

K596Q — WFS1 Molecular Atlas Card

Variant type: Missense

Substitution: Lysine (K) → Glutamine (Q) at position 596

Domain context: Cytoplasmic loop 5 / pre-lumenal

ALPHAMISSENSE

- **Pathogenicity score:** 0.381
- **Class:** ambiguous

ALPHAFOLD CONFIDENCE

- **pLDDT at residue 596:** 62.06

> **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:** Uncertain significance
 - **Review status:** criteria provided, multiple submitters, no conflicts
 - **cDNA change:** c.1786A>C
 - **ClinVar accession:** VCV002976544
 - **Last evaluated:** 2025/09/04 00:00
 - **Submissions:** 1
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.647035Z.

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