# Resource Summary Report

Generated by nidm-terms on Jul 29, 2026

## Lowes Syndrome Mutation Database

RRID:SCR_002907
Type: Tool

### Proper Citation

Lowes Syndrome Mutation Database (RRID:SCR_002907)

### Resource Information

**URL:** http://www.ncbi.nlm.nih.gov/lovd/home.php?select_db=OCRL

**Proper Citation:** Lowes Syndrome Mutation Database (RRID:SCR_002907)

**Description:** The Lowe Syndrome Mutation Database is now being maintained by the National Center for Biotechnology Information (NCBI) at the National Institutes of Health. A database of mutations causing Lowe syndrome. Information on new mutations may be submitted online. Lowe oculocerebrorenal syndrome is an X-linked disorder caused by mutations in the OCRL1 gene, which encodes a 105-kDa Golgi protein with phosphatidylinositol (4,5) bisphosphate 5-phosphatase activity. genetics

**Synonyms:** Lowe Syndrome Mutation Database

**Resource Type:** service resource, data or information resource, data repository, database, storage service resource

**Keywords:** mutation

**Related Condition:** Lowe syndrome

**Funding:** NHGRI

**Availability:** THIS RESOURCE IS NO LONGER IN SERVICE

**Resource Name:** Lowes Syndrome Mutation Database

**Resource ID:** SCR_002907

**Alternate IDs:** nif-0000-00155

**Old URLs:** http://research.nhgri.nih.gov/lowe/

**License:** The Lowe Syndrome Mutation Database is now being maintained by the National Center for Biotechnology Information (NCBI) at the National Institutes of Health,

The community can contribute to this resource

**Record Creation Time:** 20220129T080216+0000

**Record Last Update:** 20260729T044041+0000

## Ratings and Alerts

No rating or validation information has been found for Lowes Syndrome Mutation Database.

No alerts have been found for Lowes Syndrome Mutation Database.

## Data and Source Information

**Source:** SciCrunch Registry

## Usage and Citation Metrics

We found 1 mentions in open access literature.

**Listed below are recent publications.** The full list is available at nidm-terms .

Noakes CJ, et al. (2011) The PH domain proteins IPIP27A and B link OCRL1 to receptor recycling in the endocytic pathway. Molecular biology of the cell, 22(5), 606.