p14ARF mutant 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. ↗

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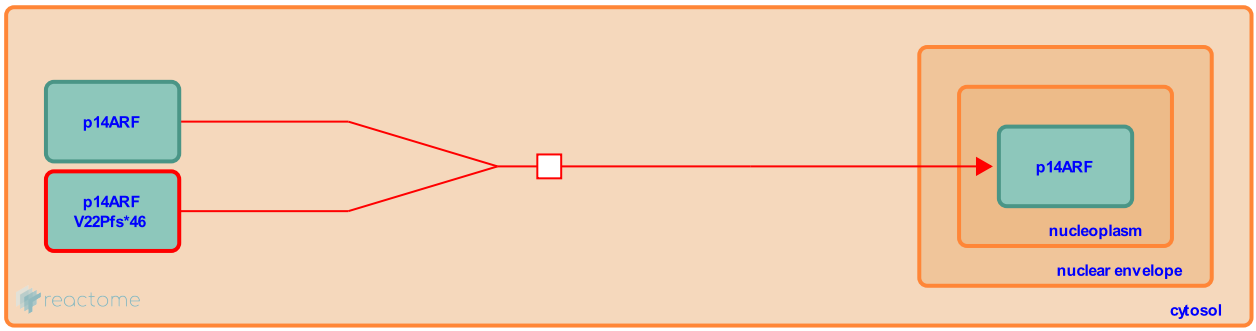
Stable identifier: R-HSA-9646295

Type: transition

Compartments: cytosol

Diseases: cancer



A germline mutation affecting exon 1beta of the CDKN2A locus is associated with a familial melanoma syndrome. The mutation represents an insertion of 16 nucleotides (CGGCCCGCCGCGAGTG) between coding bases 60 and 61 in exon 1beta. This insertion results in a frameshift, starting at arginine codon at position 21 of p14ARF, with the first amino acid changed being valine at position 22, and ending with a premature stop codon at position 67. The mutant protein p14ARF V22Pfs*46 (p14ARF 60ins16) is unable to translocate to the nucleus and stabilize TP53 (Rizos, Puig et al. 2001).

Literature references

Puig, S., Milà, M., Darmanian, AP., Badenas, C., Kefford, RF., Rizos, H. et al. (2001). A melanoma-associated germline mutation in exon 1beta inactivates p14ARF. *Oncogene, 20*, 5543-7. ↗

Editions

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