Defective NEIL1 variants do not cleave Tg

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03/05/2024

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.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

Literature references

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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. ↗

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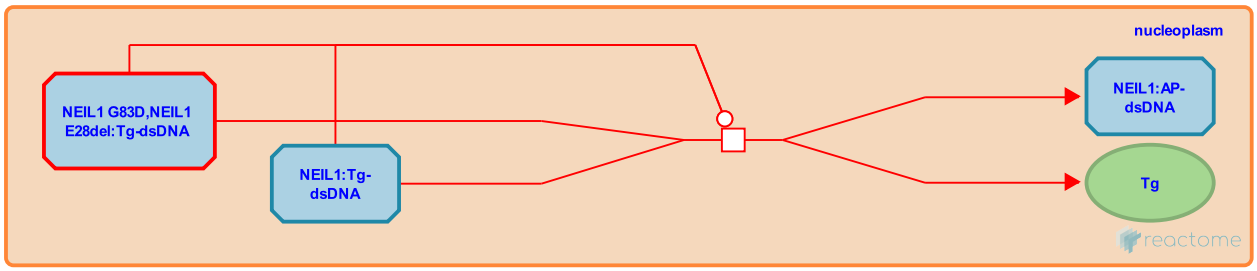
Stable identifier: R-HSA-9629166

Type: transition

Compartments: nucleoplasm

Diseases: cancer



NEIL1 G83D variant is a low frequency polymorphism, estimated to occur in ~1% of US population (Roy et al. 2007), and is associated with primary sclerosing cholangitis and cholangiocarcinoma (Forsbring et al. 2009). NEIL1 G83D does not cleave thymine glycol (Tg) from damaged DNA (Prakash et al. 2014).
NEIL1 E28del, an in-frame deletion variant of NEIL1 reported in gastric (stomach) cancer, where glutamate at position 28 is deleted, does not cleave Tg from damaged DNA (Shinmura et al. 2008).

Literature references

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Editions

2018-11-27	Authored	Orlic-Milacic, M.
2019-01-03	Reviewed	Sampath, H.
2019-01-05	Edited	Orlic-Milacic, M.