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

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: 20250421T053332+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 46 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF.

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.

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.

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.

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.