RARERESEARCH.AI · WFS1 MOLECULAR ATLAS · SYNONYMOUS

# R139= — WFS1 Molecular Atlas Card

**Variant type:** Synonymous (silent)

**Codon:** position 139 (Arginine, R) — amino acid unchanged

**Domain context:** N-terminal cytoplasmic (intrinsically disordered)

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## SCHEMA CATEGORY: SILENT — SILENT — BUT NEAR AN EXON BOUNDARY (SPLICE EFFECT POSSIBLE)

No amino-acid change (R139 is unchanged), so there is no protein-level structural or stability effect. However, this codon sits within 3 residues of the exon junction near protein position 140 — close enough that the nucleotide change could perturb splicing. Worth a SpliceAI check (Wave 2); otherwise expected to be benign at the protein level.

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## CLINICAL EVIDENCE

- **Classification:** Likely benign
- **Review status:** criteria provided, multiple submitters, no conflicts
- **Associated conditions:** Wolfram syndrome 1
- **cDNA change:** c.417C>T
- **ClinVar accession:** VCV000682544
- **Last evaluated:** 2025/11/06 00:00
- **Submissions:** 1

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*Card generated by `wolfram-atlas-batch` (synonymous pipeline) on 2026-06-08T02:51:05.929783Z.*

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