AIFM1: c.1195G>A (p.Gly399Ser) chrX:129270050

PROVEAN

number of clusters: 30

number of supporting sequences used: 112

G399S -5.961 Deleterious

SIFT

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| User Input | ENSP | Pos | Ref | Subst | Prediction | SIFT Score | Median Information Content | # Seqs |
| ENST00000287295,G399S    | ENSP00000287295    | 399 | G | S | DAMAGING | 0 | 2.2 | 177 |

**PolyPhen-2 report for O95831 G399S**

**Query**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Protein Acc** | **Position** | **AA1** | **AA2** | **Description** |
| [O95831](http://www.uniprot.org/uniprot/O95831) | 399 | G | S | Canonical; RecName: Full=Apoptosis-inducing factor 1, mitochondrial; EC=1.-.-.-; AltName: Full=Programmed cell death protein 8; Flags: Precursor; Length: 613 |

**Results**

|  |  |
| --- | --- |
| **Prediction/Confidence** | ***PolyPhen-2 v2.2.2r398*** |

**HumDiv**

This mutation is predicted to be **PROBABLY DAMAGING** with a score of **1.000** (sensitivity: **0.00**; specificity: **1.00**)



|  |  |
| --- | --- |
| mutation t*@*sting |  |

|  |  |  |
| --- | --- | --- |
| Prediction | disease causing | **Model: *simple\_aae*, prob: 0.999999999999398**     ([explain](http://doro.charite.de/MutationTaster/info/documentation.html#bayes)) |
| **Summary** | * **amino acid sequence changed**
* **protein features (might be) affected**
* **splice site changes**
 | [hyperlink](http://www.mutationtaster.org/cgi-bin/MutationTaster/MutationTaster69.cgi?transcript_stable_id_text=ENST00000287295&position_be=1195&gene=AIFM1&transcript_stable_id_radio=ENST00000287295&sequence_type=CDS&new_base=A) |
| **analysed issue** | **analysis result** |
| name of alteration | no title |
| alteration (phys. location) | chr23:129270130C>T [show variant in all transcripts](http://www.mutationtaster.org/cgi-bin/MutationTaster/MT_ChrPos.cgi?chromosome=23&position=129270130&ref=C&alt=T)   [IGV](http://localhost:60151/goto?locus=chr23:129270129-129270131) |
| HGNC symbol | [AIFM1](http://www.genedistiller.org/cgi-bin/GeneDistiller_results.cgi?show_interaction=1&show_hpo=1&show_pathways=1&txid=9606&order=start_pos&show_generifs=1&show_mgd_phenotypes=1&show_GO=1&show_pfam=1&show_transcripts=1&show_omim=1&show_synonyms=1&show_interpro=1&show_paralogs=1&show_proteinfamilies=1&genesymbol=AIFM1) |
| Ensembl transcript ID | [ENST00000287295](http://grch37.ensembl.org/Homo_sapiens/transview?db=core;transcript=ENST00000287295) |
| Genbank transcript ID | [NM\_004208](http://www.ncbi.nlm.nih.gov/nuccore/NM_004208) |
| UniProt peptide | [O95831](http://www.uniprot.org/uniprot/O95831) |
| alteration type | single base exchange |
| alteration region | CDS |
| DNA changes | c.1195G>AcDNA.1426G>Ag.29732G>A |
| AA changes | G399S Score: 56 [explain score(s)](http://doro.charite.de/MutationTaster/info/documentation.html#grantham) |
| position(s) of altered AAif AA alteration in CDS | 399 |
| frameshift | no |
| known variant | Variant was neither found in ExAC nor 1000G.[Search ExAC.](http://exac.broadinstitute.org/variant/X-129270130-C-T) |