

Anna Wedell

List of Publications by Year in descending order

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Version: 2024-02-01

54
papers

1,928
citations

279487

23
h-index

264894

42
g-index

55
all docs

55
docs citations

55
times ranked

3373
citing authors

#	ARTICLE	IF	CITATIONS
1	PatientMatcher: A customizable Python-based open-source tool for matching undiagnosed rare disease patients via the Matchmaker Exchange network. <i>Human Mutation</i> , 2022, , .	1.1	5
2	Outcome at age 7 of epilepsy presenting in the first 2% years of life. A population-based study. <i>Epilepsia</i> , 2022, 63, 2096-2107.	2.6	5
3	Clinical Presentation, Genetic Etiology, and Coenzyme Q10 Levels in 55 Children with Combined Enzyme Deficiencies of the Mitochondrial Respiratory Chain. <i>Journal of Pediatrics</i> , 2021, 228, 240-251.e2.	0.9	6
4	Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in <i>NDUFB7</i> . <i>Human Mutation</i> , 2021, 42, 378-384.	1.1	8
5	Case Report: A Novel Mutation in the Mitochondrial MT-ND5 Gene Is Associated With Leber Hereditary Optic Neuropathy (LHON). <i>Frontiers in Neurology</i> , 2021, 12, 652590.	1.1	4
6	Novel Mutation m.10372A>G in <i>MT-ND3</i> Causing Sensorimotor Axonal Polyneuropathy. <i>Neurology: Genetics</i> , 2021, 7, e566.	0.9	3
7	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	3.6	116
8	Protocol for the derivation, culturing, and differentiation of human iPS-cell-derived neuroepithelial stem cells to study neural differentiation in vitro. <i>STAR Protocols</i> , 2021, 2, 100528.	0.5	11
9	First Year of TREC-Based National SCID Screening in Sweden. <i>International Journal of Neonatal Screening</i> , 2021, 7, 59.	1.2	8
10	Stable Isotope Labeling of Amino Acids in Flies (SILAF) Reveals Differential Phosphorylation of Mitochondrial Proteins Upon Loss of OXPHOS Subunits. <i>Molecular and Cellular Proteomics</i> , 2021, 20, 100065.	2.5	6
11	<i>FBXL4</i> deficiency increases mitochondrial removal by autophagy. <i>EMBO Molecular Medicine</i> , 2020, 12, e11659.	3.3	44
12	<i>SLC12A2</i> mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. <i>Neurology: Genetics</i> , 2020, 6, e478.	0.9	20
13	Epilepsy syndromes, etiologies, and the use of next-generation sequencing in epilepsy presenting in the first 2 years of life: A population-based study. <i>Epilepsia</i> , 2020, 61, 2486-2499.	2.6	24
14	Expanded Screening of One Million Swedish Babies with R4S and CLIR for Post-Analytical Evaluation of Data. <i>International Journal of Neonatal Screening</i> , 2020, 6, 42.	1.2	13
15	Loqusdb: added value of an observations database of local genomic variation. <i>BMC Bioinformatics</i> , 2020, 21, 273.	1.2	5
16	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. <i>Genome Medicine</i> , 2019, 11, 68.	3.6	88
17	Diagnostic pitfalls in vitamin B6-dependent epilepsy caused by mutations in the <i>PLPBP</i> gene. <i>JIMD Reports</i> , 2019, 50, 1-8.	0.7	16
18	MCEE Mutations in an Adult Patient with Parkinson's Disease, Dementia, Stroke and Elevated Levels of Methylmalonic Acid. <i>International Journal of Molecular Sciences</i> , 2019, 20, 2631.	1.8	5

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19	Heterogeneity of disease-causing variants in the Swedish galactosemia population: Identification of 16 novel <i>GALT</i> variants. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 1008-1018.	1.7	3
20	SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. <i>Stem Cell Reports</i> , 2019, 12, 696-711.	2.3	32
21	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. <i>Nature Communications</i> , 2019, 10, 1396.	5.8	11
22	Absence of TXNIP in Humans Leads to Lactic Acidosis and Low Serum Methionine Linked to Deficient Respiration on Pyruvate. <i>Diabetes</i> , 2019, 68, 709-723.	0.3	22
23	RNA modification landscape of the human mitochondrial tRNALys regulates protein synthesis. <i>Nature Communications</i> , 2018, 9, 3966.	5.8	61
24	Chorea, psychosis, acanthocytosis, and prolonged survival associated with <i>ELAC2</i> mutations. <i>Neurology</i> , 2018, 91, 710-712.	1.5	8
25	Detection of 6-demethoxyubiquinone in CoQ10 deficiency disorders: Insights into enzyme interactions and identification of potential therapeutics. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 216-223.	0.5	25
26	Respiratory chain complex III deficiency due to mutated BCS1L: a novel phenotype with encephalomyopathy, partially phenocopied in a Bcs1l mutant mouse model. <i>Orphanet Journal of Rare Diseases</i> , 2017, 12, 73.	1.2	20
27	Consensus recommendations for the diagnosis, treatment and follow-up of inherited methylation disorders. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 5-20.	1.7	47
28	Chorea, psychotic symptoms and long survival in a subject with <i>ELAC2</i> mutations. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2016, 87, A78.3-A79.	0.9	0
29	The Spectrum of PAH Mutations and Increase of Milder Forms of Phenylketonuria in Sweden During 1965-2014. <i>JIMD Reports</i> , 2016, 34, 19-26.	0.7	10
30	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. <i>American Journal of Human Genetics</i> , 2016, 99, 735-743.	2.6	99
31	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbach-Nishimura skeletal dysplasia due to pathogenic variants in <i>ALG9</i> . <i>European Journal of Human Genetics</i> , 2016, 24, 198-207.	1.4	29
32	Biotin and Thiamine Responsive Basal Ganglia Disease - A vital differential diagnosis in infants with severe encephalopathy. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 457-461.	0.7	18
33	Mitochondrial Polyadenylation Is a One-Step Process Required for mRNA Integrity and tRNA Maturation. <i>PLoS Genetics</i> , 2016, 12, e1006028.	1.5	43
34	The ketogenic diet compensates for <i>AGC1</i> deficiency and improves myelination. <i>Epilepsia</i> , 2015, 56, e176-81.	2.6	36
35	Mutations in <i>SLC12A5</i> in epilepsy of infancy with migrating focal seizures. <i>Nature Communications</i> , 2015, 6, 8038.	5.8	160
36	Cyclophilin D, a target for counteracting skeletal muscle dysfunction in mitochondrial myopathy. <i>Human Molecular Genetics</i> , 2015, 24, 6580-6587.	1.4	16

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37	SUV3 helicase is required for correct processing of mitochondrial transcripts. <i>Nucleic Acids Research</i> , 2015, 43, 7398-7413.	6.5	20
38	Rescue of primary ubiquinone deficiency due to a novel <i>COQ7</i> defect using 2,4-dihydroxybenzoic acid. <i>Journal of Medical Genetics</i> , 2015, 52, 779-783.	1.5	94
39	Biochemical and genetic diagnosis of 21-hydroxylase deficiency. <i>Endocrine</i> , 2015, 50, 306-314.	1.1	62
40	Intra-mitochondrial Methylation Deficiency Due to Mutations in <i>SLC25A26</i> . <i>American Journal of Human Genetics</i> , 2015, 97, 761-768.	2.6	58
41	Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. <i>BMC Genomics</i> , 2014, 15, 1090.	1.2	54
42	Nationwide Neonatal Screening for Congenital Adrenal Hyperplasia in Sweden. <i>JAMA Pediatrics</i> , 2014, 168, 567.	3.3	87
43	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. <i>American Journal of Human Genetics</i> , 2014, 95, 285-293.	2.6	110
44	Introduction to the ECR special issue on rare diseases. <i>Experimental Cell Research</i> , 2014, 325, 1.	1.2	0
45	One hundred years of congenital adrenal hyperplasia in Sweden: a retrospective, population-based cohort study. <i>Lancet Diabetes and Endocrinology</i> , 2013, 1, 35-42.	5.5	141
46	Molecular Genetics of 21-Hydroxylase Deficiency. <i>Endocrine Development</i> , 2011, 20, 80-87.	1.3	50
47	Congenital adrenal hyperplasia. <i>Clinical Biochemistry</i> , 2011, 44, 505-506.	0.8	6
48	<i>CYP21</i> mutations in simple virilizing congenital adrenal hyperplasia. <i>Journal of Molecular Medicine</i> , 2001, 79, 581-586.	1.7	20
49	A novel mutation (N233K) in the transactivating domain and the N756S mutation in the ligand binding domain of the androgen receptor gene are associated with male infertility*. <i>Clinical Endocrinology</i> , 2001, 54, 827-834.	1.2	23
50	Failure of Cortisone Acetate Treatment in Congenital Adrenal Hyperplasia because of Defective 11 β -Hydroxysteroid Dehydrogenase Reductase Activity*. <i>Journal of Clinical Endocrinology and Metabolism</i> , 1999, 84, 1210-1213.	1.8	54
51	No overrepresentation of congenital adrenal hyperplasia in patients with adrenocortical tumours. <i>Clinical Endocrinology</i> , 1999, 50, 343-346.	1.2	24
52	Novel missense mutation (P131R) in the HMG box of <i>SRY</i> in XY sex reversal. <i>Human Mutation</i> , 1998, 11, S328-S329.	1.1	11
53	Two new mutations in the porphobilinogen deaminase gene and a screening method using PCR amplification of specific alleles. <i>Human Genetics</i> , 1994, 93, 59-62.	1.8	20
54	Steroid 21-hydroxylase (P450c21): a new allele and spread of mutations through the pseudogene. <i>Human Genetics</i> , 1993, 91, 236-40.	1.8	67