**Report 5 (NCBI resources activity)**

The protein of interest: Human Glycine--tRNA ligase

The gene name: GARS1

Th location on chromosome: Chr7: 30,594,735-30,634,033 (GRCh38.p12)

There are 10563 items in dbSNP, 159 items in dbVar, 227 items in ClinVar

Filtering by choosing pathogenic missense variants recorded in ClinVar, there are 13

variants.

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| --- | --- | --- | --- | --- | --- | --- |
| Variant ID | Mutation | Mutation type | PDB ID | Location in protein | Polyphen 2 score | Prediction about impact? |
| rs201358272 | p.Ser635Leu | Polar to hydrophobic | 2Q5H | Close to active site | 0.420 | BENIGN |
| rs137852643 | p.Gly294Arg | Hydrophobic to polar | 2Q5H | Close to binding site | 1.000 | PROBABLY DAMAGING |
| rs137852645 | p.Glu125Gly | Polar to hydrophobic | 2Q5H | Close to domain | 0.618 | POSSIBLY DAMAGING |
| rs137852644 | p.Leu183Pro | Hydrophobic to Hydrophobic | 2Q5H | Close to domain | 1.000 | PROBABLY DAMAGING |
| rs137852647 | p.Asp554Asn | Polar to Polar | 2Q5H | Close to active site | 0.048 | BENIGN |

PDB ID of the wild type protein: 2Q5H

Using iCn3D to show the structure of the mutation rs201358272

