HNF1A-dependent synthesis of HNF4A

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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

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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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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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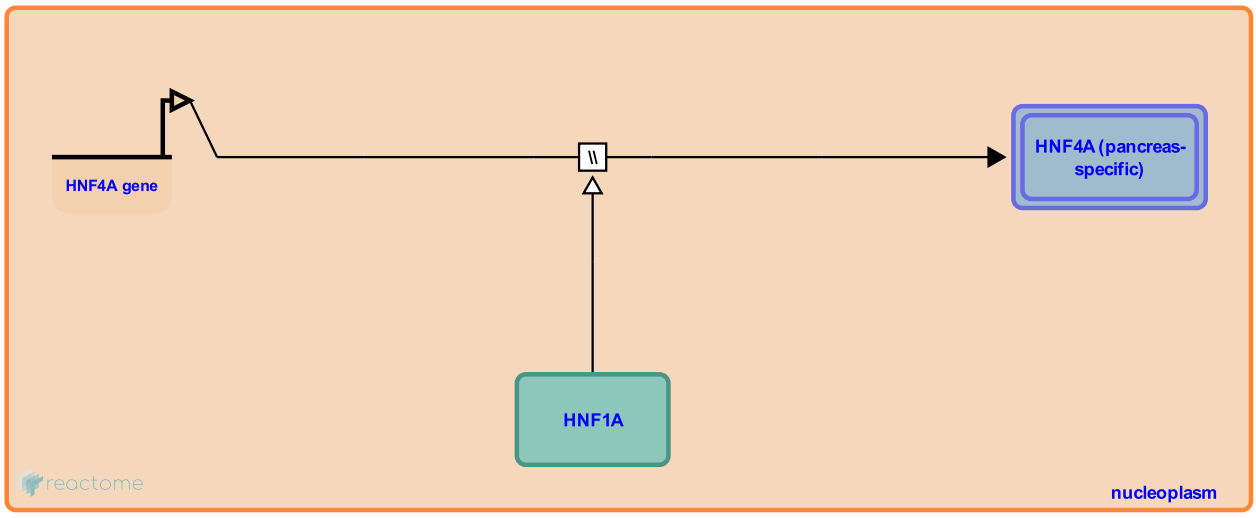
Stable identifier: R-HSA-211466

Type: omitted

Compartments: nucleoplasm

Inferred from: HNF1a regulates Hnf4a (Mus musculus)



The HNF4A gene is transcribed from either of two promoters, P1 and P2, the resulting mRNA is translated, and the protein products localize in the nucleoplasm. Transcription is positively regulated by HNF1A. Many of the molecular details of these events have not been studied experimentally in humans, but are inferred from mouse model systems (Boj et al. 2001). Transcription in mouse and human pancreatic beta cells is P2-dependent and in humans yields three isoforms of mature HNF4A protein. A point mutation in the human P2 genomic DNA sequence is associated with MODY (maturity onset diabetes of the young), consistent with the hypothesis that P2-mediated transcription is essential for HNF4A expression and normal beta cell function (Hansen et al. 2002).

Editions

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