIM Gene Phenotypes - Dark Green Can Be Disease-causing
602235/KCNQ2
GeneReviews
KCNQ2
ClinGen Gene-Disease Validity Classification
levelopmental disorder
on-epileptic myoclonus
ileptic encephalopathy
ClinVar Short Variants < 50bp (1 items filtered out)
C>A
G>A
dup
A>C
A>T
A>G
A>G
G>T
C>A
C>T
T>G
T>C
G>C
G>A
ClinVar SNVs submitted interpretations and evidence
ClinVar interp
P
LP
VUS
LB
B
OTH
Human Gene Mutation Database - Public Version Dec 2023
KCNQ2:CM1817029
KCNQ2:CM095412
KCNQ2:CM186393
KCNQ2:CM1311201
KCNQ2:CM154627
LOVD: Leiden Open Variation Database, short < 50 bp variants and insertions of any length
_172107.2):c.636C>A
KCNQ2(NM_172107.2):c.634G>T
KCNQ2(NM_172107.2):c.629G>A
KCNQ2(NM_172107.2):c.629G>C
KCNQ2(NM_172107.2):c.628C>T
UniProt/SwissProt Amino Acid Substitutions
204-873del in BFNS1
R210C
Avada Variants extracted from full text publications
D212G
Genomenon Mastermind Variants extracted from full text publications
G>T
G>C
C>A
T>G
T>C
T>A
A>C
T>C
C>T
C>G
G>A
G>T
G>C
G>A
A>G
del
T>G
T>C
gnomAD Predicted Loss of Function Constraint Metrics By Gene (pLI) v2.1.1
KCNQ2
gnomAD Predicted Regional Missense Constraint Metrics (O/E scores from ExAc Dataset) v2.1.1
KCNQ2
Mean Proportion
gnomAD pext Mean Proportion
gnomAD Predicted Missense Constraint Metrics By Gene (Z-scores) v2.1.1
KCNQ2
Genome Aggregation Database (gnomAD) Exome Variants v2.1.1
chr20-62076075-G-T
Genome Aggregation Database (gnomAD) Genome Variants v2.1.1
Mutation: A
REVEL: Mutation is A
Mutation: C
REVEL: Mutation is C
Mutation: G
REVEL: Mutation is G
Mutation: T
REVEL: Mutation is T
Clingen CSpec Variant Interpretation VCEP Specifications
REVEL: Positions with >1 score due to overlapping transcripts (mouseover for details)