

# V580E — WFS1 Molecular Atlas Card

**Variant type:** Missense

**Substitution:** Valine (V) → Glutamic acid (E) at position 580

**Domain context:** Transmembrane helix 9

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## ALPHAMISSENSE

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- **Pathogenicity score:** 0.3885
- **Class:** ambiguous

## ALPHAFOLD CONFIDENCE

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- **pLDDT at residue 580:** 86.75

> **DynaMut2  $\Delta\Delta G$ :** not yet computed for this variant — AlphaMissense + AlphaFold

> confidence shown above. Stability  $\Delta\Delta G$  and the wild-type/mutant structural

> comparison backfill behind this note.

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## CLINICAL EVIDENCE

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- **Classification:** Uncertain significance/Uncertain risk allele
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Inborn genetic diseases; Wolfram syndrome 1; Autosomal dominant nonsyndromic hearing loss 6; WFS1-Related Spectrum Disorders; Cataract 41; Type 2 diabetes mellitus; Wolfram-like syndrome
- **cDNA change:** c.1739T>A
- **ClinVar accession:** VCV000904987
- **Last evaluated:** 2025/10/21 00:00
- **Submissions:** 1

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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.633111Z.

AlphaMissense (Cheng et al. 2023) · AlphaFold model v6 · UniProt O76024.