RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · SYNONYMOUS

L822= — WFS1 Molecular Atlas Card

Variant type: Synonymous (silent)

Codon: position 822 (Leucine, L) — amino acid unchanged

Domain context: C-terminal ER-lumenal (calcium binding, calmodulin, chaperone)

SCHEMA CATEGORY: SILENT — SILENT — NO AMINO-ACID CHANGE

No amino-acid change (L822 is unchanged): the codon is altered but the protein sequence is identical to wild-type. No structural, stability or AlphaMissense effect applies. Synonymous variants are typically benign unless they affect splicing or regulatory elements; this one is not adjacent to an exon boundary.

CLINICAL EVIDENCE

- **Classification:** Conflicting classifications of pathogenicity
- **Review status:** criteria provided, conflicting classifications
- **Associated conditions:** WFS1-Related Spectrum Disorders; Autosomal dominant nonsyndromic hearing loss 6
- **cDNA change:** c.2466C>T
- **ClinVar accession:** VCV000745408
- **Last evaluated:** 2026/01/15 00:00
- **Submissions:** 1

Card generated by `wolfram-atlas-batch` (synonymous pipeline) on 2026-06-08T02:56:32.568399Z.

WFS1: UniProt O76024, AlphaFold v6. Synonymous variants carry no protein-structural effect.