Allan M Lund

List of Publications by Year
in descending order

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77	2,045	230014	325983
papers	citations	27 h-index	40 g-index

84	84	84	2367
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Pyridoxine or pyridoxalâ€5â€phosphate treatment for seizures in glycosylphosphatidylinositol deficiency: A cohort study. Developmental Medicine and Child Neurology, 2022, 64, 789-798.	1.1	6
2	The effect of casein glycomacropeptide versus free synthetic amino acids for early treatment of phenylketonuria in a mice model. PLoS ONE, 2022, 17, e0261150.	1.1	3
3	The impact of rifaximin on inflammation and metabolism in alcoholic hepatitis: A randomized clinical trial. PLoS ONE, 2022, 17, e0264278.	1.1	14
4	Influence of early identification and therapy on longâ€term outcomes in earlyâ€onset <scp>MTHFR</scp> deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 848-861.	1.7	7
5	Cystathionine Î²â€synthase deficiency in the <scp>Eâ€HOD registryâ€part</scp> I: pyridoxine responsiveness as a determinant of biochemical and clinical phenotype at diagnosis. Journal of Inherited Metabolic Disease, 2021, 44, 677-692.	1.7	20
6	Use of Molecular Genetic Analyses in Danish Routine Newborn Screening. International Journal of Neonatal Screening, 2021, 7, 50.	1.2	12
7	Long-term safety and outcomes in hereditary tyrosinaemia type 1 with nitisinone treatment: a 15-year non-interventional, multicentre study. Lancet Diabetes and Endocrinology,the, 2021, 9, 427-435.	5.5	19
8	Increased risk of sudden death in untreated primary carnitine deficiency. Journal of Inherited Metabolic Disease, 2020, 43, 290-296.	1.7	21
9	Cystic fibrosis newborn screening in Denmark: Experience from the first 2 years. Pediatric Pulmonology, 2020, 55, 549-555.	1.0	26
10	The SPARKLE registry: protocol for an international prospective cohort study in patients with alpha-mannosidosis. Orphanet Journal of Rare Diseases, 2020, 15, 271.	1.2	4
11	Use of the Bruininks-Oseretsky test of motor proficiency (BOT-2) to assess efficacy of velmanase alfa as enzyme therapy for alpha-mannosidosis. Molecular Genetics and Metabolism Reports, 2020, 23, 100586.	0.4	6
12	Impaired lipolysis in propionic acidemia: A new metabolic myopathy?. JIMD Reports, 2020, 53, 16-21.	0.7	10
13	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	1.1	14
14	Impaired Fat Oxidation During Exercise in Long-Chain Acyl-CoA Dehydrogenase Deficiency Patients and Effect of IV-Glucose. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3610-3613.	1.8	1
15	Including Classical Galactosaemia in the Expanded Newborn Screening Panel Using Tandem Mass Spectrometry for Galactose-1-Phosphate. International Journal of Neonatal Screening, 2019, 5, 19.	1.2	7
16	Real-World Outcomes with Lomitapide Use in Paediatric Patients with Homozygous Familial Hypercholesterolaemia. Advances in Therapy, 2019, 36, 1786-1811.	1.3	35
17	Questions about a vegan diet should be included in differential diagnostics of neurologically abnormal infants with failure to thrive. Acta Paediatrica, International Journal of Paediatrics, 2019, 108, 1377-1379.	0.7	2
18	Functional disruption of pyrimidine nucleoside transporter CNT1 results in a novel inborn error of metabolism with high excretion of uridine and cytidine. Journal of Inherited Metabolic Disease, 2019, 42, 494-500.	1.7	6

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19	Recognition of alpha-mannosidosis in paediatric and adult patients: Presentation of a diagnostic algorithm from an international working group. Molecular Genetics and Metabolism, 2019, 126, 470-474.	0.5	20
20	Evaluation of dietary treatment and amino acid supplementation in organic acidurias and ureaâ€cycle disorders: On the basis of information from a European multicenter registry. Journal of Inherited Metabolic Disease, 2019, 42, 1162-1175.	1.7	30
21	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	1.7	37
22	Decreased plasma l-arginine levels in organic acidurias (MMA and PA) and decreased plasma branched-chain amino acid levels in urea cycle disorders as a potential cause of growth retardation: Options for treatment. Molecular Genetics and Metabolism, 2019, 126, 397-405.	0.5	26
23	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	1.7	53
24	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	1.7	35
25	A splice-site variant in the lncRNA gene cosegregates in the large Volkmann cataract family. Molecular Vision, 2019, 25, 1-11.	1.1	1
26	The impact of consanguinity on the frequency of inborn errors of metabolism. Molecular Genetics and Metabolism Reports, 2018, 15, 6-10.	0.4	24
27	Comprehensive longâ€term efficacy and safety of recombinant human alphaâ€mannosidase (velmanase) Tj ETQq1 1 0.784314 rgBT /Ov 1225-1233.	1.7	35
28	Hyperglucagonemia correlates with plasma levels of non-branched-chain amino acids in patients with liver disease independent of type 2 diabetes. American Journal of Physiology - Renal Physiology, 2018, 314, G91-G96.	1.6	61
29	Simultaneous quantification of succinylacetone and nitisinone for therapeutic drug monitoring in the treatment of Tyrosinemia type 1. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2018, 1072, 259-266.	1.2	5
30	Health Related Quality of Life, Disability, and Pain in Alpha Mannosidosis. FIRE Forum for International Research in Education, 2018, 6, 232640981879685.	0.7	4
31	L-Carnitine Improves Skeletal Muscle Fat Oxidation in Primary Carnitine Deficiency. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 4580-4588.	1.8	15
32	International working group identifies need for newborn screening for mucopolysaccharidosis type I but states that existing hurdles must be overcome. Acta Paediatrica, International Journal of Paediatrics, 2018, 107, 2059-2065.	0.7	10
33	Compound heterozygous mutations in two different domains of ALDH18A1 do not affect the amino acid levels in a patient with hereditary spastic paraplegia. Neurogenetics, 2018, 19, 145-149.	0.7	11
34	Efficacy and safety of Velmanase alfa in the treatment of patients with alphaâ€mannosidosis: results from the core and extension phase analysis of a phase III multicentre, doubleâ€blind, randomised, placeboâ€controlled trial. Journal of Inherited Metabolic Disease, 2018, 41, 1215-1223.	1.7	34
35	Easyâ€toâ€use algorithm would provide faster diagnoses for mucopolysaccharidosis type I and enable patients to receive earlier treatment. Acta Paediatrica, International Journal of Paediatrics, 2018, 107, 1402-1408.	0.7	11
36	Comparison of Glycomacropeptide with Phenylalanine Free-Synthetic Amino Acids in Test Meals to PKU Patients: No Significant Differences in Biomarkers, Including Plasma Phe Levels. Journal of Nutrition and Metabolism, 2018, 2018, 1-11.	0.7	14

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37	Osteogenesis imperfecta and the teeth, eyes, and earsâ€"a study of non-skeletal phenotypes in adults. Osteoporosis International, 2018, 29, 2781-2789.	1.3	29
38	Cryopreservation of ovarian tissue may be considered in young girls with galactosemia. Journal of Assisted Reproduction and Genetics, 2018, 35, 1209-1217.	1.2	28
39	Extreme neonatal hyperbilirubinemia, acute bilirubin encephalopathy, and kernicterus spectrum disorder in children with galactosemia. Pediatric Research, 2018, 84, 228-232.	1.1	11
40	Prevalence of Mucopolysaccharidosis Types I, II, and VI in the Pediatric and Adult Population with Carpal Tunnel Syndrome (CTS). Retrospective and Prospective Analysis of Patients Treated for CTS. JIMD Reports, 2017, 36, 29-33.	0.7	5
41	Primary Carnitine Deficiency: Is Foetal Development Affected and Can Newborn Screening Be Improved?. JIMD Reports, 2017, 36, 35-40.	0.7	11
42	The D313Y variant in the GLA gene â€“ no evidence of a pathogenic role in Fabry disease. Scandinavian Journal of Clinical and Laboratory Investigation, 2017, 77, 617-621.	0.6	19
43	Neonatal Screening for Primary Carnitine Deficiency: Lessons Learned from the Faroe Islands. International Journal of Neonatal Screening, 2017, 3, 1.	1.2	10
44	Splenomegaly â€“ Diagnostic validity, work-up, and underlying causes. PLoS ONE, 2017, 12, e0186674.	1.1	29
45	Impact of age at onset and newborn screening on outcome in organic acidurias. Journal of Inherited Metabolic Disease, 2016, 39, 341-353.	1.7	60
46	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. Journal of Inherited Metabolic Disease, 2016, 39, 661-672.	1.7	52
47	Skeletal phenotypes in adult patients with osteogenesis imperfectaâ€"correlations with COL1A1/COL1A2 genotype and collagen structure. Osteoporosis International, 2016, 27, 3331-3341.	1.3	30
48	Alphaâ€mannosidosis: characterization of CNS pathology and correlation between CNS pathology and cognitive function. Clinical Genetics, 2016, 89, 489-494.	1.0	11
49	Defective hepatic bicarbonate production due to carbonic anhydrase VA deficiency leads to early-onset life-threatening metabolic crisis. Genetics in Medicine, 2016, 18, 991-1000.	1.1	31
50	Cognitive profile and activities of daily living: 35 patients with alphaâ€mannosidosis. Journal of Inherited Metabolic Disease, 2015, 38, 1119-1127.	1.7	16
51	Alpha-mannosidosis: correlation between phenotype, genotype and mutant MAN2B1 subcellular localisation. Orphanet Journal of Rare Diseases, 2015, 10, 70.	1.2	35
52	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. Journal of Inherited Metabolic Disease, 2015, 38, 1041-1057.	1.7	186
53	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074.	1.7	175
54	Abnormal Newborn Screening in a Healthy Infant of a Mother with Undiagnosed Medium-Chain Acyl-CoA Dehydrogenase Deficiency. JIMD Reports, 2015, 23, 67-70.	0.7	7

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55	Carnitine levels in 26,462 individuals from the nationwide screening program for primary carnitine deficiency in the Faroe Islands. Journal of Inherited Metabolic Disease, 2014, 37, 215-222.	1.7	64
56	Primary Carnitine deficiency in the Faroe Islands: health and cardiac status in 76 adult patients diagnosed by screening. Journal of Inherited Metabolic Disease, 2014, 37, 223-230.	1.7	31
57	Residual OCTN2 transporter activity, carnitine levels and symptoms correlate in patients with primary carnitine deficiency. Molecular Genetics and Metabolism Reports, 2014, 1, 241-248.	0.4	20
58	Alpha-mannosidosis - a review of genetic, clinical findings and options of treatment. Pediatric Endocrinology Reviews, 2014, 12 Suppl 1, 185-91.	1.2	15
59	Enzyme replacement therapy for alphaâ€mannosidosis: 12 months followâ€up of a single centre, randomised, multiple dose study. Journal of Inherited Metabolic Disease, 2013, 36, 1015-1024.	1.7	33
60	Primary carnitine deficiency and pivalic acid exposure causing encephalopathy and fatal cardiac events. Journal of Inherited Metabolic Disease, 2013, 36, 35-41.	1.7	46
61	Patients With Medium-Chain Acylâ€“Coenzyme A Dehydrogenase Deficiency Have Impaired Oxidation of Fat During Exercise but No Effect of<scp>l</scp>-Carnitine Supplementation. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 1667-1675.	1.8	24
62	Biochemical screening of 504,049 newborns in Denmark, the Faroe Islands and Greenland â€" Experience and development of a routine program for expanded newborn screening. Molecular Genetics and Metabolism, 2012, 107, 281-293.	0.5	55
63	MCAD deficiency in Denmark. Molecular Genetics and Metabolism, 2012, 106, 175-188.	0.5	33
64	Normal Levels of Plasma Free Carnitine and Acylcarnitines in Follow-Up Samples from a Presymptomatic Case of Carnitine Palmitoyl Transferase 1 (CPT1) Deficiency Detected Through Newborn Screening in Denmark. JIMD Reports, 2011, 3, 11-15.	0.7	9
65	Clinical and biochemical monitoring of patients with fatty acid oxidation disorders. Journal of Inherited Metabolic Disease, 2010, 33, 495-500.	1.7	25
66	Carnitine transporter and holocarboxylase synthetase deficiencies in The Faroe Islands. Journal of Inherited Metabolic Disease, 2007, 30, 341-349.	1.7	43
67	Variations in IBD (ACAD8) in Children with Elevated C4-Carnitine Detected by Tandem Mass Spectrometry Newborn Screening. Pediatric Research, 2006, 60, 315-320.	1.1	32
68	Anthropometry of patients with osteogenesis imperfecta. Archives of Disease in Childhood, 1999, 80, 524-528.	1.0	55
69	Osteogenesis imperfecta: Mosaicism and refinement of the genotype-phenotype map in OI type III. , 1999, 13, 503-503.		17
70	Bone mineral content and collagen defects in osteogenesis imperfecta. Acta Paediatrica, International Journal of Paediatrics, 1999, 88, 1083-8.	0.7	10
71	Collagen-derived markers of bone metabolism in osteogenesis imperfecta. Acta Paediatrica, International Journal of Paediatrics, 1998, 87, 1131-7.	0.7	25
72	Dental manifestations of osteogenesis imperfecta and abnormalities of collagen I metabolism. Journal of Craniofacial Genetics and Developmental Biology, 1998, 18, 30-7.	0.1	26

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73	Parental mosaicism and autosomal dominant mutations causing structural abnormalities of collagen I are frequent in families with osteogenesis imperfecta type III/IV. Acta Paediatrica, International Journal of Paediatrics, 1997, 86, 711-718.	0.7	40
74	(G586V) substitutions in the Î±1 and Î±2 chains of collagen I: Effect of Î±-chain stoichiometry on the phenotype of osteogenesis imperfecta?. , 1997, 9, 431-436.		4
75	Deletion of a Gly-Pro-Pro repeat in the proÎ±2(I) chain of procollagen I in a family with dominant osteogenesis imperfecta type IV. Human Genetics, 1996, 97, 287-290.	1.8	9
76	GENETIC COUNSELLING AND PRENATAL DIAGNOSIS OF OSTEOGENESIS IMPERFECTA CAUSED BY PATERNAL MOSAICISM. , 1996, 16, 1032-1038.		14
77	Variable clinical expression in a family with OI type IV due to deletion of three base pairs in COL1A1. Clinical Genetics, 1996, 50, 304-309.	1.0	17