

# R611H — WFS1 Molecular Atlas Card

**Variation type:** Missense

**Substitution:** Arginine (R) → Histidine (H) at position 611

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

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## ALPHAMISSENSE

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- **Pathogenicity score:** 0.0882
- **Class:** likely benign

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## ALPHAFOLD CONFIDENCE

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- **pLDDT at residue 611:** 58.38

> **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:** Benign/Likely benign
  - **Review status:** criteria provided, multiple submitters, no conflicts
  - **Associated conditions:** WFS1-Related Spectrum Disorders; Autosomal dominant nonsyndromic hearing loss 6; Wolfram syndrome 1
  - **cDNA change:** c.1832G>A
  - **ClinVar accession:** VCV000045442
  - **Last evaluated:** 2026/02/04 00:00
  - **Submissions:** 2
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Card generated by `wolfram-atlas-batch` (missense AlphaMissense mint) on 2026-06-08T02:27:33.659344Z.

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