NEIL1 Q282TER does not translocate to the nucleus

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Introduction

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

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Literature references

Fabregat, A., Sidiropoulos, K., Viteri, G., Forner, O., Marin-Garcia, P., Arnau, V. et al. (2017). Reactome pathway analysis: a high-performance in-memory approach. *BMC bioinformatics, 18*, 142. ↗

Sidiropoulos, K., Viteri, G., Sevilla, C., Jupe, S., Webber, M., Orlic-Milacic, M. et al. (2017). Reactome enhanced pathway visualization. *Bioinformatics, 33*, 3461-3467. ↗

Fabregat, A., Jupe, S., Matthews, L., Sidiropoulos, K., Gillespie, M., Garapati, P. et al. (2018). The Reactome Pathway Knowledgebase. *Nucleic Acids Res, 46*, D649-D655. ↗

Fabregat, A., Korninger, F., Viteri, G., Sidiropoulos, K., Marin-Garcia, P., Ping, P. et al. (2018). Reactome graph database: Efficient access to complex pathway data. *PLoS computational biology, 14*, e1005968. ↗

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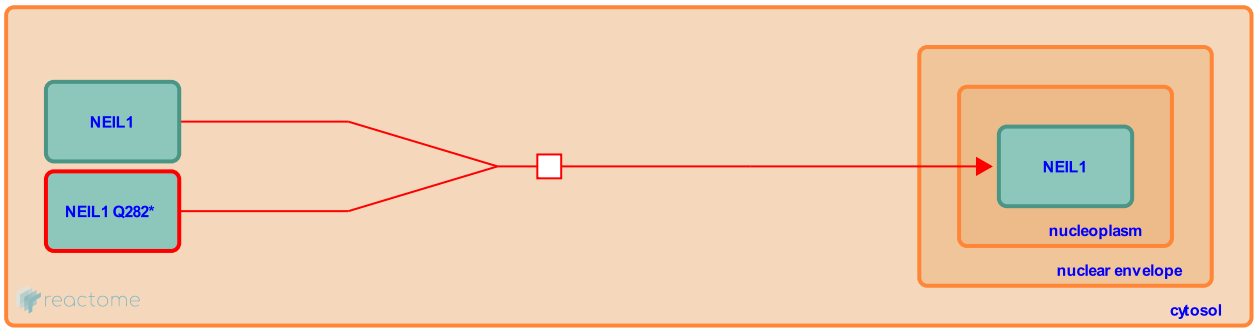
Stable identifier: R-HSA-9629917

Type: transition

Compartments: cytosol

Diseases: cancer



A rare NEIL1 variant reported in the Japanese population results in a truncated NEIL1 protein, NEIL1 Q282TER, which lacks the putative nuclear localization signal (NLS). NEIL1 Q282TER localizes to the cytosol and, due to this mislocalization, is not able to access damaged DNA substrates. Several NEIL truncation mutations that remove the NLS have been reported in different cancer types and correlate with increased mutational loads, but they have not been functionally studied. NEIL1 Q282TER has not yet been associated with any specific cancer type or cancer predisposition, but is the only functionally studied NEIL1 truncation mutant that lacks the NLS (Shinmura et al. 2015).

Literature references

Shinmura, K., Inoue, Y., Nakamura, S., Goto, M., Sugimura, H., Kato, H. et al. (2015). NEIL1 p.Gln282Stop variant is predominantly localized in the cytoplasm and exhibits reduced activity in suppressing mutations. *Gene, 571*, 33-42. ↗

Editions

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