

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: data or information resource, database, disease-related portal, portal, topical 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: 20260905T132450+0000

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.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 52 mentions in open access literature.

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

Fu H, et al. (2025) A Novel Missense Variant of the ABCD1 Gene in X-Linked Adrenoleukodystrophy in Chinese Family. *Molecular genetics & genomic medicine*, 13(10), e70148.

Zhang Y, et al. (2025) Genotypic and Phenotypic Characteristics of Pediatric X-Adrenoleukodystrophy in a Chinese Cohort. *Neuropsychiatric disease and treatment*, 21, 677.

Mansouri M, et al. (2025) X-Linked Adrenoleukodystrophy in a Moroccan Patient: Genetic Diagnosis Leads to Presymptomatic Testing and Family Counseling. *Balkan journal of medical genetics : BJMG*, 28(1), 20250002.

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.

He R, et al. (2023) Novel mutations in the ABCD1 gene caused adrenomyeloneuropathy in the Chinese population. *Frontiers in neurology*, 14, 1126729.

Dong B, et al. (2022) Identification of Two Novel Mutations of ABCD1 Gene in Pedigrees with X-Linked Adrenoleukodystrophy and Review of the Literature. *International journal of endocrinology*, 2022, 5479781.

Campopiano R, et al. (2021) A Large Family with p.Arg554His Mutation in ABCD1: Clinical Features and Genotype/Phenotype Correlation in Female Carriers. *Genes*, 12(5).

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.

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.