Anna Wedell

List of Publications by Year
in descending order

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	Mutations in SLC12A5 in epilepsy of infancy with migrating focal seizures. Nature Communications, 2015, 6, 8038.	5.8	160
2	One hundred years of congenital adrenal hyperplasia in Sweden: a retrospective, population-based cohort study. Lancet Diabetes and Endocrinology,the, 2013, 1, 35-42.	5.5	141
3	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	3.6	116
4	Neu-Laxova Syndrome Is a Heterogeneous Metabolic Disorder Caused by Defects in Enzymes of the L-Serine Biosynthesis Pathway. American Journal of Human Genetics, 2014, 95, 285-293.	2.6	110
5	Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. American Journal of Human Genetics, 2016, 99, 735-743.	2.6	99
6	Rescue of primary ubiquinone deficiency due to a novelCOQ7defect using 2,4â€“dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783.	1.5	94
7	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. Genome Medicine, 2019, 11, 68.	3.6	88
8	Nationwide Neonatal Screening for Congenital Adrenal Hyperplasia in Sweden. JAMA Pediatrics, 2014, 168, 567.	3.3	87
9	Steroid 21-hydroxylase (P450c21): a new allele and spread of mutations through the pseudogene. Human Genetics, 1993, 91, 236-40.	1.8	67
10	Biochemical and genetic diagnosis of 21-hydroxylase deficiency. Endocrine, 2015, 50, 306-314.	1.1	62
11	RNA modification landscape of the human mitochondrial tRNALys regulates protein synthesis. Nature Communications, 2018, 9, 3966.	5.8	61
12	Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768.	2.6	58
13	Failure of Cortisone Acetate Treatment in Congenital Adrenal Hyperplasia because of Defective 11Î²-Hydroxysteroid Dehydrogenase Reductase Activity*. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 1210-1213.	1.8	54
14	Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. BMC Genomics, 2014, 15, 1090.	1.2	54
15	Molecular Genetics of 21- Hydroxylase Deficiency. Endocrine Development, 2011, 20, 80-87.	1.3	50
16	Consensus recommendations for the diagnosis, treatment and followâ€up of inherited methylation disorders. Journal of Inherited Metabolic Disease, 2017, 40, 5-20.	1.7	47
17	<scp>FBXL</scp> 4 deficiency increases mitochondrial removal by autophagy. EMBO Molecular Medicine, 2020, 12, e11659.	3.3	44
18	Mitochondrial Polyadenylation Is a One-Step Process Required for mRNA Integrity and tRNA Maturation. PLoS Genetics, 2016, 12, e1006028.	1.5	43

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19	The ketogenic diet compensates for <scp>AGC</scp>1 deficiency and improves myelination. Epilepsia, 2015, 56, e176-81.	2.6	36
20	SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. Stem Cell Reports, 2019, 12, 696-711.	2.3	32
21	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbachâ€“Nishimura skeletal dysplasia due to pathogenic variants in ALG9. European Journal of Human Genetics, 2016, 24, 198-207.	1.4	29
22	Detection of 6-demethoxyubiquinone in CoQ10 deficiency disorders: Insights into enzyme interactions and identification of potential therapeutics. Molecular Genetics and Metabolism, 2017, 121, 216-223.	0.5	25
23	No overrepresentation of congenital adrenal hyperplasia in patients with adrenocortical tumours. Clinical Endocrinology, 1999, 50, 343-346.	1.2	24
24	Epilepsy syndromes, etiologies, and the use of nextâ€generation sequencing in epilepsy presenting in the first 2 years of life: A populationâ€based study. Epilepsia, 2020, 61, 2486-2499.	2.6	24
25	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*. Clinical Endocrinology, 2001, 54, 827-834.	1.2	23
26	Absence of TXNIP in Humans Leads to Lactic Acidosis and Low Serum Methionine Linked to Deficient Respiration on Pyruvate. Diabetes, 2019, 68, 709-723.	0.3	22
27	Two new mutations in the porphobilinogen deaminase gene and a screening method using PCR amplification of specific alleles. Human Genetics, 1994, 93, 59-62.	1.8	20
28	CYP21 mutations in simple virilizing congenital adrenal hyperplasia. Journal of Molecular Medicine, 2001, 79, 581-586.	1.7	20
29	SUV3 helicase is required for correct processing of mitochondrial transcripts. Nucleic Acids Research, 2015, 43, 7398-7413.	6.5	20
30	Respiratory chain complex III deficiency due to mutated BCS1L: a novel phenotype with encephalomyopathy, partially phenocopied in a Bcs1l mutant mouse model. Orphanet Journal of Rare Diseases, 2017, 12, 73.	1.2	20
31	SLC12A2 mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. Neurology: Genetics, 2020, 6, e478.	0.9	20
32	Biotin and Thiamine Responsive Basal Ganglia Disease â€“ A vital differential diagnosis in infants with severe encephalopathy. European Journal of Paediatric Neurology, 2016, 20, 457-461.	0.7	18
33	Cyclophilin D, a target for counteracting skeletal muscle dysfunction in mitochondrial myopathy. Human Molecular Genetics, 2015, 24, 6580-6587.	1.4	16
34	Diagnostic pitfalls in vitamin B6â€dependent epilepsy caused by mutations in the PLPBP gene. JIMD Reports, 2019, 50, 1-8.	0.7	16
35	Expanded Screening of One Million Swedish Babies with R4S and CLIR for Post-Analytical Evaluation of Data. International Journal of Neonatal Screening, 2020, 6, 42.	1.2	13
36	Novel missense mutation (P131R) in the HMG box of SRY in XY sex reversal. Human Mutation, 1998, 11, S328-S329.	1.1	11

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37	Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. Nature Communications, 2019, 10, 1396.	5.8	11
38	Protocol for the derivation, culturing, and differentiation of human iPS-cell-derived neuroepithelial stem cells to study neural differentiation inÂvitro. STAR Protocols, 2021, 2, 100528.	0.5	11
39	The Spectrum of PAH Mutations and Increase of Milder Forms of Phenylketonuria in Sweden During 1965â€“2014. JIMD Reports, 2016, 34, 19-26.	0.7	10
40	Chorea, psychosis, acanthocytosis, and prolonged survival associated with ELAC2 mutations. Neurology, 2018, 91, 710-712.	1.5	8
41	Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in NDUFB7. Human Mutation, 2021, 42, 378-384.	1.1	8
42	First Year of TREC-Based National SCID Screening in Sweden. International Journal of Neonatal Screening, 2021, 7, 59.	1.2	8
43	Congenital adrenal hyperplasia. Clinical Biochemistry, 2011, 44, 505-506.	0.8	6
44	Clinical Presentation, Genetic Etiology, and Coenzyme Q10 Levels in 55 Children with Combined Enzyme Deficiencies of the Mitochondrial Respiratory Chain. Journal of Pediatrics, 2021, 228, 240-251.e2.	0.9	6
45	Stable Isotope Labeling of Amino Acids in Flies (SILAF) Reveals Differential Phosphorylation of Mitochondrial Proteins Upon Loss of OXPHOS Subunits. Molecular and Cellular Proteomics, 2021, 20, 100065.	2.5	6
46	MCEE Mutations in an Adult Patient with Parkinsonâ€™s Disease, Dementia, Stroke and Elevated Levels of Methylmalonic Acid. International Journal of Molecular Sciences, 2019, 20, 2631.	1.8	5
47	Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273.	1.2	5
48	PatientMatcher: A customizable Pythonâ€based openâ€source tool for matching undiagnosed rare disease patients via the Matchmaker Exchange network. Human Mutation, 2022, , .	1.1	5
49	Outcome at age 7 of epilepsy presenting in the first 2â€‰years of life. A populationâ€based study. Epilepsia, 2022, 63, 2096-2107.	2.6	5
50	Case Report: A Novel Mutation in the Mitochondrial MT-ND5 Gene Is Associated With Leber Hereditary Optic Neuropathy (LHON). Frontiers in Neurology, 2021, 12, 652590.	1.1	4
51	Heterogeneity of diseaseâ€causing variants in the Swedish galactosemia population: Identification of 16 novel GALT variants. Journal of Inherited Metabolic Disease, 2019, 42, 1008-1018.	1.7	3
52	Novel Mutation m.10372A>G in MT-ND3 Causing Sensorimotor Axonal Polyneuropathy. Neurology: Genetics, 2021, 7, e566.	0.9	3
53	Introduction to the ECR special issue on rare diseases. Experimental Cell Research, 2014, 325, 1.	1.2	0
54	J10â€…Chorea, psychotic symptoms and long survival in a subject with ELAC2 mutations. Journal of Neurology, Neurosurgery and Psychiatry, 2016, 87, A78.3-A79.	0.9	0