

# Resource Summary Report

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## X-linked Adrenoleukodystrophy Database

RRID:SCR\_002548

Type: Tool

### Proper Citation

X-linked Adrenoleukodystrophy Database (RRID:SCR\_002548)

### Resource Information

**URL:** <http://www.x-ald.nl/>

**Proper Citation:** X-linked Adrenoleukodystrophy Database (RRID:SCR\_002548)

**Description:** X-ALD database initiated July 1999 by Hugo W. Moser, M.D. and Stephan Kemp, Ph.D. The primary aims of the database are: to catalogue and facilitate the analysis of X-ALD mutations; to provide background information on X-ALD; to provide links to X-ALD patient organizations; and to help with contacting and finding (local) X-ALD health care professionals. The purpose of the X-linked adrenoleukodystrophy database is to provide general educational information about X-ALD.

**Resource Type:** database, portal, data or information resource, topical portal, disease-related portal

**Keywords:** inherited disorder, central nervous system, mutation database, gene database, x ald genomics, FASEB list

**Related Condition:** X-linked Adrenoleukodystrophy

**Funding:** Netherlands X-ALD Patient Organization

**Resource Name:** X-linked Adrenoleukodystrophy Database

**Resource ID:** SCR\_002548

**Alternate IDs:** nif-0000-21424

**Record Creation Time:** 20220129T080214+0000

**Record Last Update:** 20250409T060207+0000

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## Ratings and Alerts

No rating or validation information has been found for X-linked Adrenoleukodystrophy Database.

No alerts have been found for X-linked Adrenoleukodystrophy Database.

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## Data and Source Information

**Source:** [SciCrunch Registry](#)

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## Usage and Citation Metrics

We found 46 mentions in open access literature.

**Listed below are recent publications.** The full list is available at [RRID](#).

Takegami N, et al. (2024) Adrenomyeloneuropathy with Later Development of Cerebral Form Caused by a Hemizygous Splice-site Variant in ABCD1. *Internal medicine (Tokyo, Japan)*, 63(7), 999.

Li J, et al. (2019) Clinical, neuroimaging, biochemical, and genetic features in six Chinese patients with Adrenomyeloneuropathy. *BMC neurology*, 19(1), 227.

Chen YH, et al. (2017) Unmasking adrenoleukodystrophy in a cohort of cerebellar ataxia. *PloS one*, 12(5), e0177296.

Curiel J, et al. (2017) X-linked adrenoleukodystrophy in a chimpanzee due to an ABCD1 mutation reported in multiple unrelated humans. *Molecular genetics and metabolism*, 122(3), 130.

Yan F, et al. (2017) S149R, a novel mutation in the ABCD1 gene causing X-linked adrenoleukodystrophy. *Oncotarget*, 8(50), 87529.

van de Beek MC, et al. (2017) Lipid-induced endoplasmic reticulum stress in X-linked adrenoleukodystrophy. *Biochimica et biophysica acta. Molecular basis of disease*, 1863(9), 2255.

Wanders RJ, et al. (2017) "Role of peroxisomes in human lipid metabolism and its importance for neurological development". *Neuroscience letters*, 637, 11.

Andreoletti P, et al. (2017) Predictive Structure and Topology of Peroxisomal ATP-Binding Cassette (ABC) Transporters. *International journal of molecular sciences*, 18(7).

Mehrpour M, et al. (2016) An ABCD1 Mutation (c.253dupC) Caused Diverse Phenotypes of Adrenoleukodystrophy in an Iranian Consanguineous Pedigree. *Journal of molecular and genetic medicine : an international journal of biomedical research*, 10(2).

Horn MA, et al. (2016) Mild phenotype in an adult male with X-linked adrenoleukodystrophy - case report. *Clinical case reports*, 4(2), 177.

Ferdinandusse S, et al. (2016) The important role of biochemical and functional studies in the diagnostics of peroxisomal disorders. *Journal of inherited metabolic disease*, 39(4), 531.

Kawaguchi K, et al. (2016) ABC Transporter Subfamily D: Distinct Differences in Behavior between ABCD1-3 and ABCD4 in Subcellular Localization, Function, and Human Disease. *BioMed research international*, 2016, 6786245.

Baarine M, et al. (2015) ABCD1 deletion-induced mitochondrial dysfunction is corrected by SAHA: implication for adrenoleukodystrophy. *Journal of neurochemistry*, 133(3), 380.

Wiesinger C, et al. (2015) The genetic landscape of X-linked adrenoleukodystrophy: inheritance, mutations, modifier genes, and diagnosis. *The application of clinical genetics*, 8, 109.

Karkar A, et al. (2015) A novel mutation in the ABCD1 gene of a Moroccan patient with X-linked adrenoleukodystrophy: case report. *BMC neurology*, 15, 244.

Kallabi F, et al. (2015) Splicing defects in ABCD1 gene leading to both exon skipping and partial intron retention in X-linked adrenoleukodystrophy Tunisian patient. *Neuroscience research*, 97, 7.

Kang JW, et al. (2014) Isolated cerebellar variant of adrenoleukodystrophy with a de novo adenosine triphosphate-binding cassette D1 (ABCD1) gene mutation. *Yonsei medical journal*, 55(4), 1157.

Jwa HJ, et al. (2014) A Korean boy with atypical X-linked adrenoleukodystrophy confirmed by an unpublished mutation of ABCD1. *Korean journal of pediatrics*, 57(9), 416.

Chuang CY, et al. (2014) Involvement of the carboxyl-terminal region of the yeast peroxisomal half ABC transporter Pxa2p in its interaction with Pxa1p and in transporter function. *PloS one*, 9(8), e104892.

Park HJ, et al. (2014) Clinical and genetic aspects in twelve Korean patients with adrenomyeloneuropathy. *Yonsei medical journal*, 55(3), 676.