

--- Correctly Predicted Variant ---

Top head → layer 4, head 15, score 1.0000

VariationID: 3175

VariationName: NM\_000187.4(HGD):c.808G>A (p.Gly270Arg)

VariantType: single nucleotide variant

GeneSymbol: HGD

OMIM: 607474.0

AssemblyChr: 3

Start: 120641660

Stop: 120641660

RefAlleleVCF: C

AltAlleleVCF: T

VariantLength: 1

ProteinChange: G270R

HGVS\_coding: NM\_000187.4:c.808G>A

HGVS\_protein: NP\_000178.2:p.Gly270Arg

DateLastUpdated: 2025-05-17

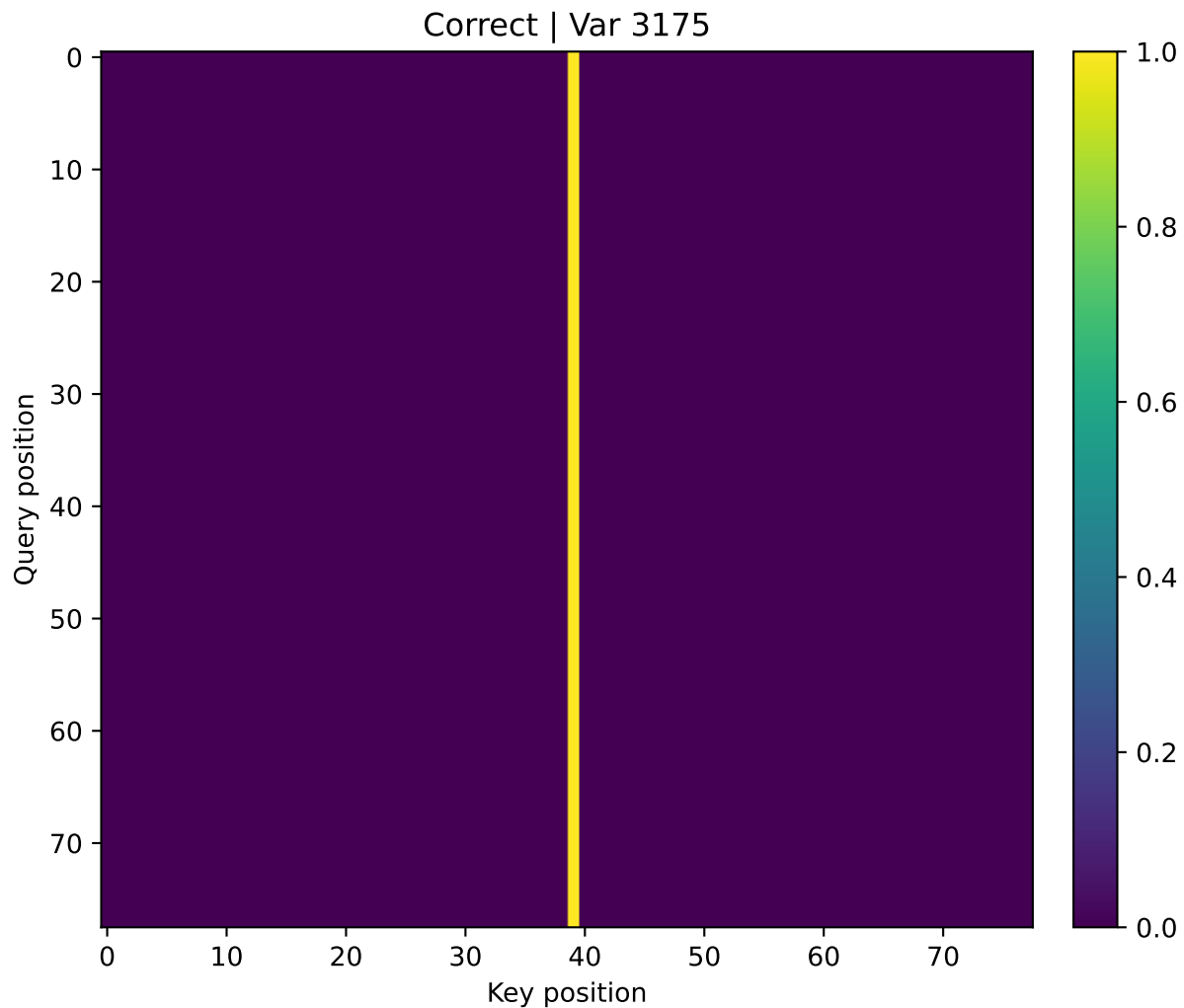
NumberOfSubmitters: 8

RecordType: classified

CLNSIG: Pathogenic/Likely\_pathogenic

actual\_label: 1

prediction: 1



--- Correctly Predicted Variant ---

Top head → layer 14, head 18, score 0.8355

VariationID: 1326860

VariationName: NM\_001204077.2(UBE4A):c.631C>T (p.Arg211Ter)

VariantType: single nucleotide variant

GeneSymbol: UBE4A

OMIM: 603753.0

AssemblyChr: 11

Start: 118372576

Stop: 118372576

RefAlleleVCF: C

AltAlleleVCF: T

VariantLength: 1

ProteinChange: R211\*

HGVS\_coding: NM\_001204077.2:c.631C>T

HGVS\_protein: NP\_001191006.1:p.Arg211Ter

DateLastUpdated: 2023-12-25

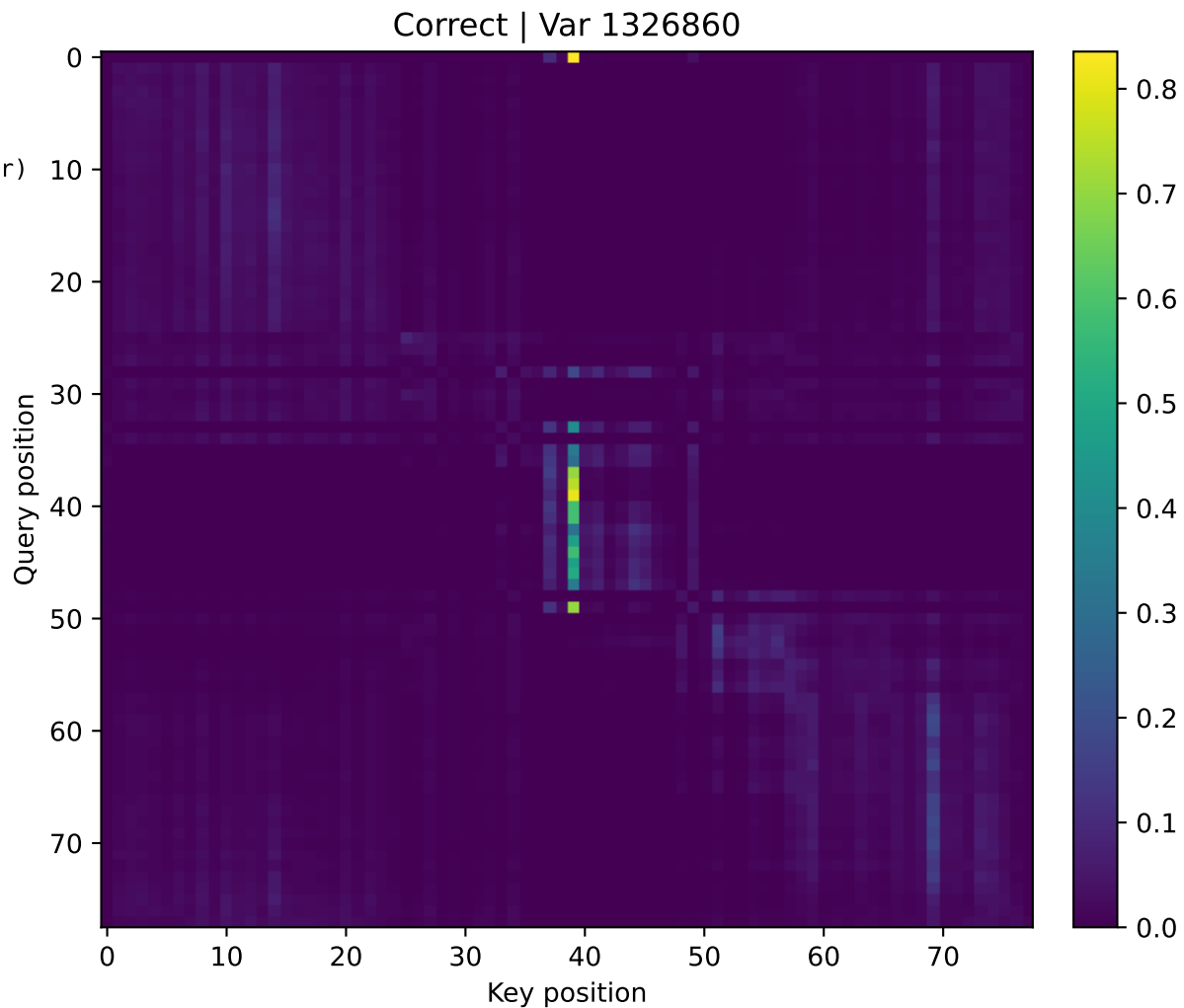
NumberOfSubmitters: 2

RecordType: classified

CLNSIG: Pathogenic

actual\_label: 1

prediction: 1



--- Correctly Predicted Variant ---

Top head → layer 11, head 16, score 0.5990

VariationID: 1146510

VariationName: NM\_006846.4(SPINK5):c.2526C>T (p.Ser842=)

VariantType: single nucleotide variant

GeneSymbol: SPINK5

OMIM: 605010.0

AssemblyChr: 5

Start: 148120379

Stop: 148120379

RefAlleleVCF: C

AltAlleleVCF: T

VariantLength: 1

ProteinChange: nan

HGVS\_coding: NM\_001127698.2:c.2526C>T

HGVS\_protein: NP\_001121170.1:p.Ser842=

DateLastUpdated: 2025-05-17

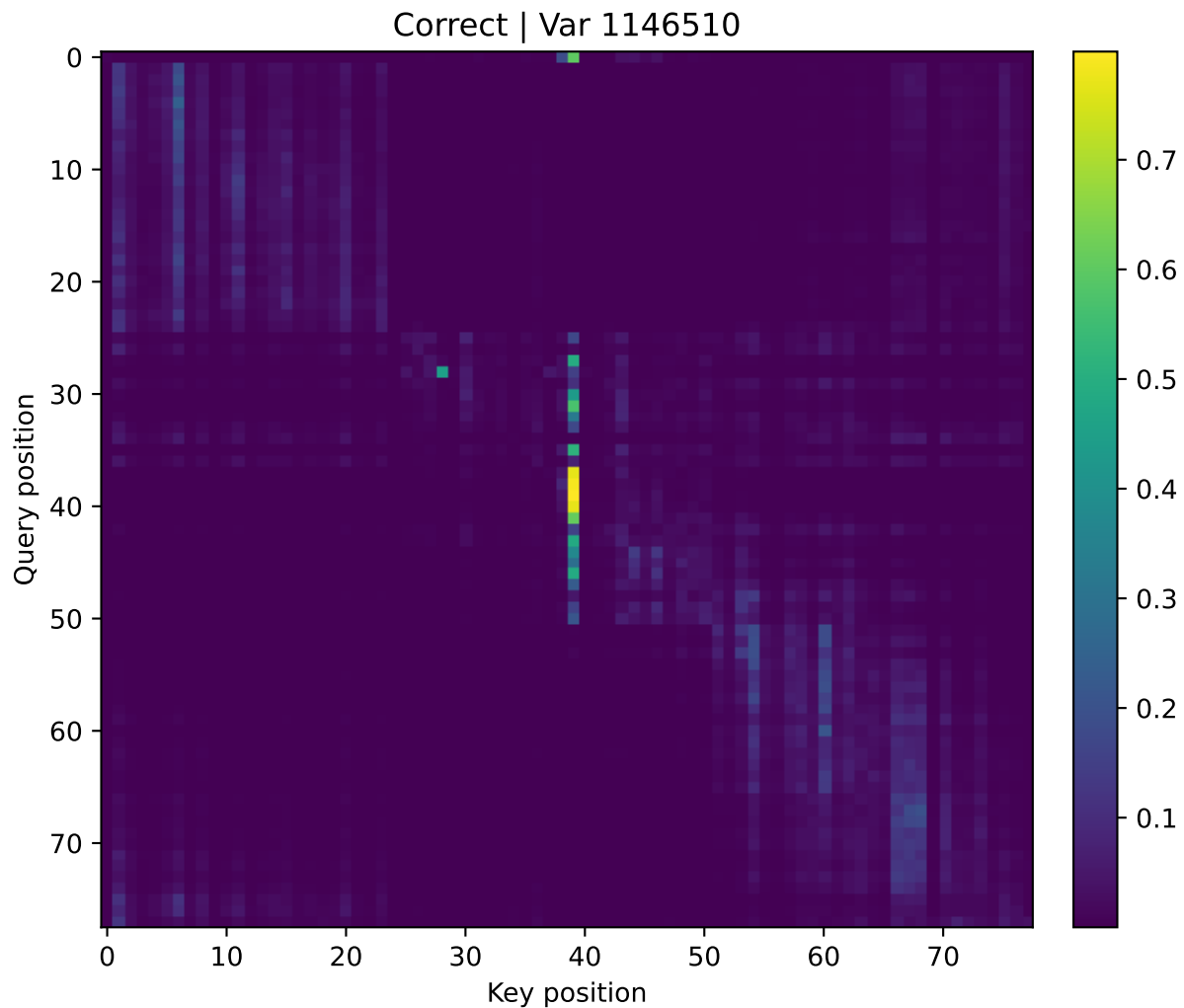
NumberOfSubmitters: 1

RecordType: classified

CLNSIG: Likely\_benign

actual\_label: 0

prediction: 0



--- Correctly Predicted Variant ---

Top head → layer 6, head 11, score 0.8463

VariationID: 862627

VariationName: NM\_000393.5(COL5A2):c.3147+1G>A

VariantType: single nucleotide variant

GeneSymbol: COL5A2

OMIM: 120190.0

AssemblyChr: 2

Start: 189049346

Stop: 189049346

RefAlleleVCF: C

AltAlleleVCF: T

VariantLength: 1

ProteinChange: nan

HGVS\_coding: NM\_000393.5:c.3147+1G>A

HGVS\_protein: nan

DateLastUpdated: 2025-02-26

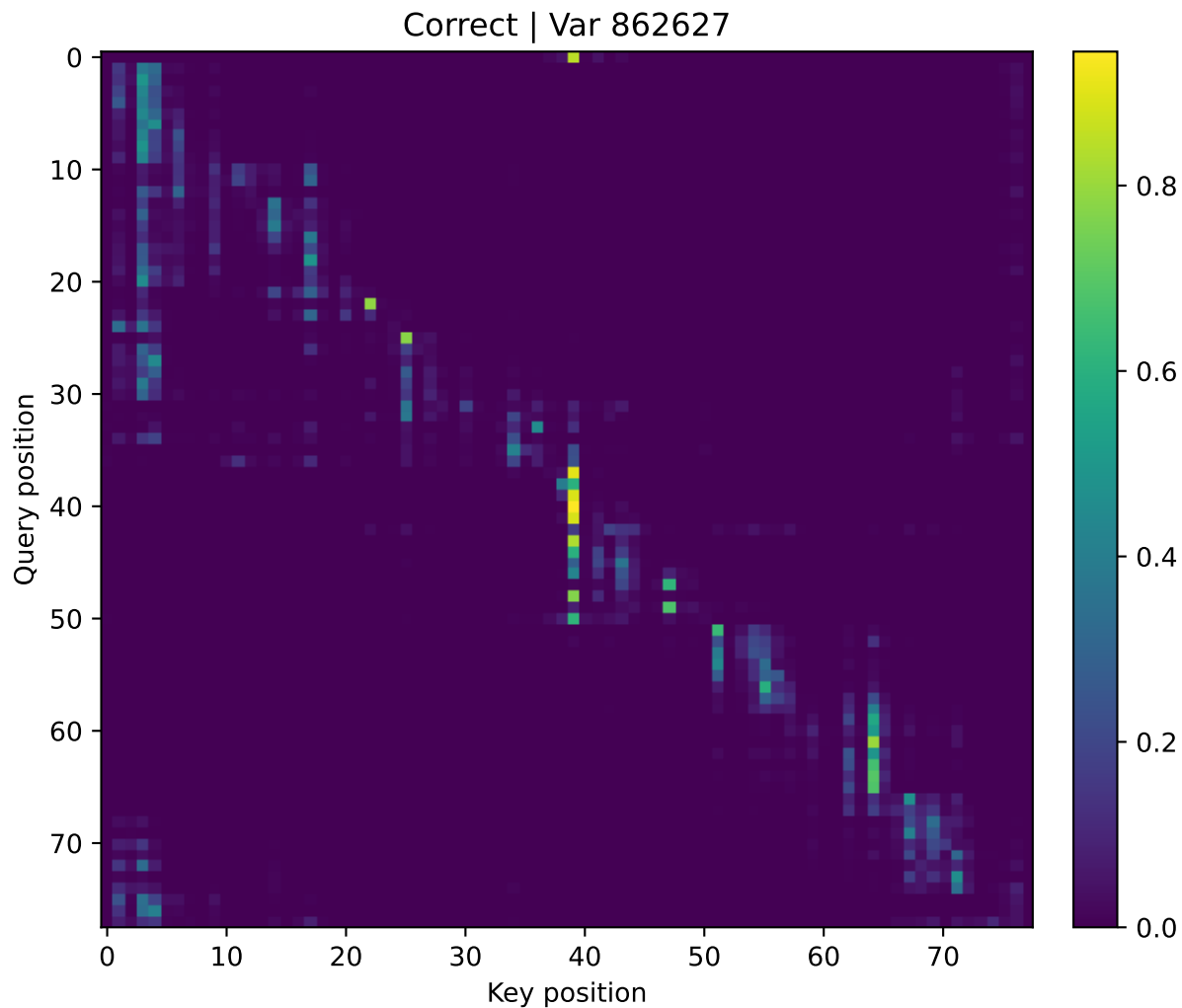
NumberOfSubmitters: 1

RecordType: classified

CLNSIG: Pathogenic

actual\_label: 1

prediction: 1



--- Correctly Predicted Variant ---

Top head → layer 13, head 16, score 0.8555

VariationID: 2586046

VariationName: NM\_014000.3(VCL):c.875-4T>G

VariantType: single nucleotide variant

GeneSymbol: VCL

OMIM: 193065.0

AssemblyChr: 10

Start: 74083362

Stop: 74083362

RefAlleleVCF: T

AltAlleleVCF: G

VariantLength: 1

ProteinChange: nan

HGVS\_coding: NM\_003373.4:c.875-4T>G

HGVS\_protein: nan

DateLastUpdated: 2025-05-19

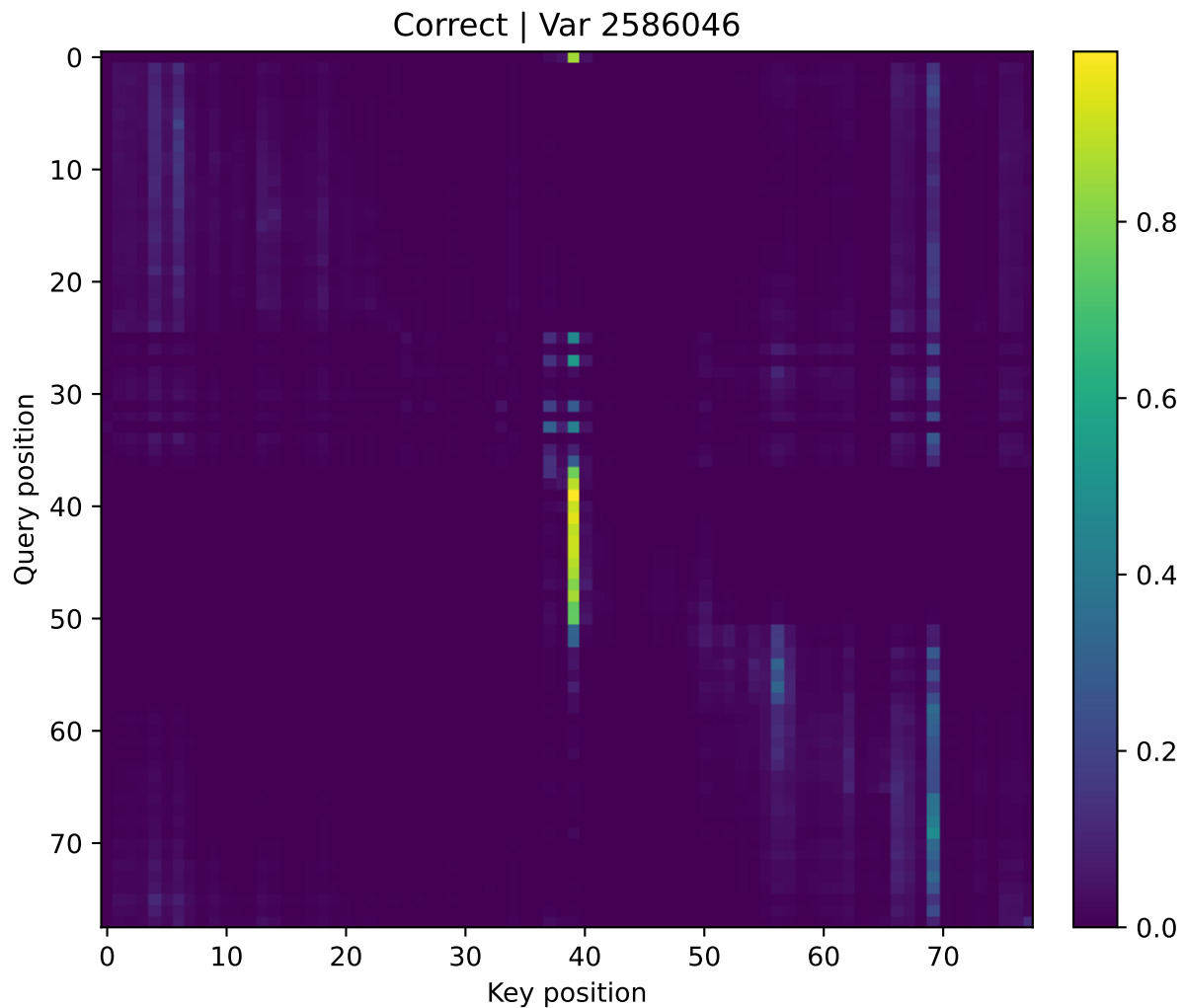
NumberOfSubmitters: 1

RecordType: classified

CLNSIG: Likely\_benign

actual\_label: 0

prediction: 0



--- Correctly Predicted Variant ---

Top head → layer 9, head 14, score 0.9269

VariationID: 3725474

VariationName: NM\_000426.4(LAMA2):c.101dup (p.His34fs)

VariantType: Duplication

GeneSymbol: LAMA2

OMIM: 156225.0

AssemblyChr: 6

Start: 128883345

Stop: 128883346

RefAlleleVCF: C

AltAlleleVCF: CA

VariantLength: 1

ProteinChange: H34fs

HGVS\_coding: NM\_000426.4:c.101dup

HGVS\_protein: NP\_000417.3:p.His34fs

DateLastUpdated: 2025-04-08

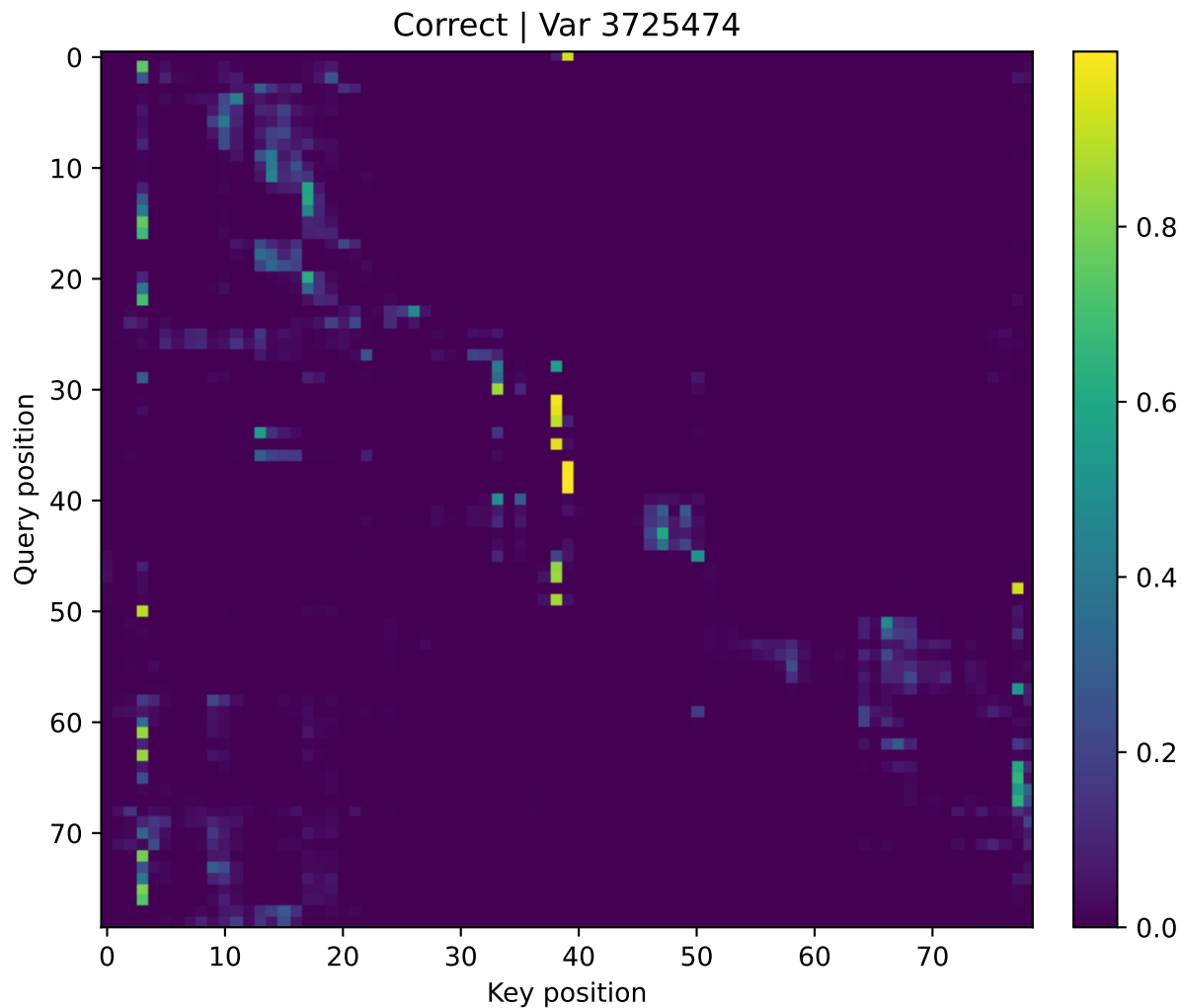
NumberOfSubmitters: 1

RecordType: classified

CLNSIG: Pathogenic

actual\_label: 1

prediction: 1



--- Correctly Predicted Variant ---

Top head → layer 10, head 13, score 0.7816

VariationID: 1935973

VariationName: NM\_006766.5(KAT6A):c.5667C>T (p.Arg1889=)

VariantType: single nucleotide variant

GeneSymbol: KAT6A

OMIM: 601408.0

AssemblyChr: 8

Start: 41932553

Stop: 41932553

RefAlleleVCF: G

AltAlleleVCF: A

VariantLength: 1

ProteinChange: nan

HGVS\_coding: NM\_006766.5:c.5667C>T

HGVS\_protein: NP\_006757.2:p.Arg1889=

DateLastUpdated: 2025-05-17

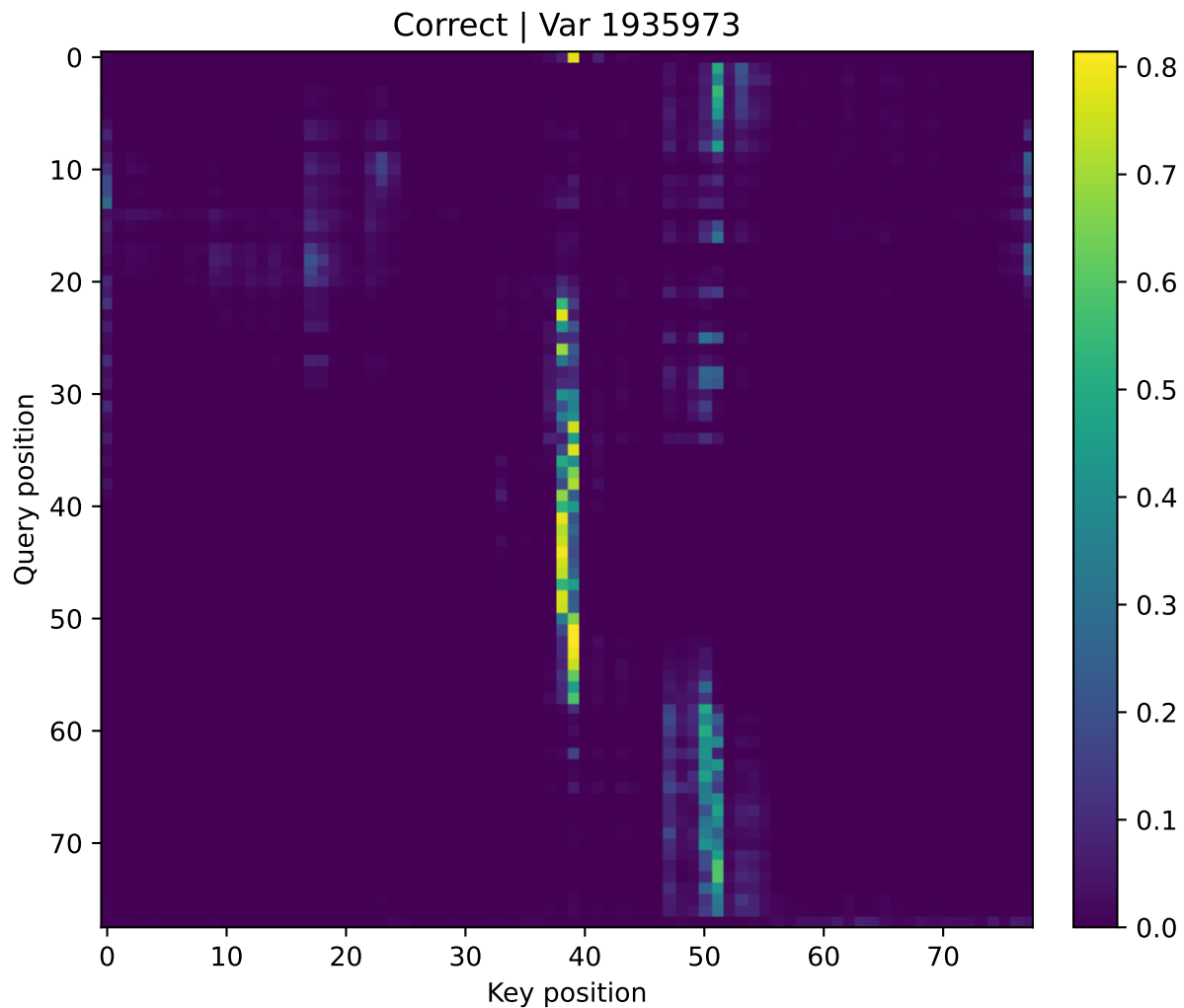
NumberOfSubmitters: 1

RecordType: classified

CLNSIG: Benign

actual\_label: 0

prediction: 0



--- Correctly Predicted Variant ---

Top head → layer 7, head 11, score 0.6800

VariationID: 2833150

VariationName: NM\_005908.4(MANBA):c.2244C>A (p.Cys748Ter)

VariantType: single nucleotide variant

GeneSymbol: MANBA

OMIM: 609489.0

AssemblyChr: 4

Start: 102634959

Stop: 102634959

RefAlleleVCF: G

AltAlleleVCF: T

VariantLength: 1

ProteinChange: C748\*

HGVS\_coding: NM\_005908.4:c.2244C>A

HGVS\_protein: NP\_005899.3:p.Cys748Ter

DateLastUpdated: 2025-05-19

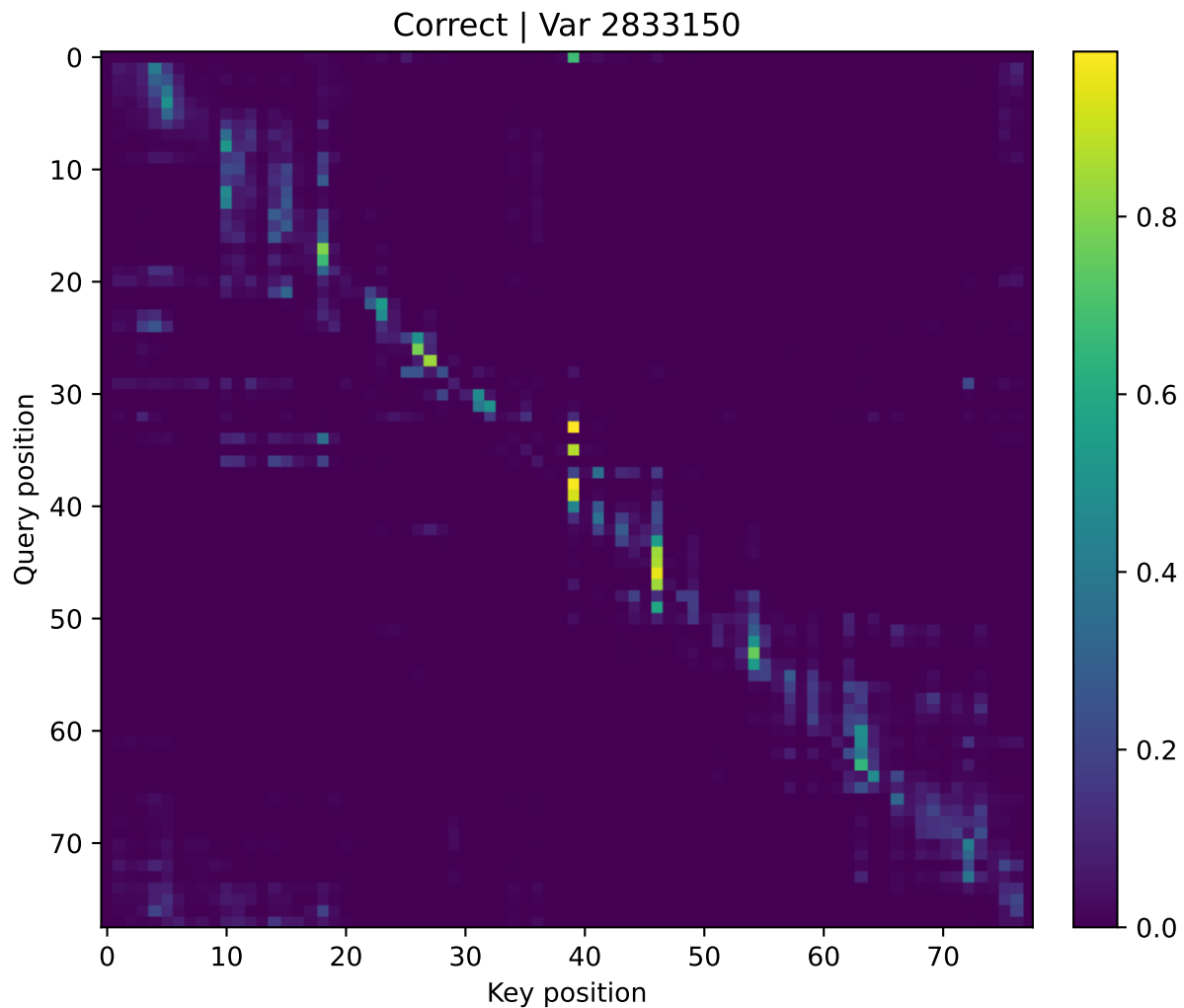
NumberOfSubmitters: 1

RecordType: classified

CLNSIG: Pathogenic

actual\_label: 1

prediction: 1





--- Correctly Predicted Variant ---

Top head → layer 12, head 11, score 0.8508

VariationID: 2869561

VariationName: NM\_000732.6(CD3D):c.271C>T (p.Arg91Ter)

VariantType: single nucleotide variant

GeneSymbol: CD3D

OMIM: 186790.0

AssemblyChr: 11

Start: 118340378

Stop: 118340378

RefAlleleVCF: G

AltAlleleVCF: A

VariantLength: 1

ProteinChange: R91\*

HGVS\_coding: NM\_000732.6:c.271C>T

HGVS\_protein: NP\_000723.1:p.Arg91Ter

DateLastUpdated: 2025-05-19

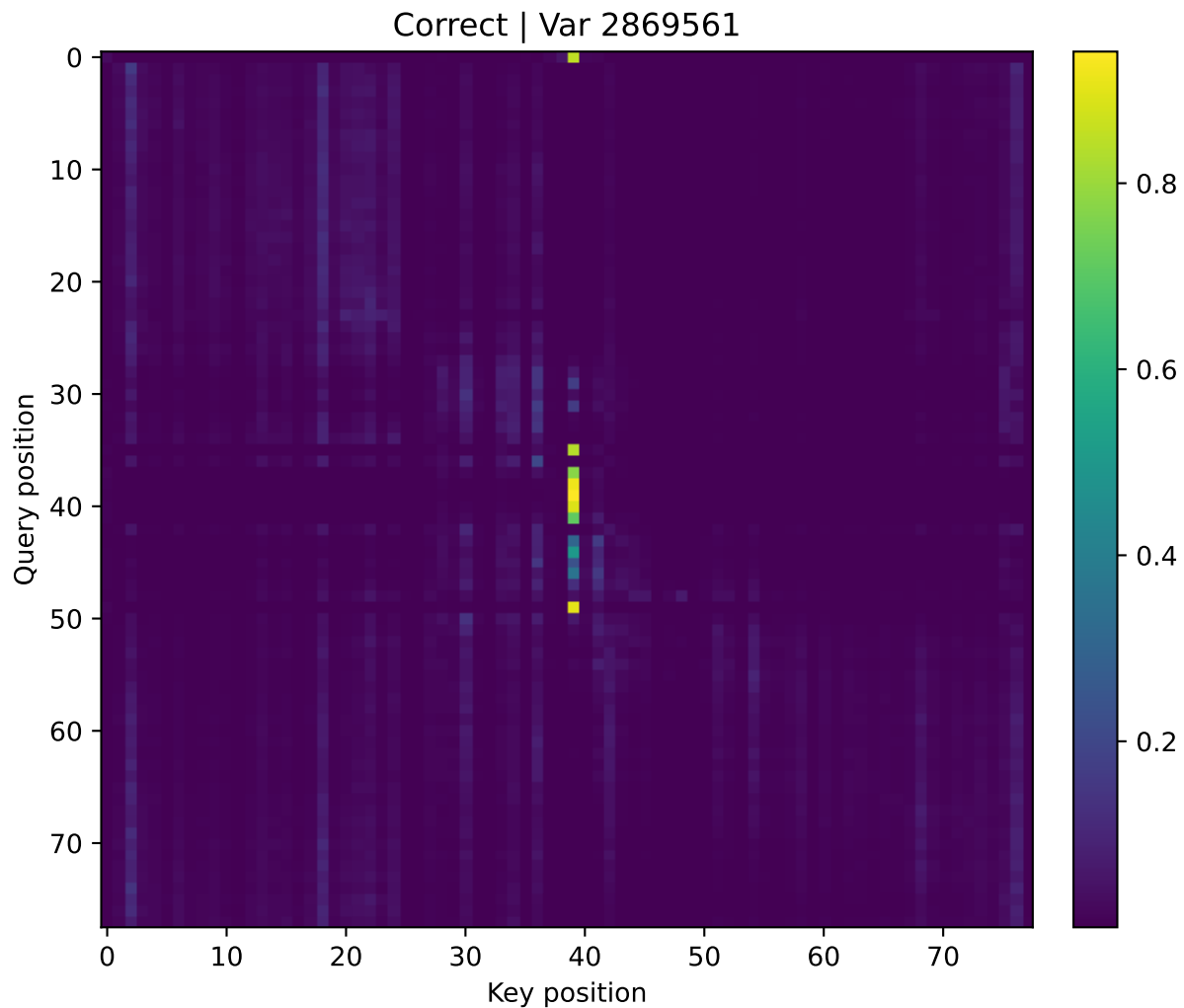
NumberOfSubmitters: 1

RecordType: classified

CLNSIG: Pathogenic

actual\_label: 1

prediction: 1



--- Correctly Predicted Variant ---

Top head → layer 9, head 14, score 0.8459

VariationID: 378862

VariationName: NM\_025216.3(WNT10A):c.1168G>T (p.Glu390Ter)

VariantType: single nucleotide variant

GeneSymbol: WNT10A

OMIM: 606268.0

AssemblyChr: 2

Start: 218893185

Stop: 218893185

RefAlleleVCF: G

AltAlleleVCF: T

VariantLength: 1

ProteinChange: E390\*

HGVS\_coding: NM\_025216.3:c.1168G>T

HGVS\_protein: NP\_079492.2:p.Glu390Ter

DateLastUpdated: 2025-05-17

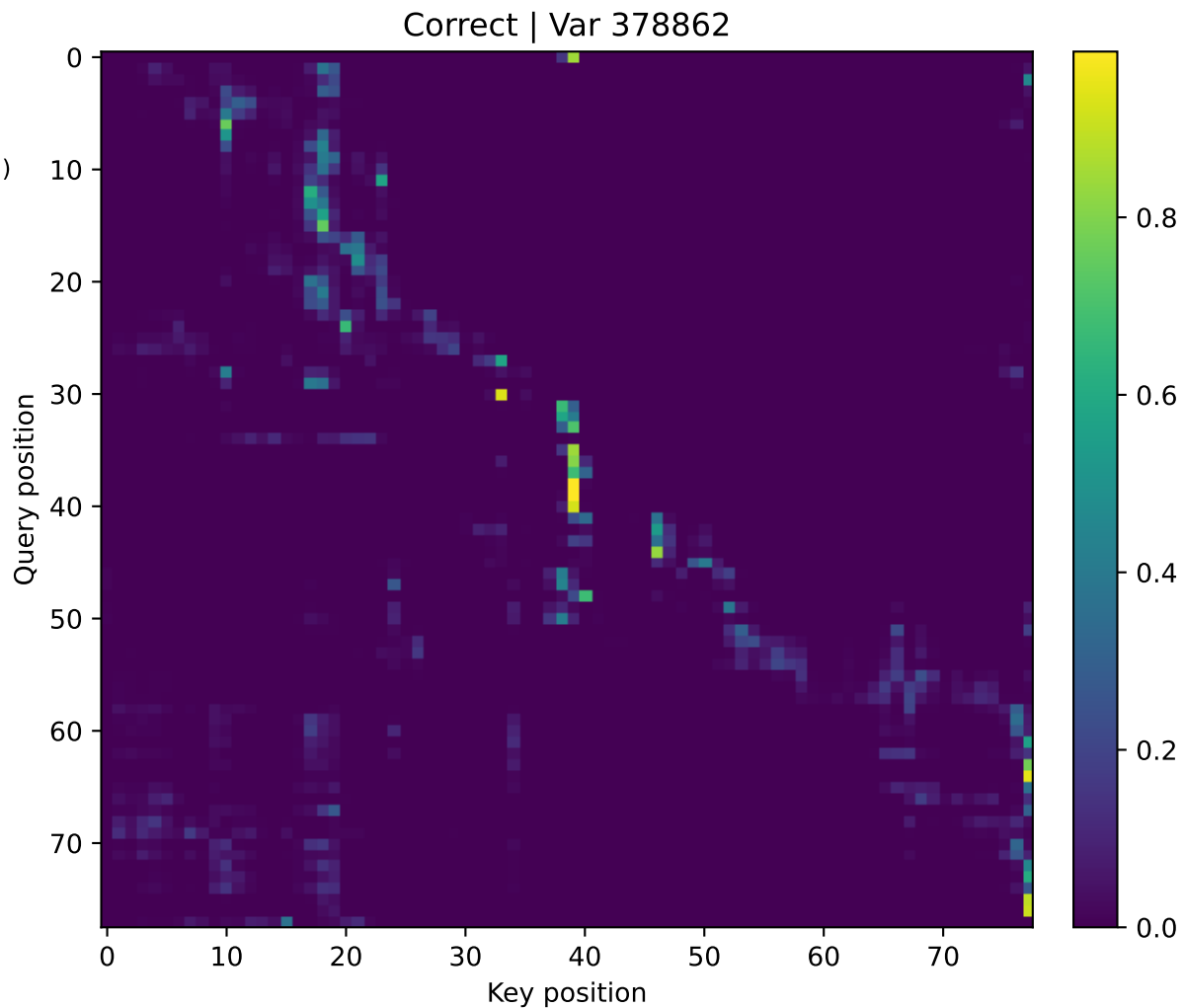
NumberOfSubmitters: 3

RecordType: classified

CLNSIG: Pathogenic/Likely\_pathogenic

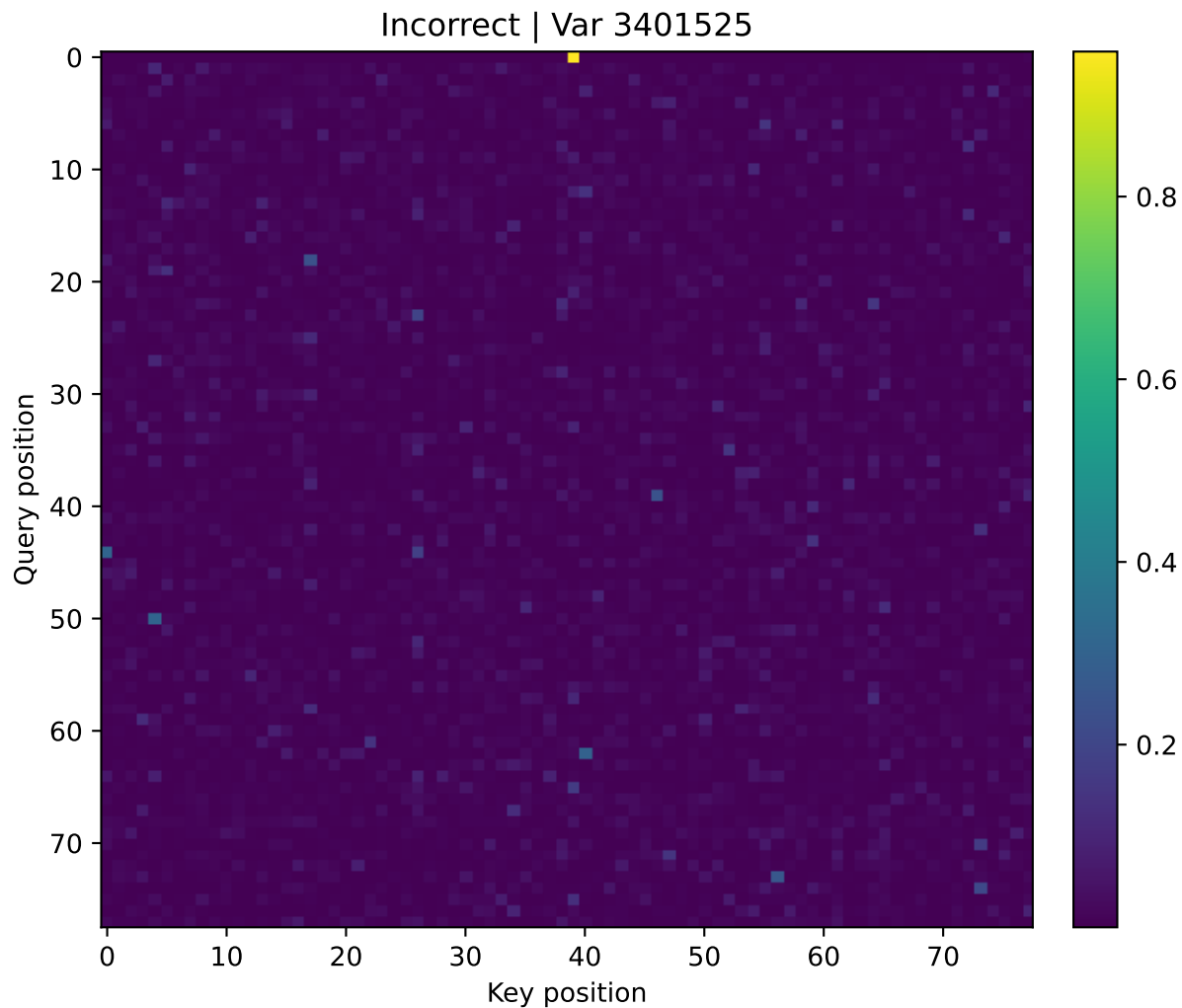
actual\_label: 1

prediction: 1



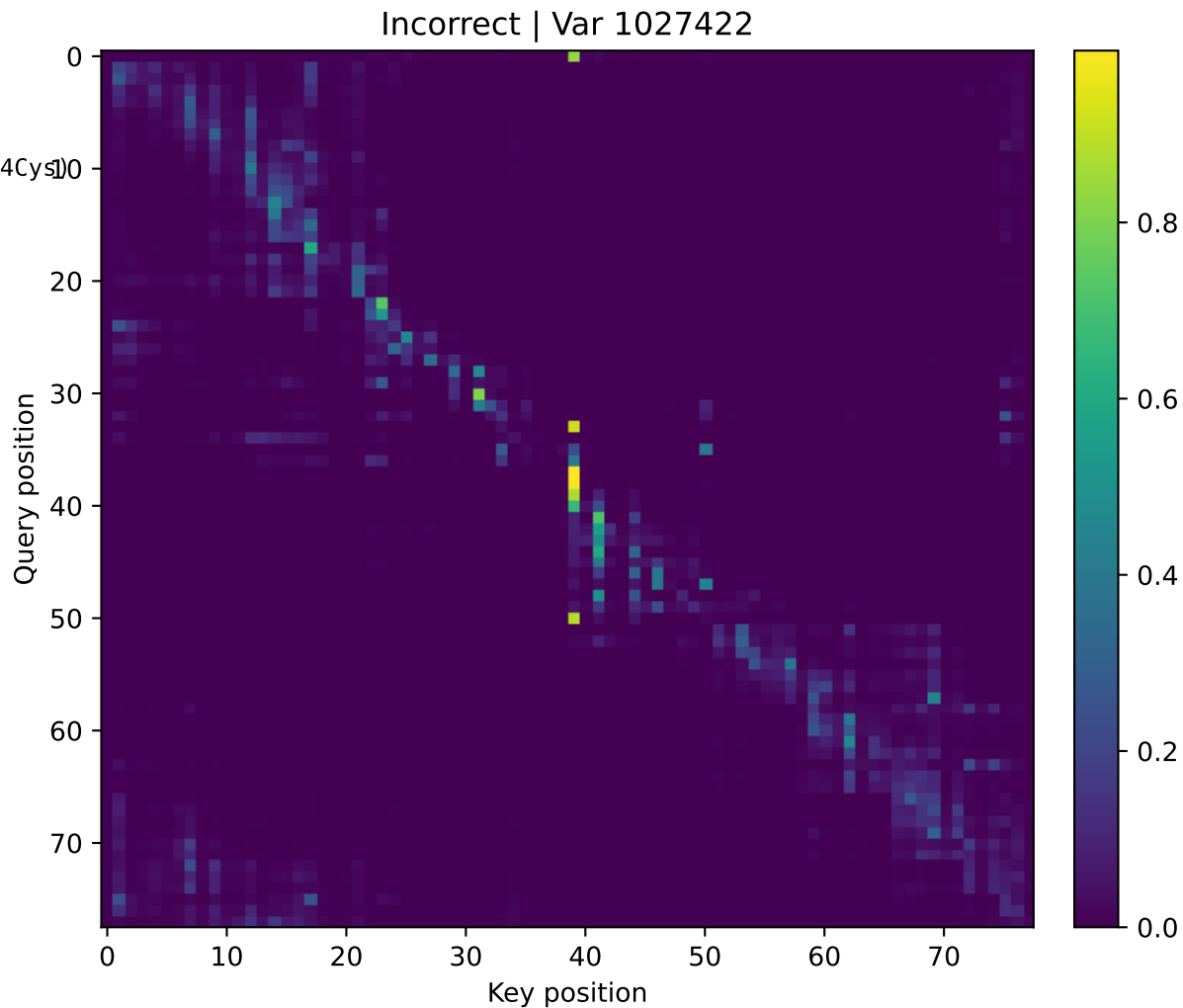
--- Incorrectly Predicted Variant ---  
Top head → layer 0, head 7, score 0.9588

VariationID: 3401525  
VariationName: NM\_001150.3(ANPEP):c.1775G>A (p.Arg592Lys)  
VariantType: single nucleotide variant  
GeneSymbol: ANPEP  
OMIM: 151530.0  
AssemblyChr: 15  
Start: 89801155  
Stop: 89801155  
RefAlleleVCF: C  
AltAlleleVCF: T  
VariantLength: 1  
ProteinChange: R592K  
HGVS\_coding: NM\_001150.3:c.1775G>A  
HGVS\_protein: NP\_001141.2:p.Arg592Lys  
DateLastUpdated: 2025-01-19  
NumberOfSubmitters: 1  
RecordType: classified  
CLNSIG: Likely\_benign  
actual\_label: 0  
prediction: 1



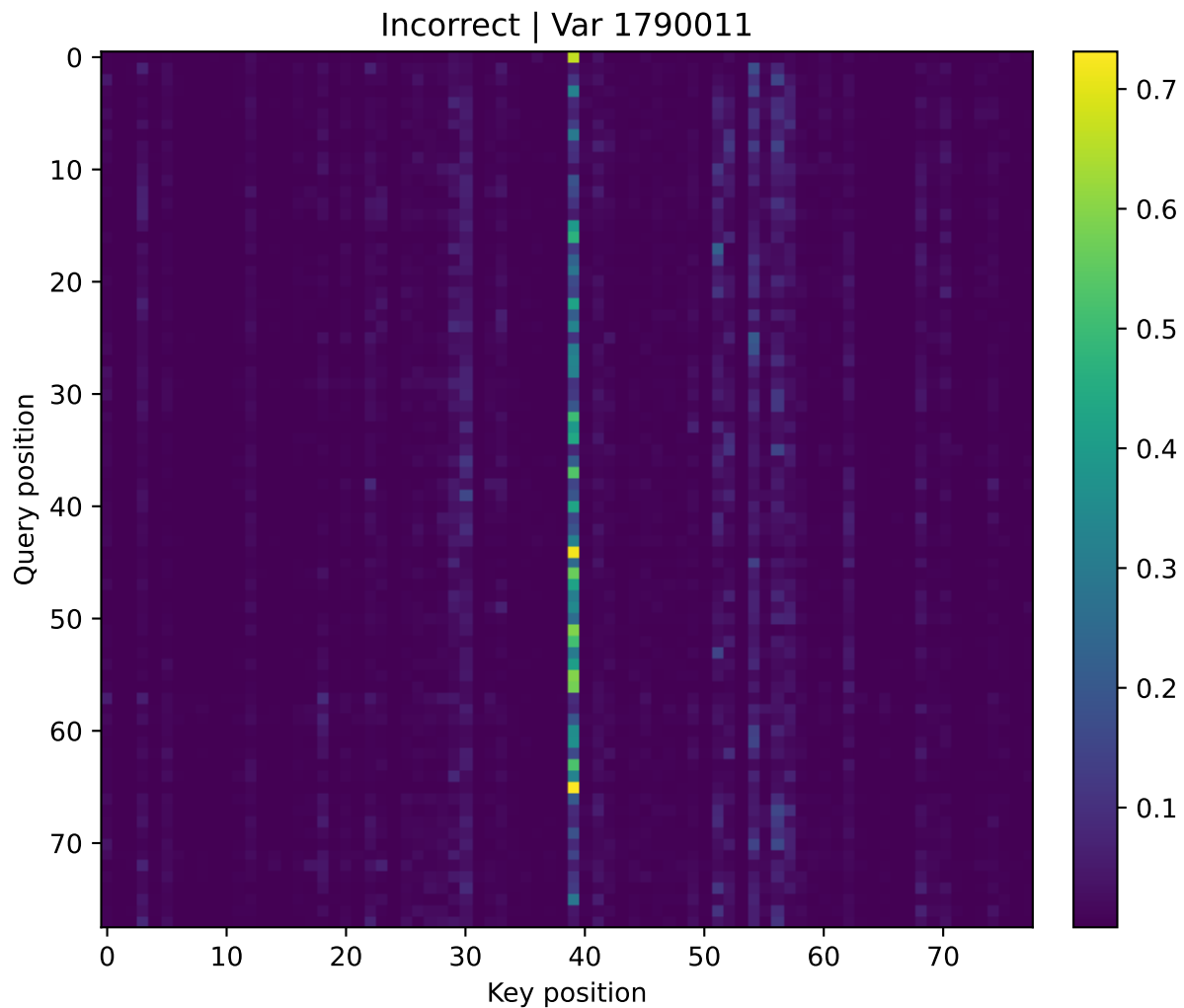
--- Incorrectly Predicted Variant ---  
Top head → layer 7, head 11, score 0.8344

VariationID: 1027422  
VariationName: NM\_001130438.3(SPTAN1):c.4870C>T (p.Arg1624Cys)  
VariantType: single nucleotide variant  
GeneSymbol: SPTAN1  
OMIM: 182810.0  
AssemblyChr: 9  
Start: 128611810  
Stop: 128611810  
RefAlleleVCF: C  
AltAlleleVCF: T  
VariantLength: 1  
ProteinChange: R1599C  
HGVS\_coding: NM\_001130438.3:c.4870C>T  
HGVS\_protein: NP\_001123910.1:p.Arg1624Cys  
DateLastUpdated: 2023-12-25  
NumberOfSubmitters: 1  
RecordType: classified  
CLNSIG: Likely\_pathogenic  
actual\_label: 1  
prediction: 0



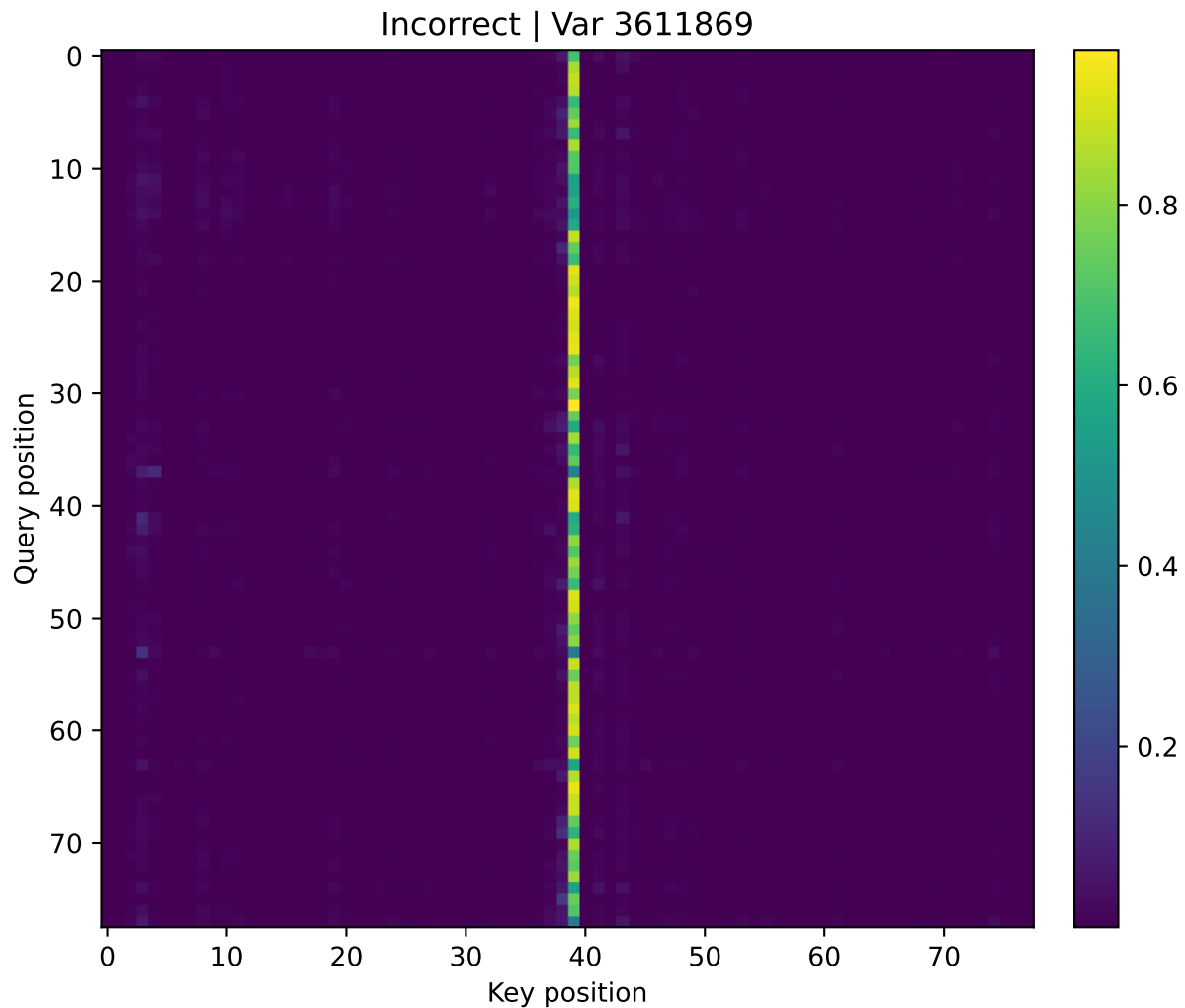
--- Incorrectly Predicted Variant ---  
Top head → layer 1, head 6, score 0.6653

VariationID: 1790011  
VariationName: NM\_020774.4(MIB1):c.2351A>G (p.Asn784Ser)  
VariantType: single nucleotide variant  
GeneSymbol: MIB1  
OMIM: 608677.0  
AssemblyChr: 18  
Start: 21847083  
Stop: 21847083  
RefAlleleVCF: A  
AltAlleleVCF: G  
VariantLength: 1  
ProteinChange: N784S  
HGVS\_coding: NM\_020774.4:c.2351A>G  
HGVS\_protein: NP\_065825.1:p.Asn784Ser  
DateLastUpdated: 2025-05-17  
NumberOfSubmitters: 1  
RecordType: classified  
CLNSIG: Likely\_benign  
actual\_label: 0  
prediction: 1



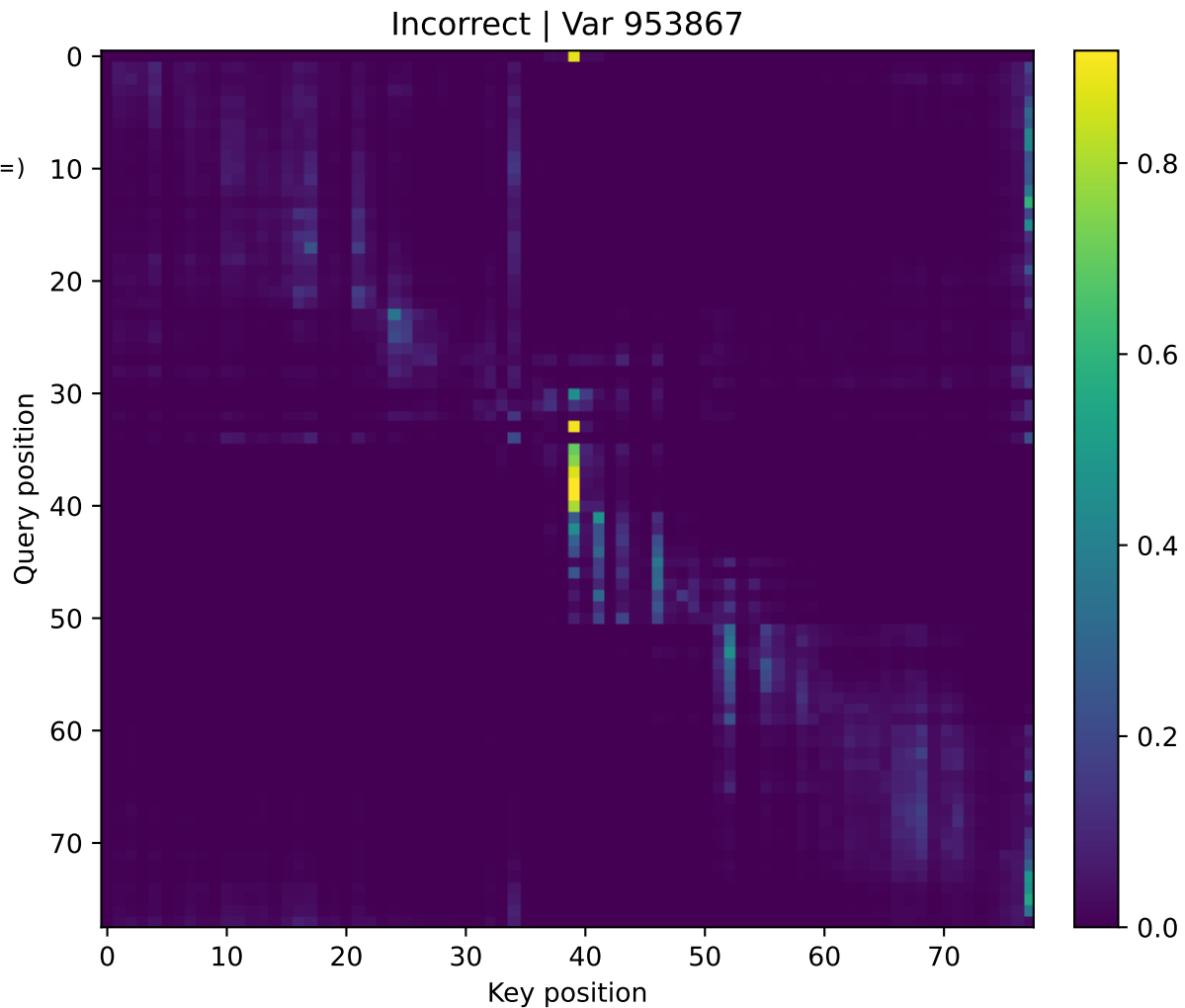
--- Incorrectly Predicted Variant ---  
Top head → layer 1, head 3, score 0.6875

VariationID: 3611869  
VariationName: NM\_001039141.3(TRIOBP):c.5380-12T>C  
VariantType: single nucleotide variant  
GeneSymbol: LOC126863145  
OMIM: 609761.0  
AssemblyChr: 22  
Start: 37754865  
Stop: 37754865  
RefAlleleVCF: T  
AltAlleleVCF: C  
VariantLength: 1  
ProteinChange: nan  
HGVS\_coding: NM\_001039141.3:c.5380-12T>C  
HGVS\_protein: nan  
DateLastUpdated: 2025-04-08  
NumberOfSubmitters: 1  
RecordType: classified  
CLNSIG: Likely\_benign  
actual\_label: 0  
prediction: 1



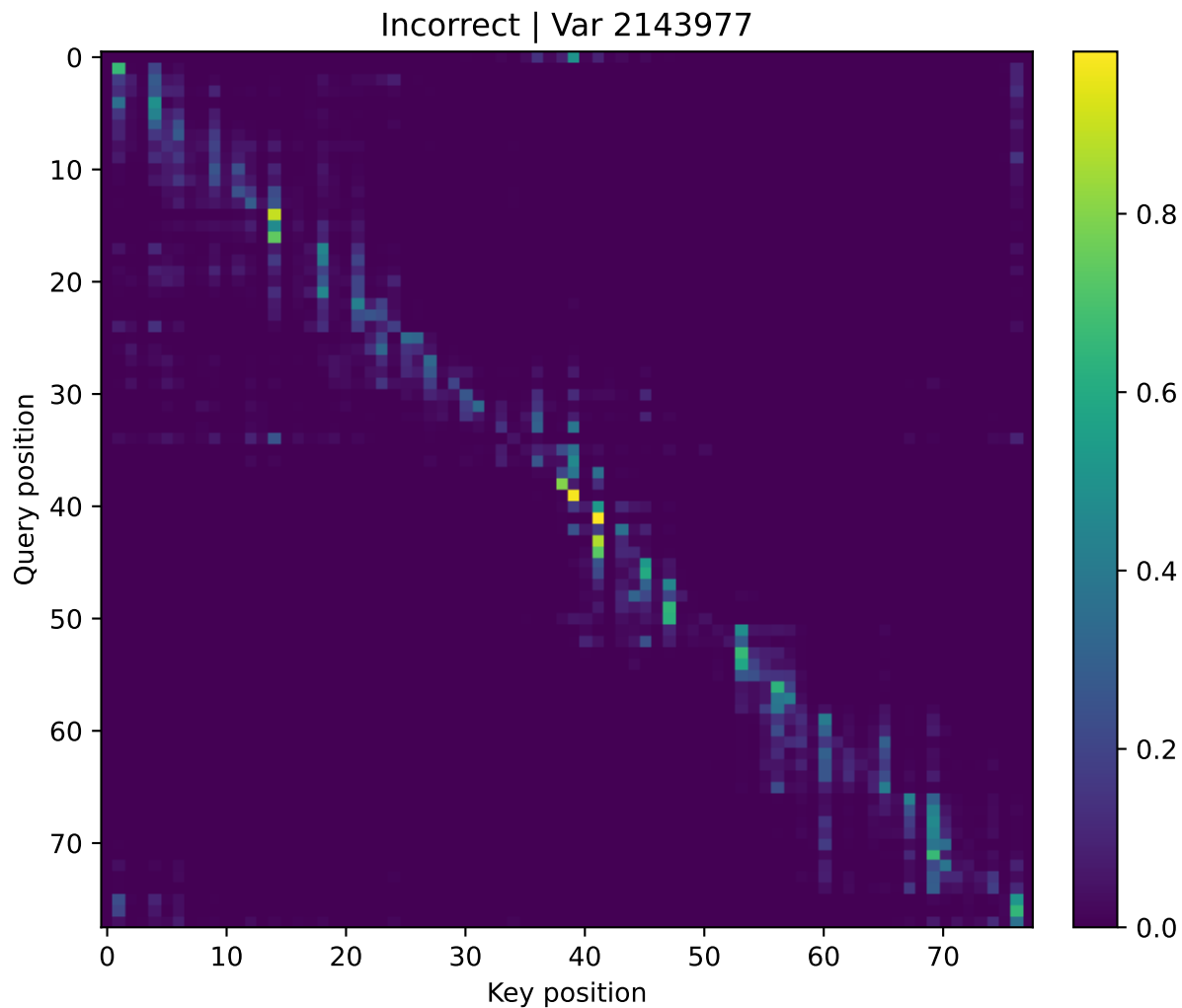
--- Incorrectly Predicted Variant ---  
Top head → layer 11, head 17, score 0.8835

VariationID: 953867  
VariationName: NM\_001113378.2(FANCI):c.3171G>A (p.Leu1057=)  
VariantType: single nucleotide variant  
GeneSymbol: FANCI  
OMIM: 611360.0  
AssemblyChr: 15  
Start: 89305227  
Stop: 89305227  
RefAlleleVCF: G  
AltAlleleVCF: A  
VariantLength: 1  
ProteinChange: nan  
HGVS\_coding: NM\_001113378.2:c.3171G>A  
HGVS\_protein: NP\_001106849.1:p.Leu1057=  
DateLastUpdated: 2025-03-22  
NumberOfSubmitters: 1  
RecordType: classified  
CLNSIG: Likely\_benign  
actual\_label: 0  
prediction: 1



--- Incorrectly Predicted Variant ---  
Top head → layer 6, head 14, score 0.5040

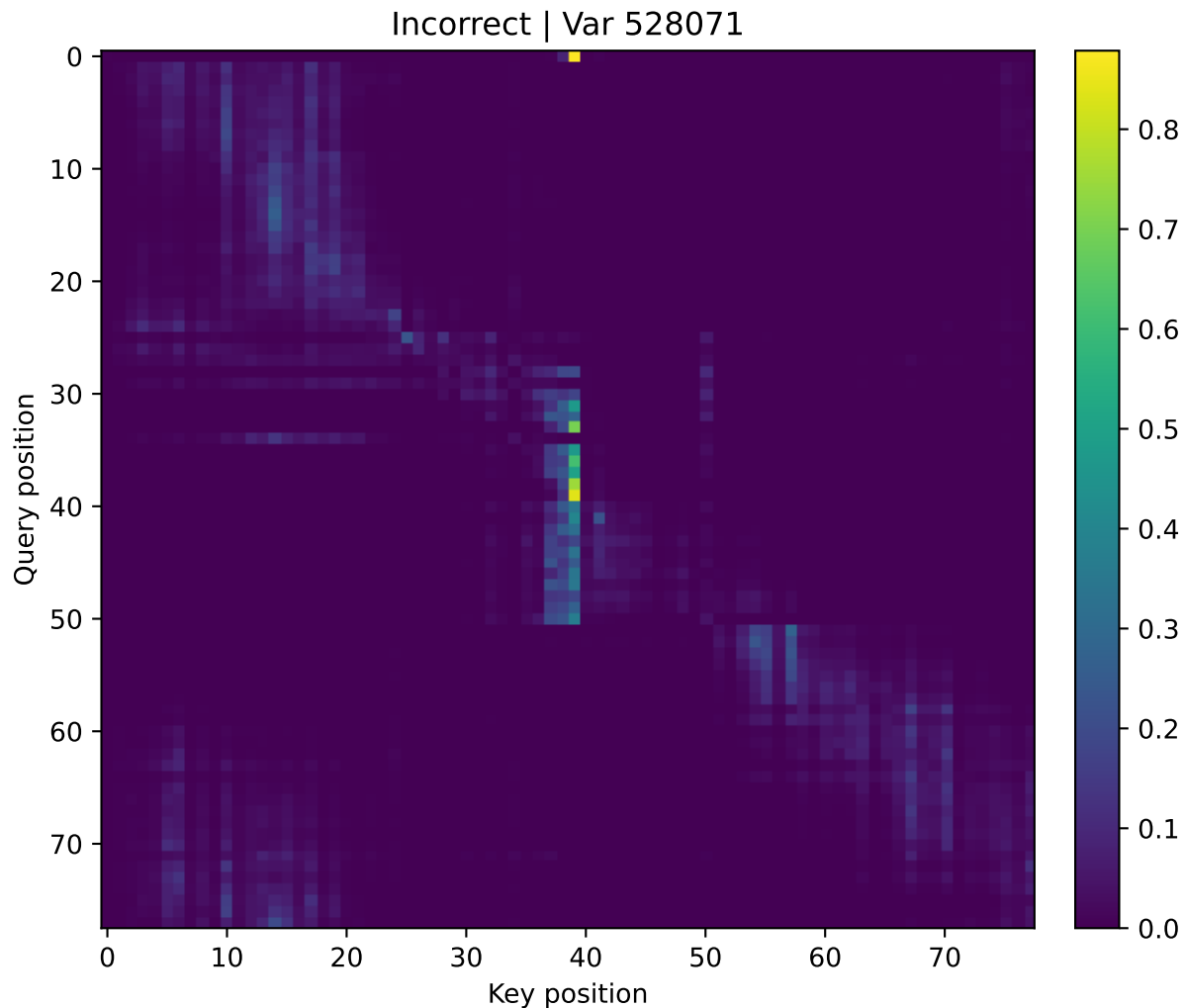
VariationID: 2143977  
VariationName: NM\_000245.4(MET):c.3633-13G>A  
VariantType: single nucleotide variant  
GeneSymbol: MET  
OMIM: 164860.0  
AssemblyChr: 7  
Start: 116783291  
Stop: 116783291  
RefAlleleVCF: G  
AltAlleleVCF: A  
VariantLength: 1  
ProteinChange: nan  
HGVS\_coding: NM\_001127500.3:c.3687-13G>A  
HGVS\_protein: nan  
DateLastUpdated: 2025-05-19  
NumberOfSubmitters: 2  
RecordType: classified  
CLNSIG: Likely\_benign  
actual\_label: 0  
prediction: 1





--- Incorrectly Predicted Variant ---  
Top head → layer 11, head 18, score 0.8786

VariationID: 528071  
VariationName: NM\_001114753.3(ENG):c.683C>A (p.Ser228Ter)  
VariantType: single nucleotide variant  
GeneSymbol: ENG  
OMIM: 131195.0  
AssemblyChr: 9  
Start: 127825701  
Stop: 127825701  
RefAlleleVCF: G  
AltAlleleVCF: T  
VariantLength: 1  
ProteinChange: S228\*  
HGVS\_coding: NM\_000118.4:c.683C>A  
HGVS\_protein: NP\_000109.1:p.Ser228Ter  
DateLastUpdated: 2025-02-26  
NumberOfSubmitters: 1  
RecordType: classified  
CLNSIG: Pathogenic  
actual\_label: 1  
prediction: 0



--- Incorrectly Predicted Variant ---

Top head → layer 6, head 11, score 0.7928

VariationID: 1993076

VariationName: NM\_205768.3(ZBTB18):c.362T>G (p.Val121Gly)

VariantType: single nucleotide variant

GeneSymbol: ZBTB18

OMIM: 608433.0

AssemblyChr: 1

Start: 244054136

Stop: 244054136

RefAlleleVCF: T

AltAlleleVCF: G

VariantLength: 1

ProteinChange: V121G

HGVS\_coding: NM\_001278196.2:c.335T>G

HGVS\_protein: NP\_001265125.1:p.Val112Gly

DateLastUpdated: 2025-05-17

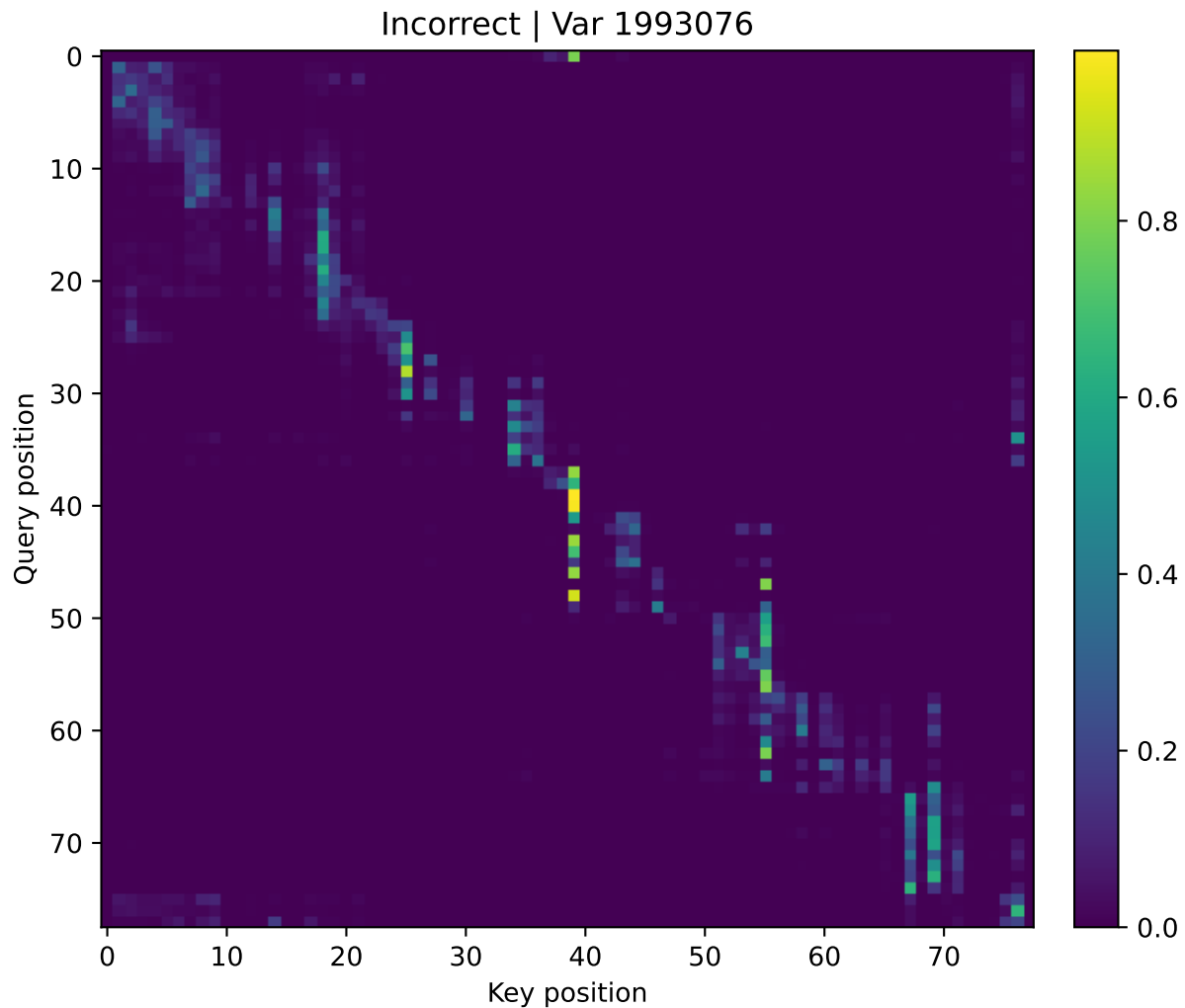
NumberOfSubmitters: 1

RecordType: classified

CLNSIG: Likely\_benign

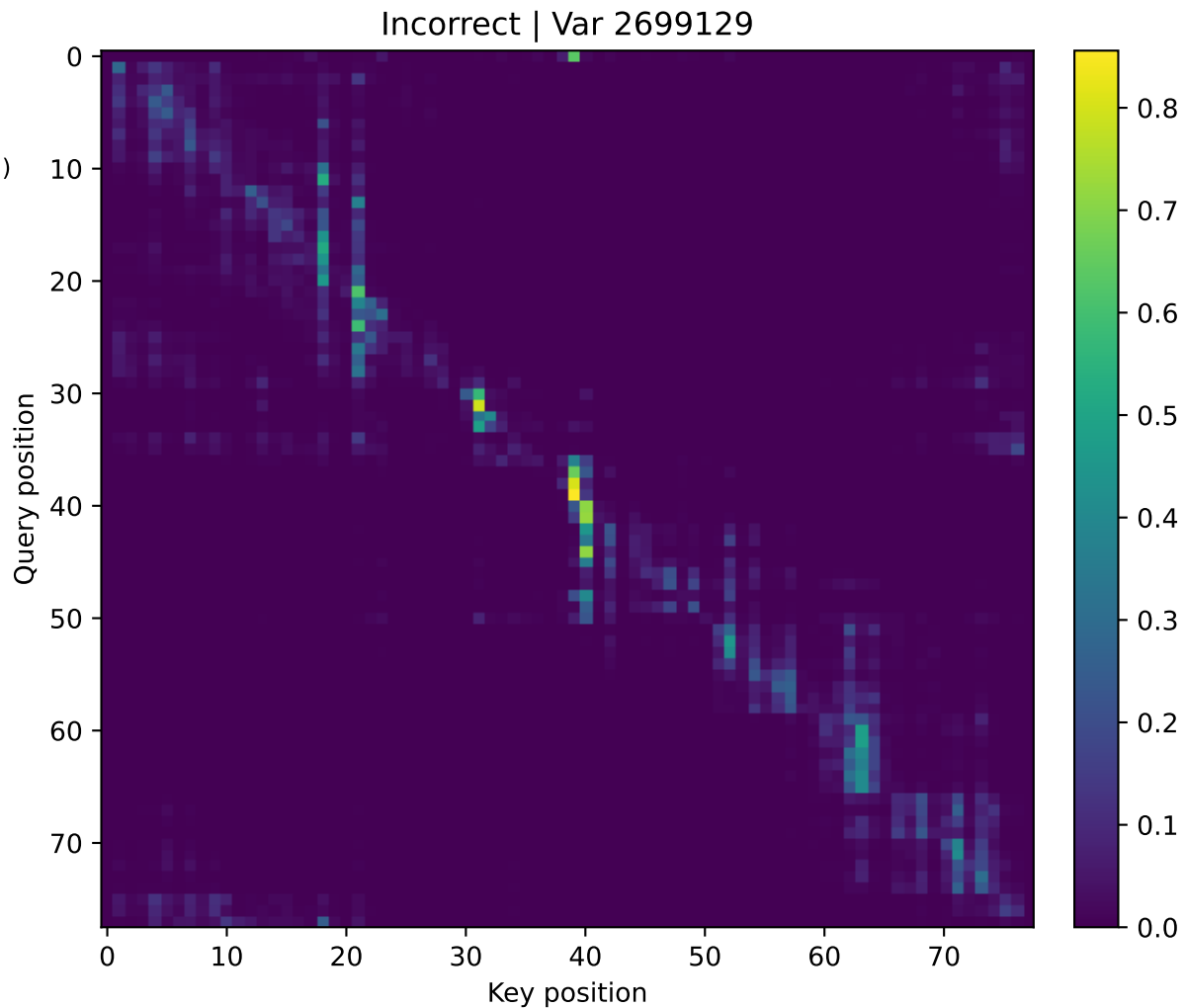
actual\_label: 0

prediction: 1



--- Incorrectly Predicted Variant ---  
Top head → layer 7, head 4, score 0.6365

VariationID: 2699129  
VariationName: NM\_005559.4(LAMA1):c.3888T>A (p.Tyr1296Ter)  
VariantType: single nucleotide variant  
GeneSymbol: LAMA1  
OMIM: 150320.0  
AssemblyChr: 18  
Start: 7009352  
Stop: 7009352  
RefAlleleVCF: A  
AltAlleleVCF: T  
VariantLength: 1  
ProteinChange: Y1296\*  
HGVS\_coding: NM\_005559.4:c.3888T>A  
HGVS\_protein: NP\_005550.2:p.Tyr1296Ter  
DateLastUpdated: 2025-05-19  
NumberOfSubmitters: 1  
RecordType: classified  
CLNSIG: Pathogenic  
actual\_label: 1  
prediction: 0



--- Incorrectly Predicted Variant ---  
Top head → layer 12, head 11, score 0.8014

VariationID: 1921276  
VariationName: NM\_024580.6(EFL1):c.1444+16G>A  
VariantType: single nucleotide variant  
GeneSymbol: EFL1  
OMIM: 617538.0  
AssemblyChr: 15  
Start: 82220062  
Stop: 82220062  
RefAlleleVCF: C  
AltAlleleVCF: T  
VariantLength: 1  
ProteinChange: nan  
HGVS\_coding: NM\_001040610.3:c.1291+16G>A  
HGVS\_protein: nan  
DateLastUpdated: 2025-05-17  
NumberOfSubmitters: 1  
RecordType: classified  
CLNSIG: Likely\_benign  
actual\_label: 0  
prediction: 1

