NEIL3 D132V does not cleave 5-guanidino-hydantoin (Gh)

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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. ↗

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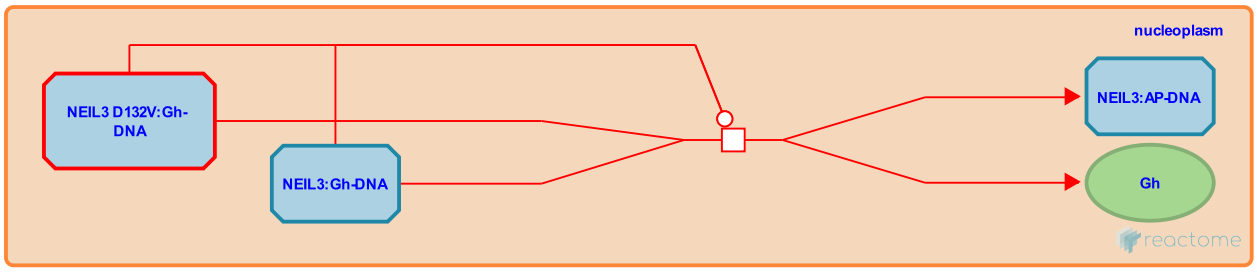
Stable identifier: R-HSA-9629245

Type: transition

Compartments: nucleoplasm

Diseases: autoimmune hypersensitivity disease



NEIL3 D132V is a NEIL3 disease variant caused by a missense mutation that results in the substitution of aspartic acid residue (D) at position 132 to valine (V). NEIL3 D132V is unable to cleave 5-guanidinohydantoin (Gh) from oxidatively damaged DNA. Individuals harbouring a homozygous NEIL3 D132V mutation are predisposed to development of autoimmune diseases (Massaad et al. 2016). Primary fibroblasts from a patient with a NEIL3 D132V homozygous mutation show an increase in telomere loss compared to control wild type fibroblasts derived from the patient's healthy sibling (Zhou et al. 2017).

Literature references

Chou, J., Megarbane, A., Ohsumi, TK., Jabara, H., Geha, RS., Al-Herz, W. et al. (2016). Deficiency of base excision repair enzyme NEIL3 drives increased predisposition to autoimmunity. *J. Clin. Invest., 126*, 4219-4236.

Editions

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