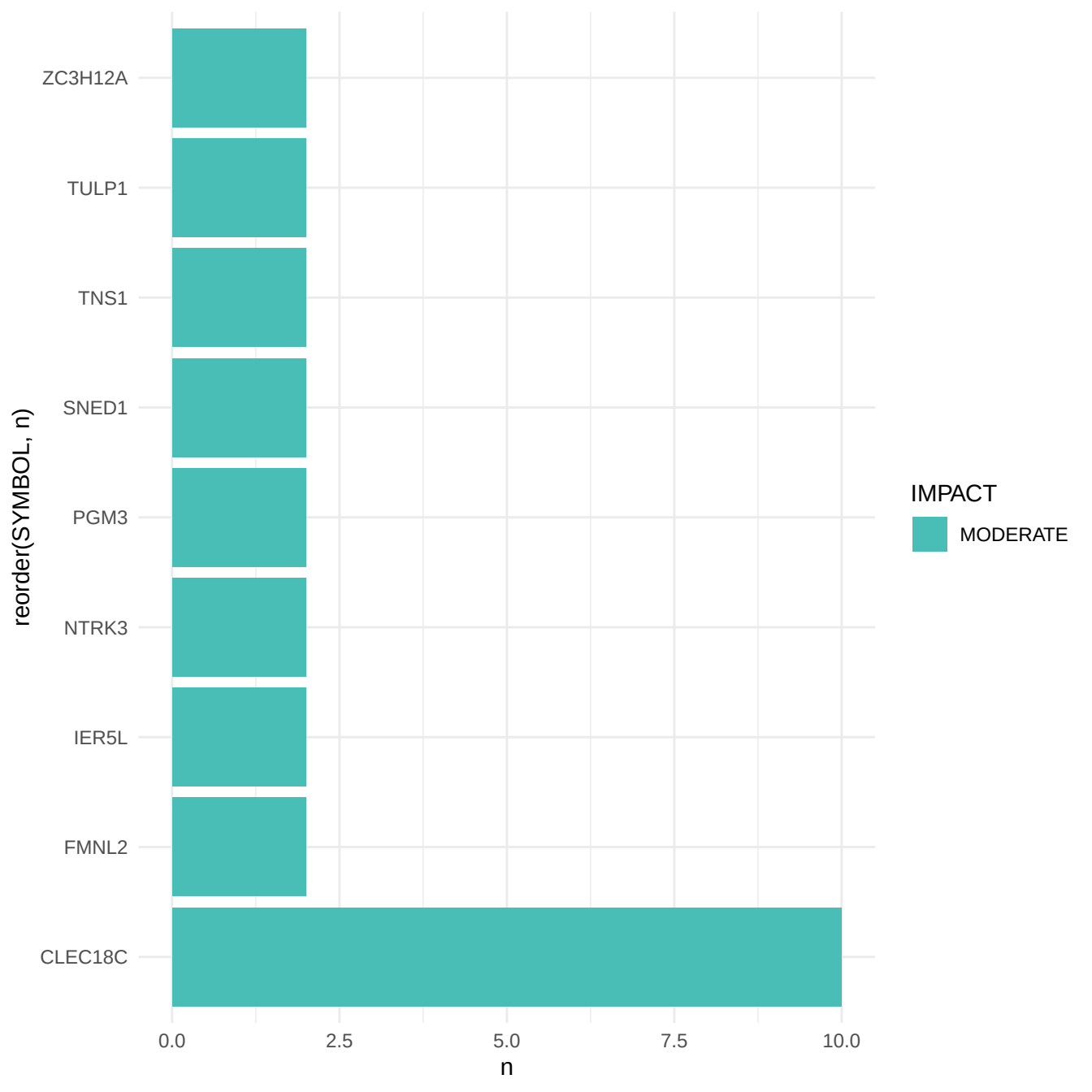
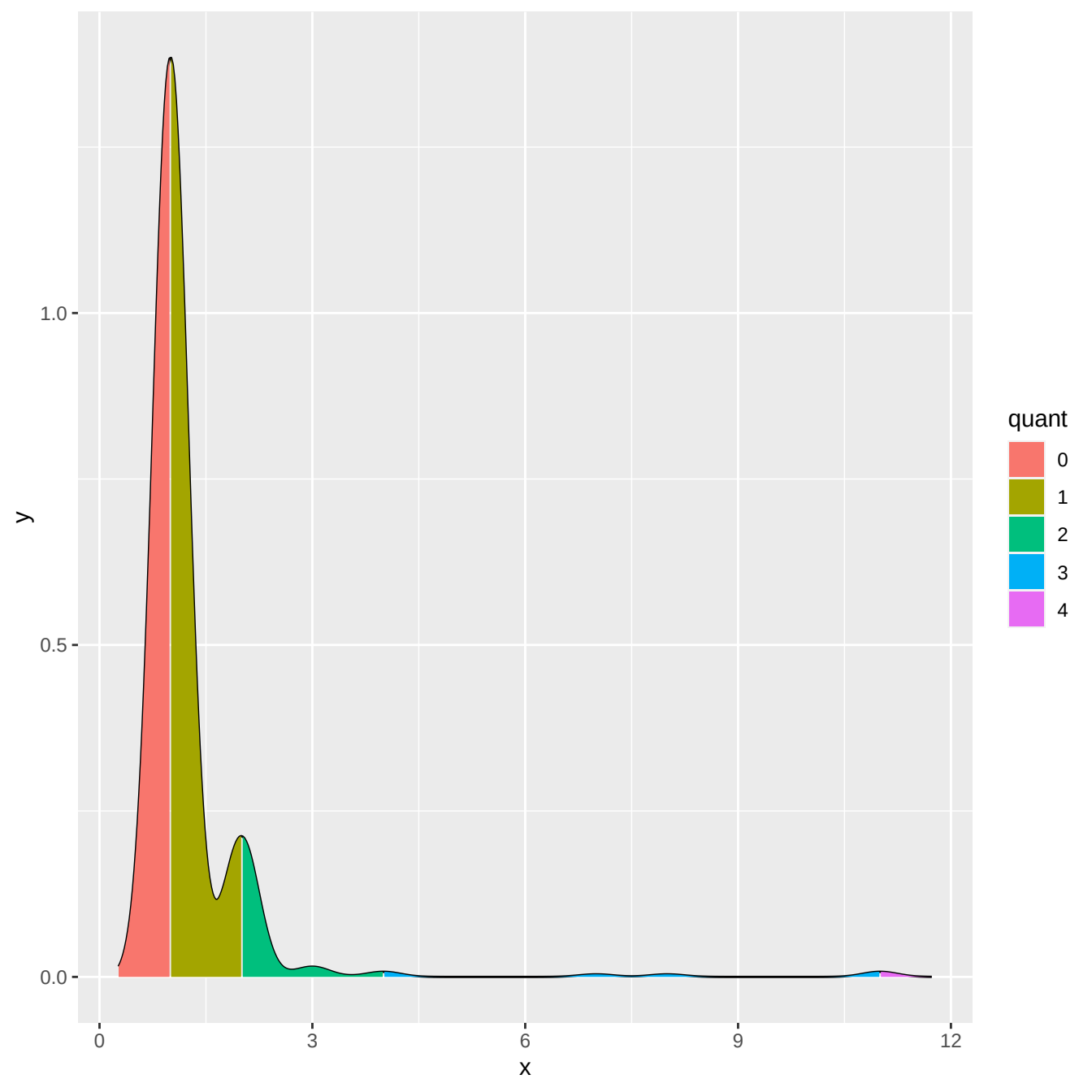
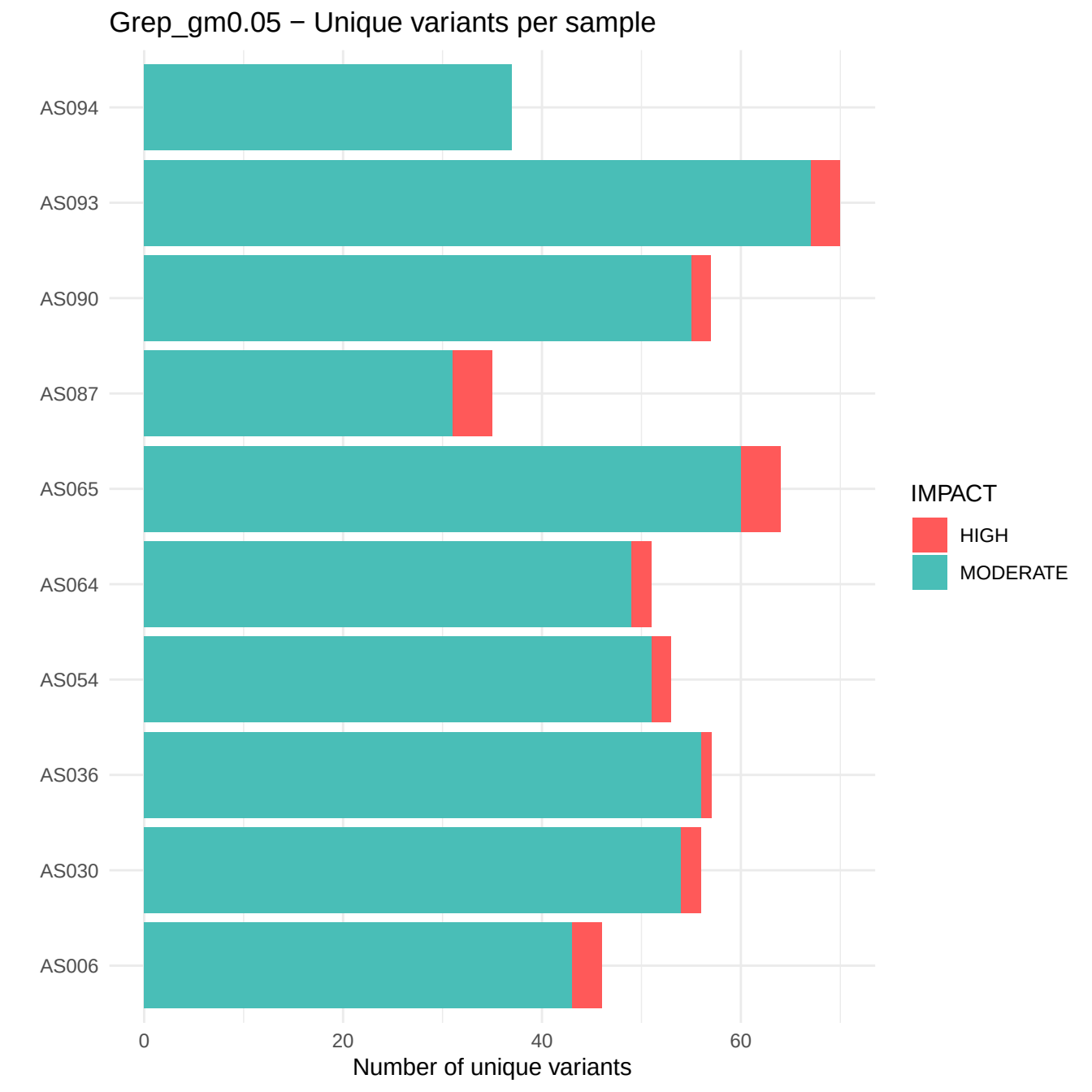
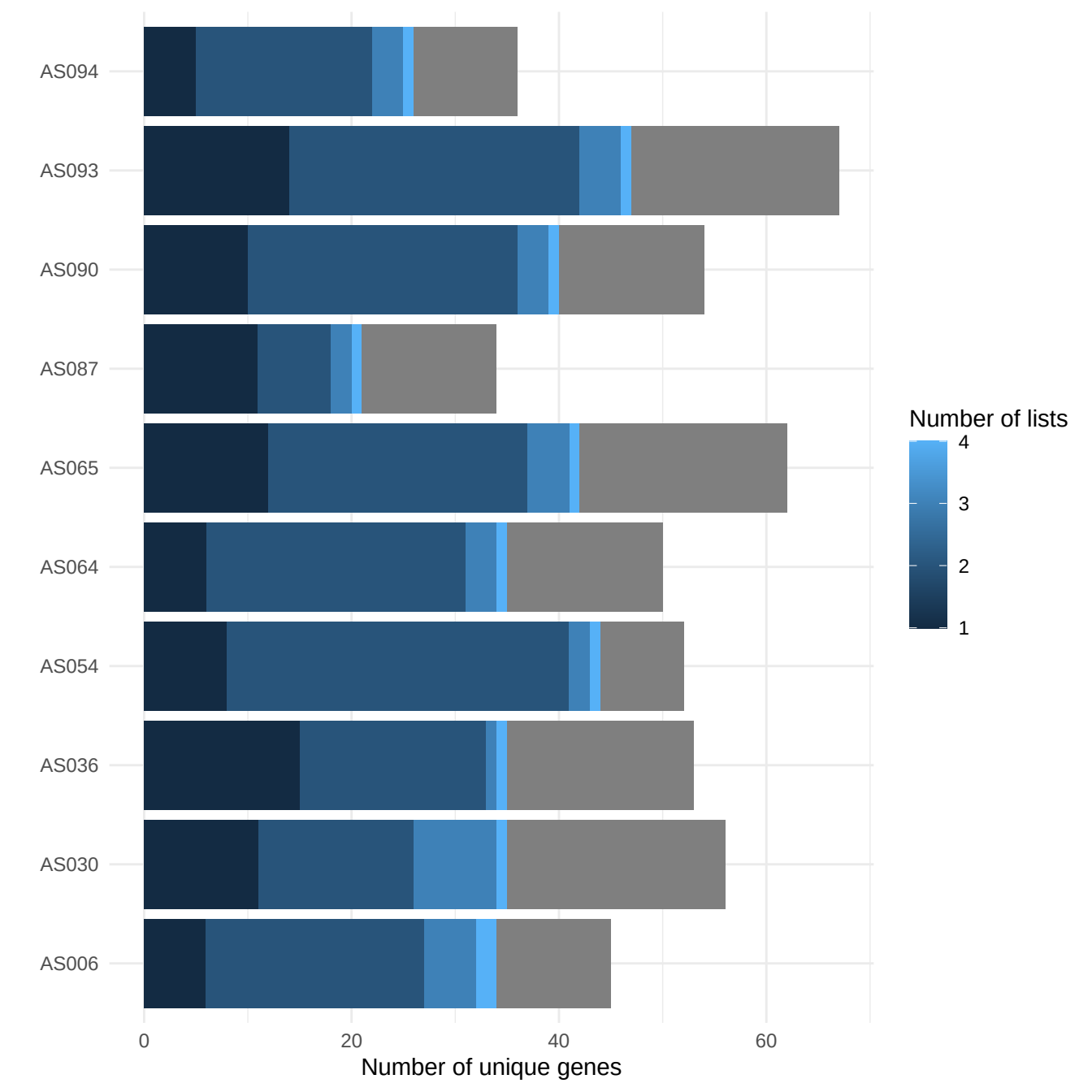
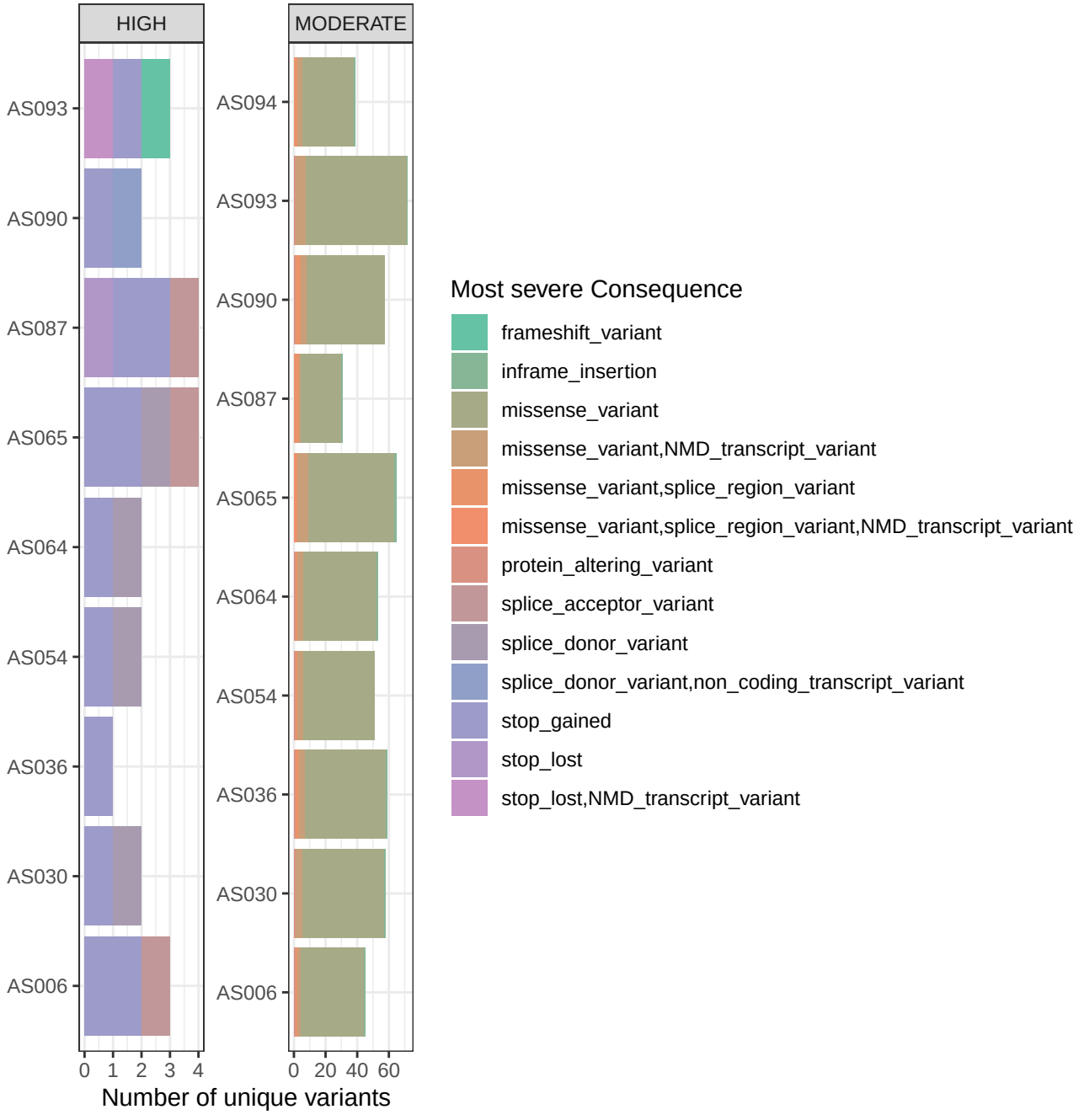
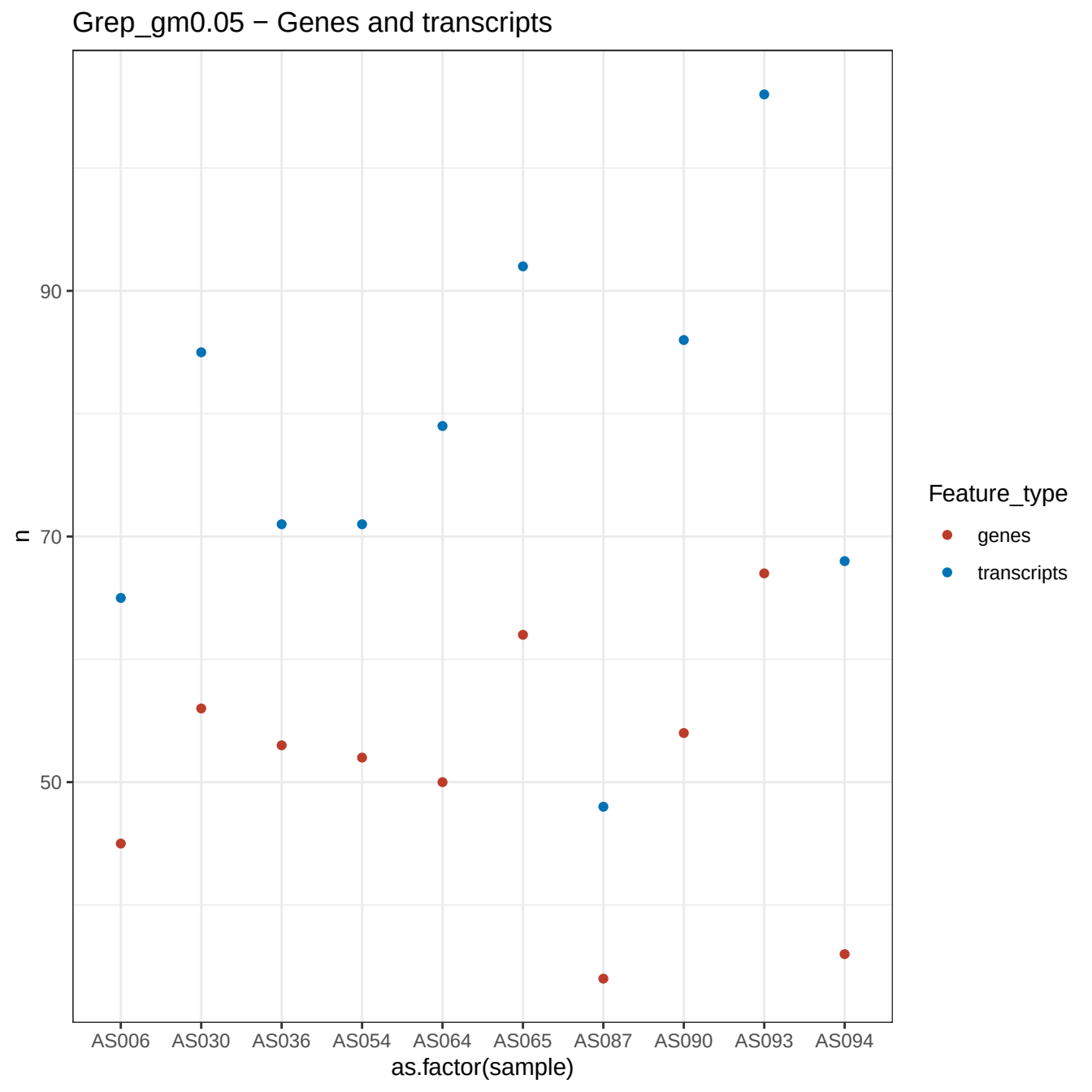

ZC3H12A
TULP1
TNS1
SNED1
PGM3
NTRK3
IER5L
FMNL2
CLEC18C
reorder(SYMBOL, n)
0.0
2.5
5.0
7.5
10.0
n
IMPACT
MODERATE

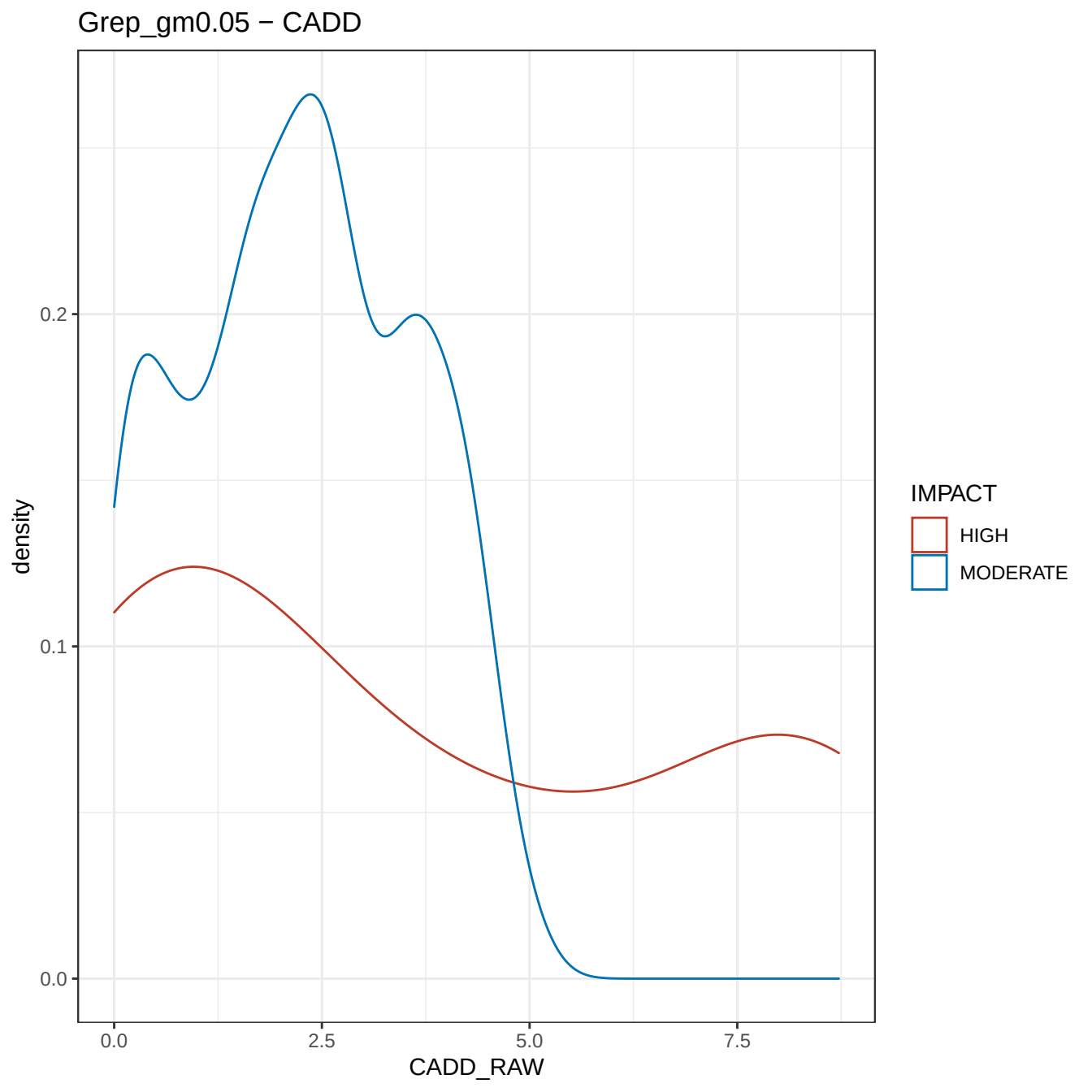

1.0
0.5
0.0
y
0
3
6
9
12
x
quant
0
1
2
3
4

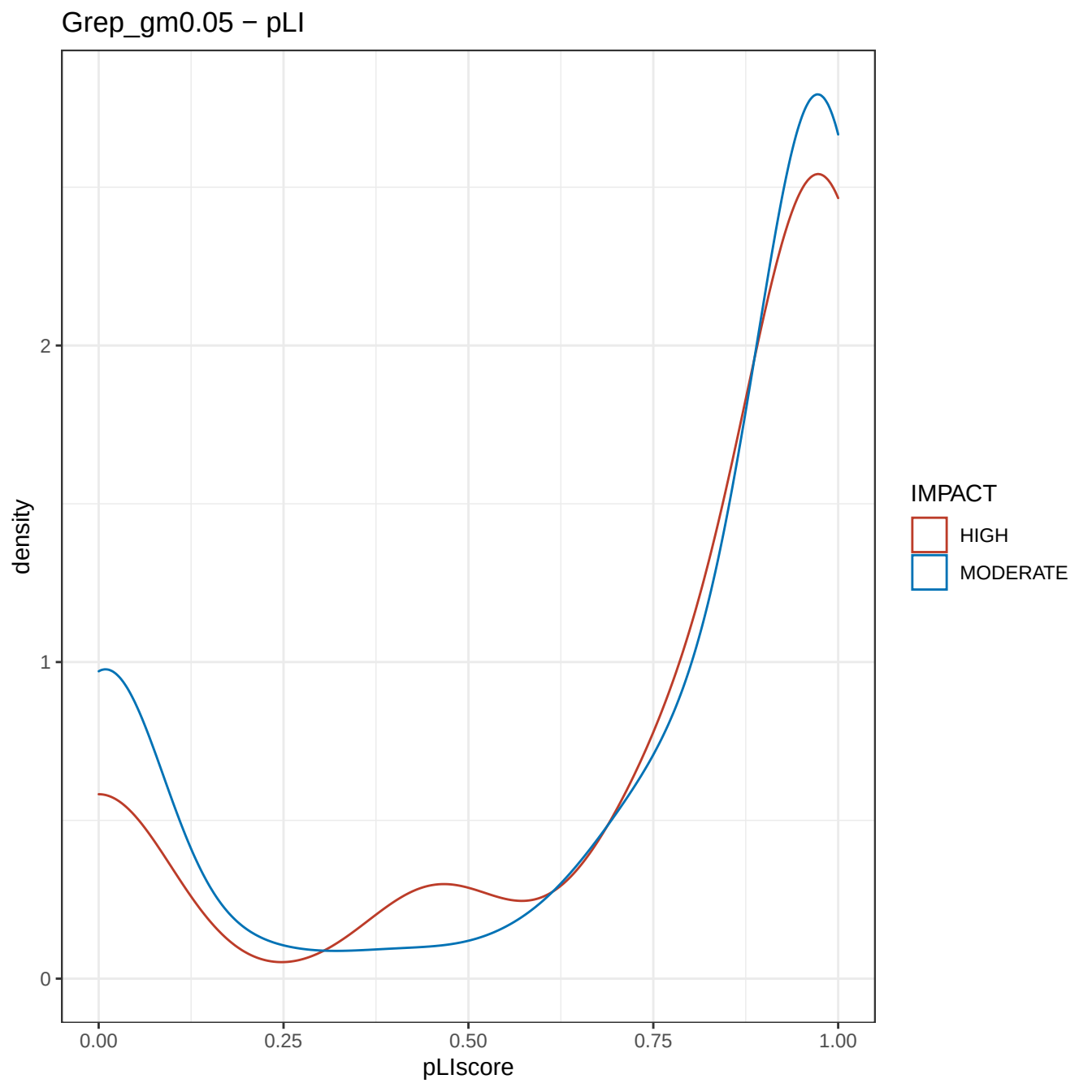

Grep_gm0.05 − Unique variants per sample
AS094
AS093
AS090
AS087
AS065
AS064
AS054
AS036
AS030
AS006
0
20
40
60
Number of unique variants
IMPACT
HIGH
MODERATE

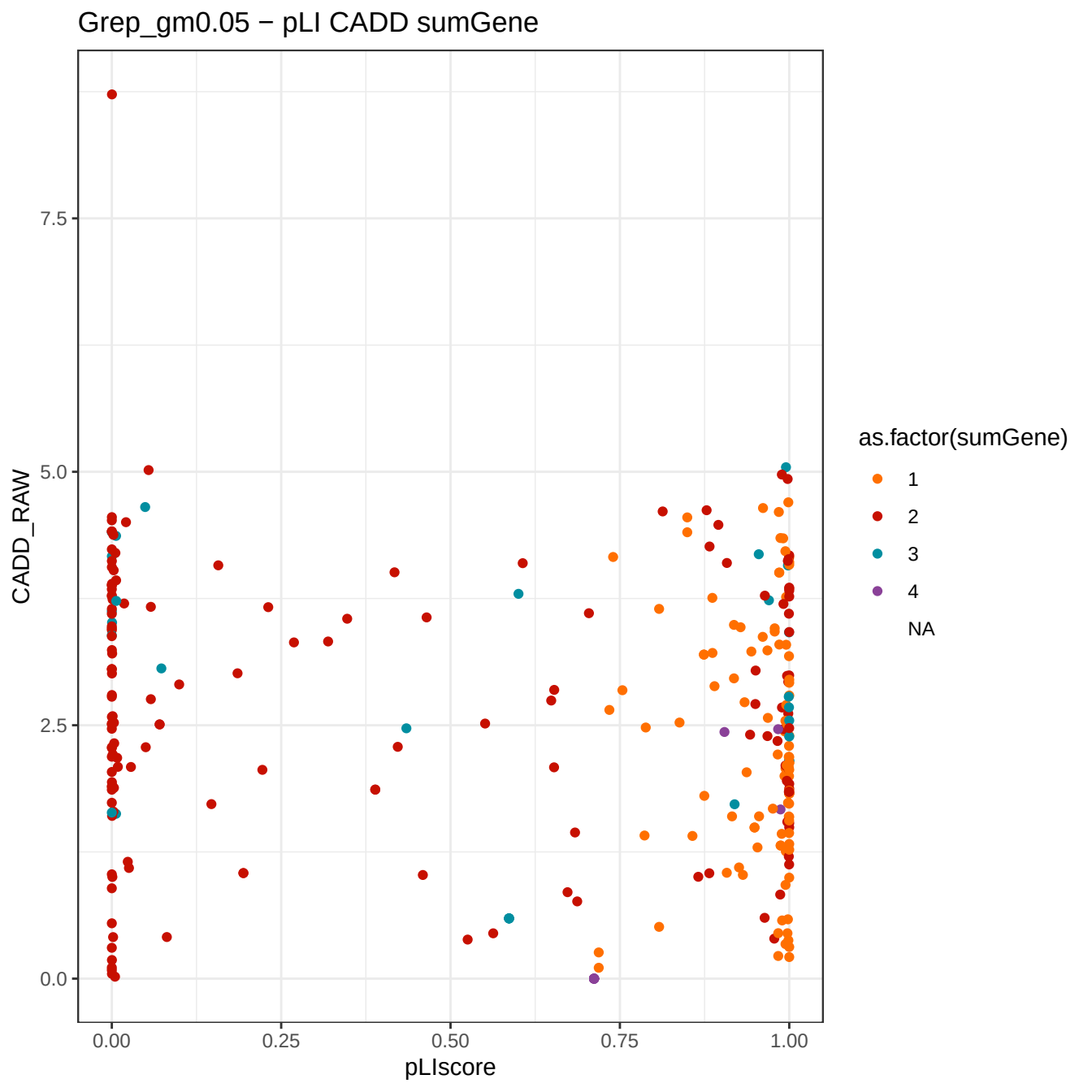

AS094
AS093
AS090
AS087
AS065
AS064
AS054
AS036
AS030
AS006
0
20
40
60
Number of unique genes
Number of lists
4
3
2
1

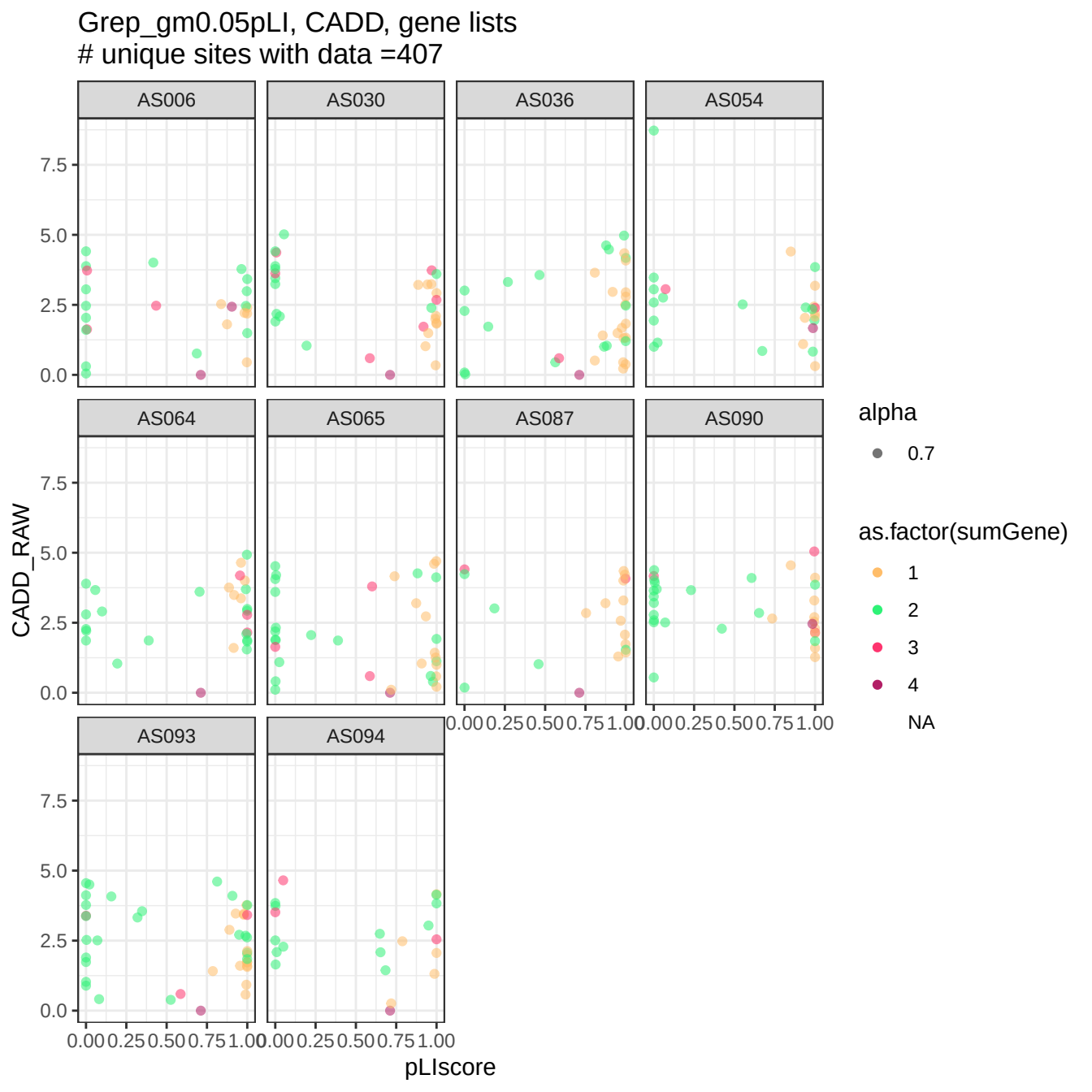

HIGH
MODERATE
AS093
AS090
AS087
AS065
AS064
AS054
AS036
AS030
AS006
AS094
AS093
AS090
AS087
AS065
AS064
AS054
AS036
AS030
AS006
0 1 2 3 4
0 20 40 60
Number of unique variants
Most severe Consequence
frameshift_variant
inframe_insertion
missense_variant
missense_variant,NMD_transcript_variant
missense_variant,splice_region_variant
missense_variant,splice_region_variant,NMD_transcript_variant
protein_altering_variant
splice_acceptor_variant
splice_donor_variant
splice_donor_variant,non_coding_transcript_variant
stop_gained
stop_lost
stop_lost,NMD_transcript_variant

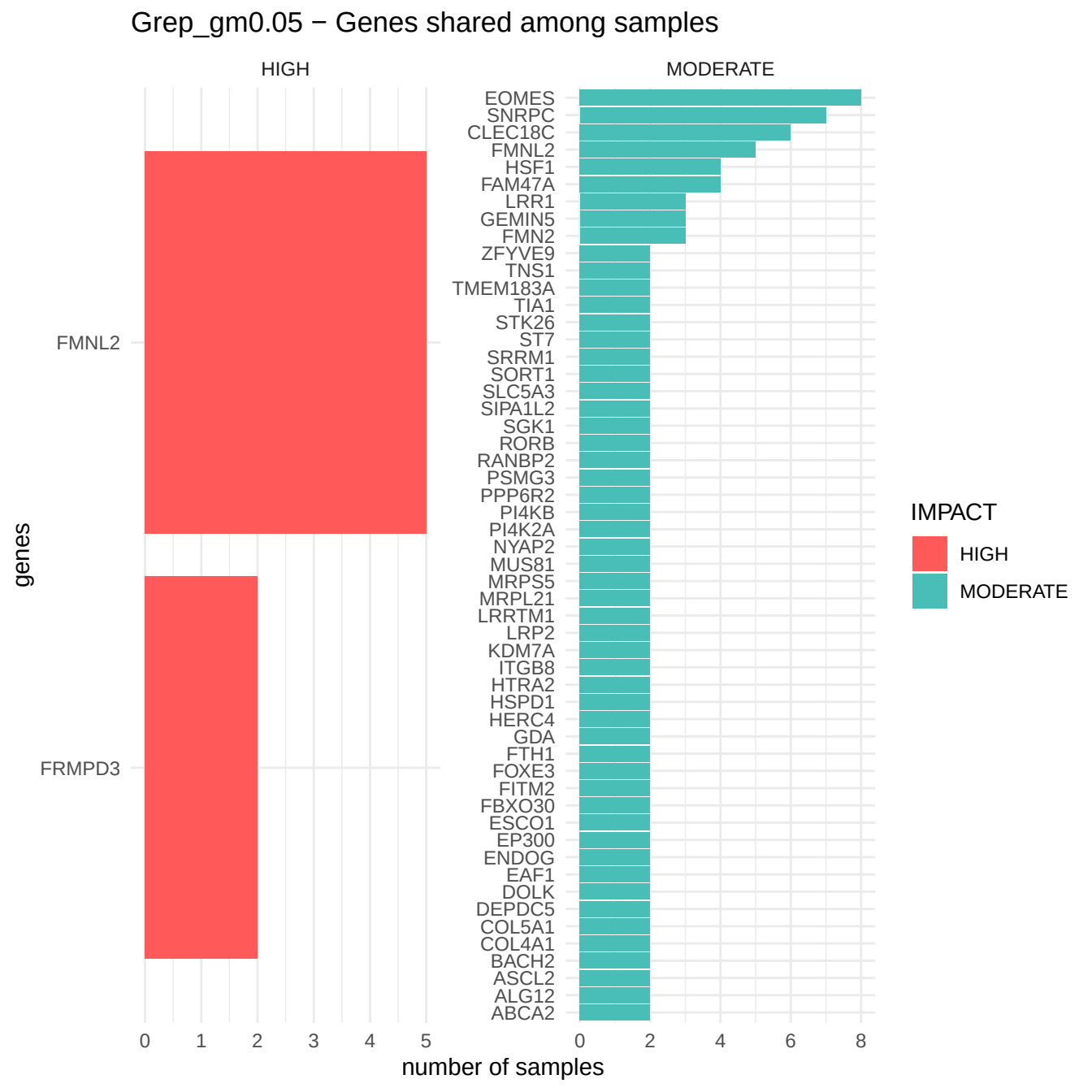

Grep_gm0.05 − Genes and transcripts
n
90
70
50
AS006
AS030
AS036
AS054
AS064
AS065
AS087
AS090
AS093
AS094
as.factor(sample)
Feature_type
genes
transcripts

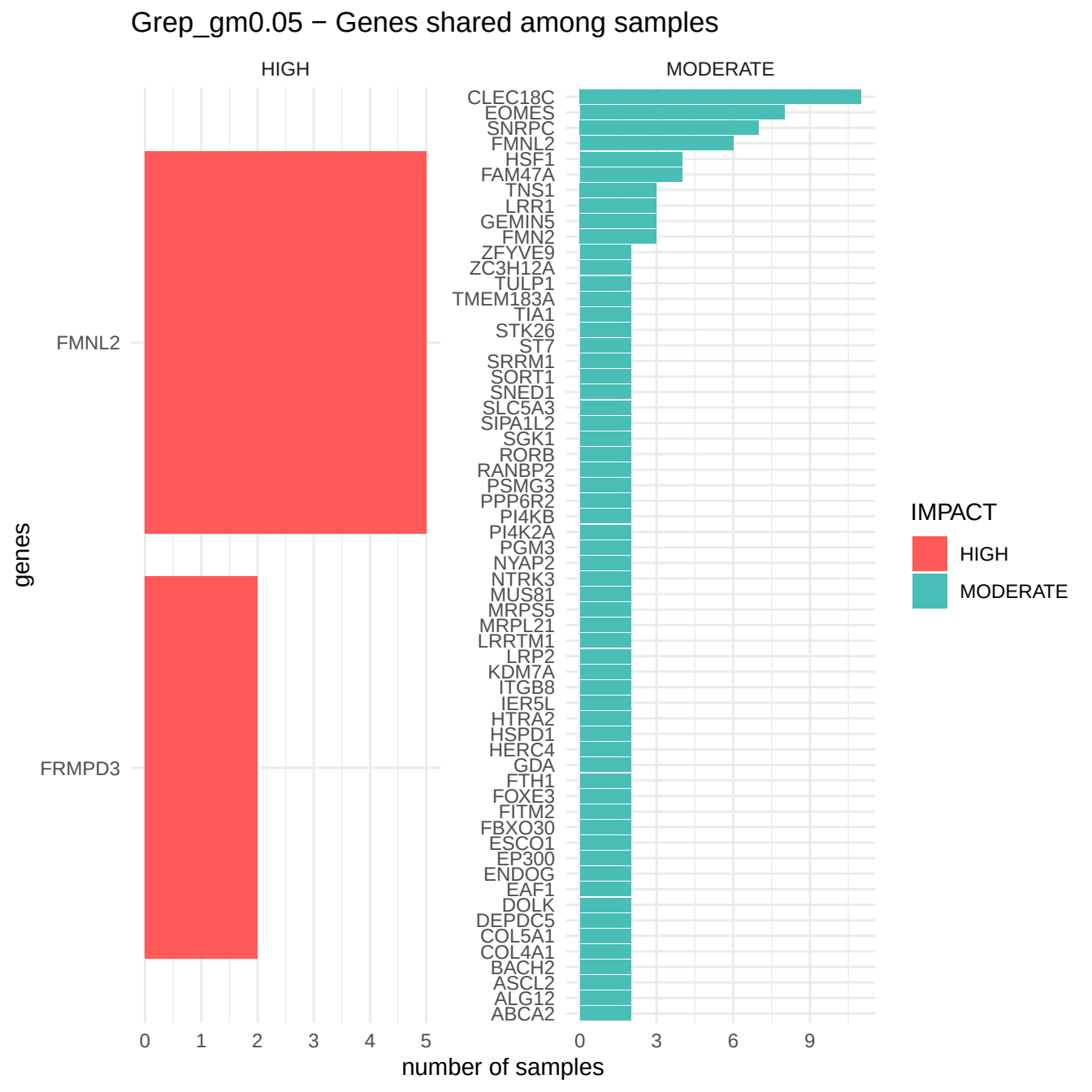

Grep_gm0.05 − CADD
density
0.2
0.1
0.0
0.0
2.5
5.0
7.5
CADD_RAW
IMPACT
HIGH
MODERATE

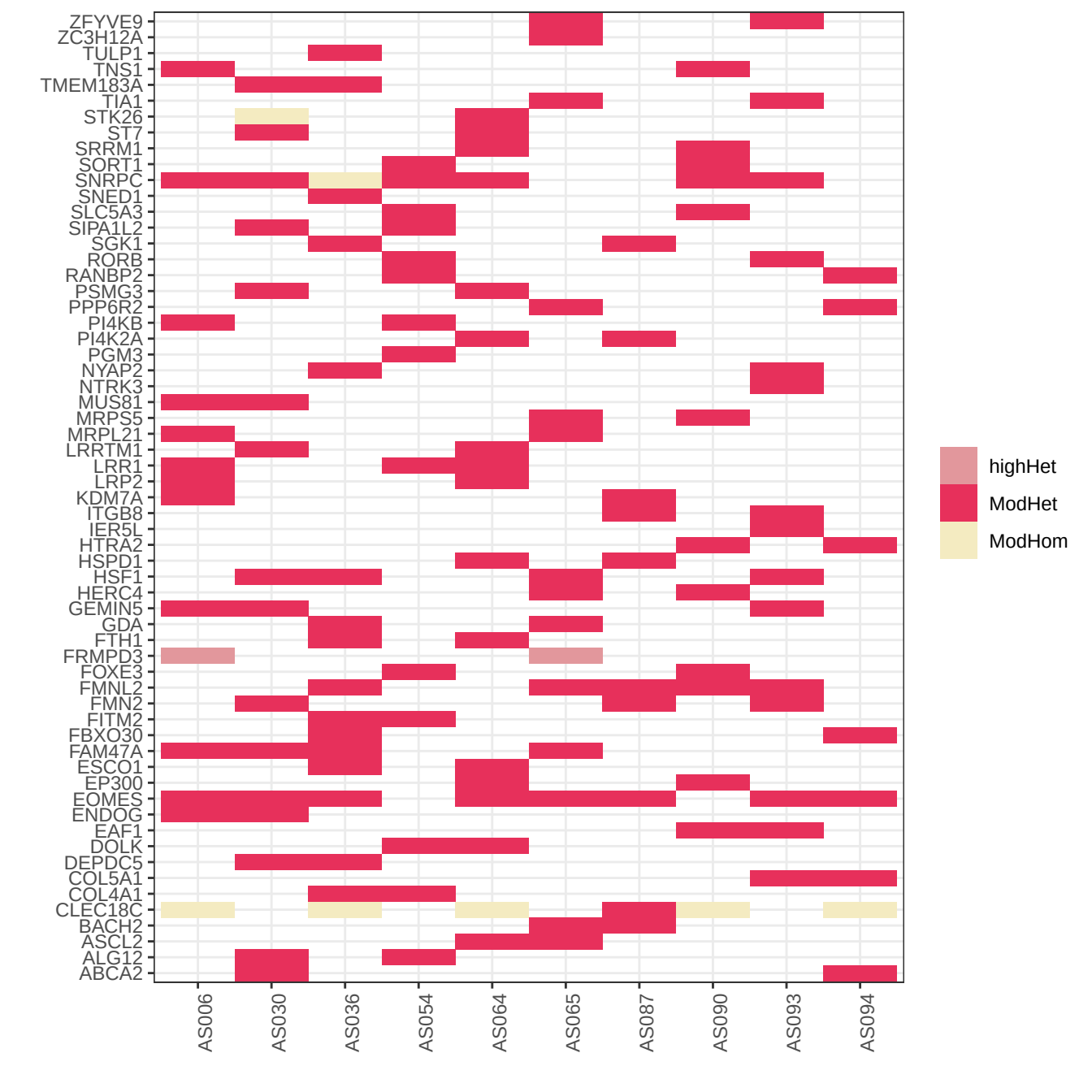

Grep_gm0.05 − pLI
density
2
1
0
0.00
0.25
0.50
0.75
1.00
pLIscore
IMPACT
HIGH
MODERATE

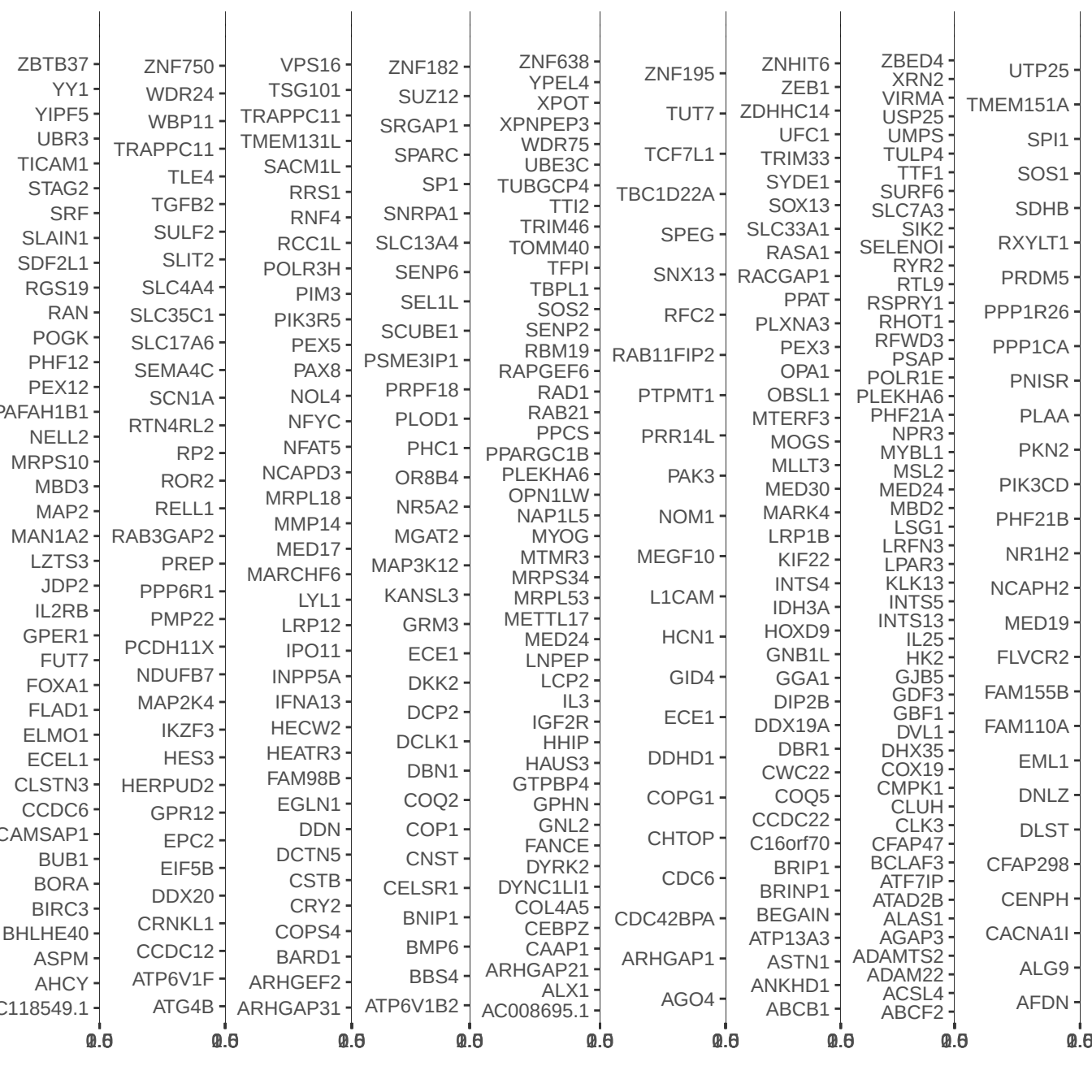

Grep_gm0.05 − pLI CADD sumGene
CADD_RAW
7.5
5.0
2.5
0.0
0.00
0.25
0.50
0.75
1.00
pLIscore
as.factor(sumGene)
1
2
3
4
NA


Grep_gm0.05pLI, CADD, gene lists
unique sites with data =407
AS006
AS030
AS036
AS054
AS064
AS065
AS087
AS090
AS093
AS094
CADD_RAW
pLIscore
0.0
2.5
5.0
7.5
0.00
0.25
0.50
0.75
1.00
alpha
0.7
as.factor(sumGene)
1
2
3
4
NA


Grep_gm0.05 − Genes shared among samples
HIGH
MODERATE
genes
FMNL2
FRMPD3
EOMES
SNRPC
CLEC18C
FMNL2
HSF1
FAM47A
LRR1
GEMIN5
FMN2
ZFYVE9
TNS1
TMEM183A
TIA1
STK26
ST7
SRRM1
SORT1
SLC5A3
SIPA1L2
SGK1
RORB
RANBP2
PSMG3
PPP6R2
PI4KB
PI4K2A
NYAP2
MUS81
MRPS5
MRPL21
LRRTM1
LRP2
KDM7A
ITGB8
HTRA2
HSPD1
HERC4
GDA
FTH1
FOXE3
FITM2
FBXO30
ESCO1
EP300
ENDOG
EAF1
DOLK
DEPDC5
COL5A1
COL4A1
BACH2
ASCL2
ALG12
ABCA2
0
1
2
3
4
5
0
2
4
6
8
number of samples
IMPACT
HIGH
MODERATE


Grep_gm0.05 − Genes shared among samples
HIGH
MODERATE
genes
FMNL2
FRMPD3
CLEC18C
EOMES
SNRPC
FMNL2
HSF1
FAM47A
TNS1
LRR1
GEMIN5
FMN2
ZFYVE9
ZC3H12A
TULP1
TMEM183A
TIA1
STK26
ST7
SRRM1
SORT1
SNED1
SLC5A3
SIPA1L2
SGK1
RORB
RANBP2
PSMG3
PPP6R2
PI4KB
PI4K2A
PGM3
NYAP2
NTRK3
MUS81
MRPS5
MRPL21
LRRTM1
LRP2
KDM7A
ITGB8
IER5L
HTRA2
HSPD1
HERC4
GDA
FTH1
FOXE3
FITM2
FBXO30
ESCO1
EP300
ENDOG
EAF1
DOLK
DEPDC5
COL5A1
COL4A1
BACH2
ASCL2
ALG12
ABCA2
0
1
2
3
4
5
0
3
6
9
number of samples
IMPACT
HIGH
MODERATE


ZFYVE9
ZC3H12A
TULP1
TNS1
TMEM183A
TIA1
STK26
ST7
SRRM1
SORT1
SNRPC
SNED1
SLC5A3
SIPA1L2
SGK1
RORB
RANBP2
PSMG3
PPP6R2
PI4KB
PI4K2A
PGM3
NYAP2
NTRK3
MUS81
MRPS5
MRPL21
LRRTM1
LRR1
LRP2
KDM7A
ITGB8
IER5L
HTRA2
HSPD1
HSF1
HERC4
GEMIN5
GDA
FTH1
FRMPD3
FOXE3
FMNL2
FMN2
FITM2
FBXO30
FAM47A
ESCO1
EP300
EOMES
ENDOG
EAF1
DOLK
DEPDC5
COL5A1
COL4A1
CLEC18C
BACH2
ASCL2
ALG12
ABCA2
AS006
AS030
AS036
AS054
AS064
AS065
AS087
AS090
AS093
AS094
highHet
ModHet
ModHom


ZBTB37
YY1
YIPF5
UBR3
TICAM1
STAG2
SRF
SLAIN1
SDF2L1
RGS19
RAN
POGK
PHF12
PEX12
PAFAH1B1
NELL2
MRPS10
MBD3
MAP2
MAN1A2
LZTS3
JDP2
IL2RB
GPER1
FUT7
FOXA1
FLAD1
ELMO1
ECEL1
CLSTN3
CCDC6
CAMSAP1
BUB1
BORA
BIRC3
BHLHE40
ASPM
AHCY
C118549.1
ZNF750
WDR24
WBP11
TRAPPC11
TLE4
TGFB2
SULF2
SLIT2
SLC4A4
SLC35C1
SLC17A6
SEMA4C
SCN1A
RTN4RL2
RP2
ROR2
RELL1
RAB3GAP2
PREP
PPP6R1
PMP22
PCDH11X
NDUFB7
MAP2K4
IKZF3
HES3
HERPUD2
GPR12
EPC2
EIF5B
DDX20
CRNKL1
CCDC12
ATP6V1F
ATG4B
VPS16
TSG101
TRAPPC11
TMEM131L
SACM1L
RRS1
RNF4
RCC1L
POLR3H
PIM3
PIK3R5
PEX5
PAX8
NOL4
NFYC
NFAT5
NCAPD3
MRPL18
MMP14
MED17
MARCHF6
LYL1
LRP12
IPO11
INPP5A
IFNA13
HECW2
HEATR3
FAM98B
EGLN1
DDN
DCTN5
CSTB
CRY2
COPS4
BARD1
ARHGEF2
ARHGAP31
ZNF182
SUZ12
SRGAP1
SPARC
SP1
SNRPA1
SLC13A4
SENP6
SEL1L
SCUBE1
PSME3IP1
PRPF18
PLOD1
PHC1
OR8B4
NR5A2
MGAT2
MAP3K12
KANSL3
GRM3
ECE1
DKK2
DCP2
DCLK1
DBN1
COQ2
COP1
CNST
CELSR1
BNIP1
BMP6
BBS4
ATP6V1B2
ZNF638
YPEL4
XPOT
XPNPEP3
WDR75
UBE3C
TUBGCP4
TTI2
TRIM46
TOMM40
TFPI
TBPL1
SOS2
SENP2
RBM19
RAPGEF6
RAD1
RAB21
PPCS
PPARGC1B
PLEKHA6
OPN1LW
NAP1L5
MYOG
MTMR3
MRPS34
MRPL53
METTL17
MED24
LNPEP
LCP2
IL3
IGF2R
HHIP
HAUS3
GTPBP4
GPHN
GNL2
FANCE
DYRK2
DYNC1LI1
COL4A5
CEBPZ
CAAP1
ARHGAP21
ALX1
AC008695.1
ZNF195
TUT7
TCF7L1
TBC1D22A
SPEG
SNX13
RFC2
RAB11FIP2
PTPMT1
PRR14L
PAK3
NOM1
MEGF10
L1CAM
HCN1
GID4
ECE1
DDHD1
COPG1
CHTOP
CDC6
CDC42BPA
ARHGAP1
AGO4
ZNHIT6
ZEB1
ZDHHC14
UFC1
TRIM33
SYDE1
SOX13
SLC33A1
RASA1
RACGAP1
PPAT
PLXNA3
PEX3
OPA1
OBSL1
MTERF3
MOGS
MLLT3
MED30
MARK4
LRP1B
KIF22
INTS4
IDH3A
HOXD9
GNB1L
GGA1
DIP2B
DDX19A
DBR1
CWC22
COQ5
CCDC22
C16orf70
BRIP1
BRINP1
BEGAIN
ATP13A3
ASTN1
ANKHD1
ABCB1
ZBED4
XRN2
VIRMA
USP25
UMPS
TULP4
TTF1
SURF6
SLC7A3
SIK2
SELENOI
RYR2
RTL9
RSPRY1
RHOT1
RFWD3
PSAP
POLR1E
PLEKHA6
PHF21A
NPR3
MYBL1
MSL2
MED24
MBD2
LSG1
LRFN3
LPAR3
KLK13
INTS5
INTS13
IL25
HK2
GJB5
GDF3
GBF1
DVL1
DHX35
COX19
CMPK1
CLUH
CLK3
CFAP47
BCLAF3
ATF7IP
ATAD2B
ALAS1
AGAP3
ADAMTS2
ADAM22
ACSL4
ABCF2
UTP25
TMEM151A
SPI1
SOS1
SDHB
RXYLT1
PRDM5
PPP1R26
PPP1CA
PNISR
PLAA
PKN2
PIK3CD
PHF21B
NR1H2
NCAPH2
MED19
FLVCR2
FAM155B
FAM110A
EML1
DNLZ
DLST
CFAP298
CENPH
CACNA1I
ALG9
AFDN