Defective SLC34A3 causes Hereditary hypophosphatemic rickets with hypercalciuria (HHRH)

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09/09/2020
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: 73

This document contains 1 pathway and 1 reaction (see Table of Contents)
Defective SLC34A3 causes Hereditary hypophosphatemic rickets with hypercalciuria (HHRH)

Stable identifier: R-HSA-5619097

Diseases: X-linked hypophosphatemia

SLC34A3 is almost exclusively expressed at the apical membranes of kidney proximal tubules and encodes a Na+/Pi cotransporter. It cotransports 2 Na+ ions with every phosphate (Pi) (electroneutral transport). Defects in SLC34A3 are the cause of hereditary hypophosphatemic rickets with hypercalciuria (HHRH; MIM:241530), an autosomal recessive form of hypophosphatemia characterised by reduced renal phosphate reabsorption and rickets (Bergwitz et al. 2006, Segawa et al. 2013, Forster et al. 2013).

Literature references


Editions

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Defective SLC34A3 does not cotransport Pi, 2Na+↗

**Location:** Defective SLC34A3 causes Hereditary hypophosphatemic rickets with hypercalciuria (HHRH)

**Stable identifier:** R-HSA-5651971

**Type:** transition

**Compartments:** extracellular region, plasma membrane

**Diseases:** X-linked hypophosphatemia

SLC34A3 is almost exclusively expressed at the apical membranes of kidney proximal tubules and encodes a Na+/Pi cotransporter. It cotransports 2 Na+ ions with every phosphate (Pi) (electroneutral transport). Defects in SLC34A3 are the cause of hereditary hypophosphatemic rickets with hypercalciuria (HHRH; MIM:241530), an autosomal recessive form of hypophosphatemia characterised by reduced renal phosphate reabsorption and rickets. Mutations causing HHRH include G191R, P303Rfs*40, C77Afs*75, R468W and G196R (Chi et al. 2014, Lorenz-Depiereux et al. 2006, Bergwitz et al. 2006).

**Literature references**


**Editions**

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