Defective SLC34A1 causes hypophosphatemic nephrolithiasis/osteoporosis 1 (NPHLOP1)

Broer, S., Jassal, B.
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: 68

This document contains 1 pathway and 1 reaction (see Table of Contents)
Defective SLC34A1 causes hypophosphatemic nephrolithiasis/osteoporosis 1 (NPHLOP1)

Stable identifier: R-HSA-5619040

Diseases: nephrolithiasis, renal tubular transport disease, hypophosphatemia

SLC34A1 and 2 encode Na+/Pi cotransporters, which cotransport divalent phosphate (PO4(2-), Pi) with 3 Na+ ions. SLC34A1 is an important Pi transporter mainly expressed in renal proximal tubules where it plays a major role in Pi homeostasis. Defects in SLC34A1 are the cause of hypophosphatemic nephrolithiasis/osteoporosis type 1 (NPHLOP1; MIM:612286), disease characterised by decreased renal phosphate absorption, hypophosphatemia, hyperphosphaturia, hypercalciuria, nephrolithiasis and implicated in the formation of renal calcium stones and/or bone demineralisation (Prie et al. 2002, Prie et al. 2004, Choi 2008, Boskey et al. 2013, Forster et al. 2013).

Literature references


Editions

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Defective SLC34A1 does not cotransport Pi, 3Na+

Location: Defective SLC34A1 causes hypophosphatemic nephrolithiasis/osteoporosis 1 (NPHLOP1)

Stable identifier: R-HSA-5651685

Type: transition

Compartments: plasma membrane, extracellular region

Diseases: nephrolithiasis, hypophosphatemia, renal tubular transport disease

SLC34A1 and 2 encode Na+/Pi cotransporters, which cotransport divalent phosphate (PO4(2-), Pi) with 3 Na+ ions. SLC34A1 is an important Pi transporter mainly expressed in renal proximal tubules where it plays a major role in Pi homeostasis. Defects in SLC34A1 are the cause of hypophosphatemic nephrolithiasis/osteoporosis type 1 (NPHLOP1; MIM:612286), disease characterised by decreased renal phosphate absorption and implicated in the formation of renal calcium stones and/or bone demineralisation. Mutations causing NPHLOP1 are A48F and V147M (Prie et al. 2002).

Literature references


Editions

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Table of Contents

Introduction 1
Defective SLC34A1 causes hypophosphatemic nephrolithiasis/osteoporosis 1 (NPHLOP1) 2
Defective SLC34A1 does not cotransport Pi, 3Na+ 3
Table of Contents 4