

Resource Summary Report

Generated by SPARC SAWG on Sep 15, 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: data or information resource, data repository, database, service resource, 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: 20260912T195548+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 [SPARC SAWG](#) .

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.