VariantValidator

Submitted Variant

NM_002693.2:c.2591A>G

- 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.2591A>G MUST be fully validated prior to use in reports: select_variants=NM_002693.3:c.2591A>G

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.89864974T>C
Chromosomal GRCh38	NC_000015.10:g.89321743T>C

Transcript and protein descriptions

Reference Sequence Type	Variant Description
Transcript	NM_002693.2:c.2591A>G
Protein three letter code	NP_002684.1:p.(Asn864Ser)
Protein single letter code	NP_002684.1:p.(N864S)

Gene Information

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