Defective SLC24A5 causes oculocutaneous albinism 6 (OCA6)

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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**


Reactome database release: 72

This document contains 1 pathway and 1 reaction (see Table of Contents)
Defective SLC24A5 causes oculocutaneous albinism 6 (OCA6)

Stable identifier: R-HSA-5619036

Diseases: oculocutaneous albinism

Five members of the NCKX (SLC24) family are all able to exchange one Ca2+ and one K+ for four Na+. SLC24A5 (NCKX5, located on the trans-Golgi membrane) is the predominant K+-dependent Na+/Ca2+ exchanger in melanocytes and is one of a handful of genes thought to play a role in determining human skin colour (Wilson et al. 2013). Defects in SLC24A5 can cause oculocutaneous albinism 6 (OCA6; MIM:113750), a disorder characterised by a reduction or complete loss of melanin in the skin, hair and eyes. Patients with this condition show accompanied eye symptoms (Kamaraj & Purohit 2014, Morice-Picard et al. 2014).

Literature references


Editions

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Defective SLC24A5 does not exchange cytosolic 4Na+ for Golgi luminal Ca2+, K+

Location: Defective SLC24A5 causes oculocutaneous albinism 6 (OCA6)

Stable identifier: R-HSA-5626356