

K193Q — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Lysine (K) → Glutamine (Q) at position 193

Domain context: N-terminal cytoplasmic (intrinsically disordered)

ALPHAMISSENSE

- **Pathogenicity score:** 0.1072
- **Class:** likely benign

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 193:** 79.19

> **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.

CLINICAL EVIDENCE

- **Classification:** Benign/Likely benign
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Monogenic diabetes; Wolfram syndrome 1; Wolfram-like syndrome; Autosomal dominant nonsyndromic hearing loss 6; Type 2 diabetes mellitus; Cataract 41; WFS1-Related Spectrum Disorders
- **cDNA change:** c.577A>C
- **ClinVar accession:** VCV000178654
- **Last evaluated:** 2026/02/01 00:00
- **Submissions:** 1

Card generated by wolfram-atlas-batch (missense AlphaMissense mint) on 2026-06-08T02:27:33.392287Z.

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