

Mutation Effects

Mutation Impact Analysis

Wild Type:

M K L V F F A R G I L S D N Q K Y Position 234

Mutant:

M K L V F F A **R** G I L S D N Q K Y

R234W

Predicted Effects

Structural Impact

- $\Delta\Delta G$: +3.2 kcal/mol
- Destabilizing

Functional Impact

- Activity: 12% WT
- Loss of function

Pathogenicity:

Clinical Interpretation

90% (Likely Pathogenic)

Conservation Score: 0.98 (Highly Conserved)

Pathogenicity prediction

Disease association scoring

Stability changes

$\Delta\Delta G$ calculation

Function impact

Activity & binding changes

Evolutionary constraints

Conservation analysis

Clinical interpretation

Variant classification