



VariantValidator

Submitted Variant

NM_002693.2:c.2243G>C

- Selected genome build: GRCh38
- Map location: 15q26.1
- Transcript Flag: RefSeq Select
- CCDS ID: [CCDS10350.1](#)

Versions

- [VariantValidator](#) 2.1.1.dev103+gced0497
- [vv_hgvs](#) 2.2.1.dev0+g69b1a7c.d20230119
- [VVDb](#) vvdb_2022_11
- [VVTA](#) vvta_2022_11_1
- [VVSeqRepo](#) VV_SR_2022_11/master

Warnings output during validation

- A more recent version of the selected reference sequence NM_002693.2 is available (NM_002693.3): NM_002693.3:c.2243G>C MUST be fully validated prior to use in reports: select_variants=NM_002693.3:c.2243G>C

Recommended Variant Descriptions

1. HGVS guidelines recommend using genomic and transcript descriptions in all publications
2. Use of the three- or one-letter amino acid alphabet is optional, but three-letter is recommended

Genomic descriptions

Reference Sequence Type	Variant Description
Chromosomal GRCh37	NC_000015.9:g.89866657C>G
Chromosomal GRCh38	NC_000015.10:g.89323426C>G

Transcript and protein descriptions

Reference Sequence Type	Variant Description
Transcript	NM_002693.2:c.2243G>C
Protein three letter code	NP_002684.1:p.(Trp748Ser)
Protein single letter code	NP_002684.1:p.(W748S)

Gene Information

Attribute	Identifier	Source
Symbol	POLG	HGNC
Name	DNA polymerase gamma, catalytic subunit	HGNC
HGNC ID	HGNC:9179	HGNC



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