# Anna Wedell

List of Publications by Year
in descending order



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|---|---|---|---|
| 54 papers | 1,928 citations | 279798 23 h-index | 265206 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. | 12.8 | 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. | 11.4 | 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. | 8.2 | 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. | 6.2 | 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. | 6.2 | 99 |
| 6 | Rescue of primary ubiquinone deficiency due to a novelCOQ7defect using 2,4â€“dihydroxybensoic acid. Journal of Medical Genetics, 2015, 52, 779-783. | 3.2 | 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. | 8.2 | 88 |
| 8 | Nationwide Neonatal Screening for Congenital Adrenal Hyperplasia in Sweden. JAMA Pediatrics, 2014, 168, 567. | 6.2 | 87 |
| 9 | Steroid 21-hydroxylase (P450c21): a new allele and spread of mutations through the pseudogene. Human Genetics, 1993, 91, 236-40. | 3.8 | 67 |
| 10 | Biochemical and genetic diagnosis of 21-hydroxylase deficiency. Endocrine, 2015, 50, 306-314. | 2.3 | 62 |
| 11 | RNA modification landscape of the human mitochondrial tRNALys regulates protein synthesis. Nature Communications, 2018, 9, 3966. | 12.8 | 61 |
| 12 | Intra-mitochondrial Methylation Deficiency Due to Mutations in SLC25A26. American Journal of Human Genetics, 2015, 97, 761-768. | 6.2 | 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. | 3.6 | 54 |
| 14 | Rapid pulsed whole genome sequencing for comprehensive acute diagnostics of inborn errors of metabolism. BMC Genomics, 2014, 15, 1090. | 2.8 | 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. | 3.6 | 47 |
| 17 | <scp>FBXL</scp> 4 deficiency increases mitochondrial removal by autophagy. EMBO Molecular Medicine, 2020, 12, e11659. | 6.9 | 44 |
| 18 | Mitochondrial Polyadenylation Is a One-Step Process Required for mRNA Integrity and tRNA Maturation. PLoS Genetics, 2016, 12, e1006028. | 3.5 | 43 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | The ketogenic diet compensates for <scp>AGC</scp>1 deficiency and improves myelination. Epilepsia, 2015, 56, e176-81. | 5.1 | 36 |
| 20 | SQSTM1/p62-Directed Metabolic Reprogramming Is Essential for Normal Neurodifferentiation. Stem Cell Reports, 2019, 12, 696-711. | 4.8 | 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. | 2.8 | 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. | 1.1 | 25 |
| 23 | No overrepresentation of congenital adrenal hyperplasia in patients with adrenocortical tumours. Clinical Endocrinology, 1999, 50, 343-346. | 2.4 | 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. | 5.1 | 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. | 2.4 | 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. | 0.6 | 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. | 3.8 | 20 |
| 28 | CYP21 mutations in simple virilizing congenital adrenal hyperplasia. Journal of Molecular Medicine, 2001, 79, 581-586. | 3.9 | 20 |
| 29 | SUV3 helicase is required for correct processing of mitochondrial transcripts. Nucleic Acids Research, 2015, 43, 7398-7413. | 14.5 | 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. | 2.7 | 20 |
| 31 | SLC12A2 mutations cause NKCC1 deficiency with encephalopathy and impaired secretory epithelia. Neurology: Genetics, 2020, 6, e478. | 1.9 | 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. | 1.6 | 18 |
| 33 | Cyclophilin D, a target for counteracting skeletal muscle dysfunction in mitochondrial myopathy. Human Molecular Genetics, 2015, 24, 6580-6587. | 2.9 | 16 |
| 34 | Diagnostic pitfalls in vitamin B6â€dependent epilepsy caused by mutations in the PLPBP gene. JIMD Reports, 2019, 50, 1-8. | 1.5 | 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. | 3.2 | 13 |
| 36 | Novel missense mutation (P131R) in the HMG box of SRY in XY sex reversal. Human Mutation, 1998, 11, S328-S329. | 2.5 | 11 |

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|---|---|---|---|
| 37 | Myoglobinopathy is an adult-onset autosomal dominant myopathy with characteristic sarcoplasmic inclusions. Nature Communications, 2019, 10, 1396. | 12.8 | 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. | 1.2 | 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. | 1.5 | 10 |
| 40 | Chorea, psychosis, acanthocytosis, and prolonged survival associated with ELAC2 mutations. Neurology, 2018, 91, 710-712. | 1.1 | 8 |
| 41 | Severe congenital lactic acidosis and hypertrophic cardiomyopathy caused by an intronic variant in NDUFB7. Human Mutation, 2021, 42, 378-384. | 2.5 | 8 |
| 42 | First Year of TREC-Based National SCID Screening in Sweden. International Journal of Neonatal Screening, 2021, 7, 59. | 3.2 | 8 |
| 43 | Congenital adrenal hyperplasia. Clinical Biochemistry, 2011, 44, 505-506. | 1.9 | 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. | 1.8 | 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. | 3.8 | 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. | 4.1 | 5 |
| 47 | Loqusdb: added value of an observations database of local genomic variation. BMC Bioinformatics, 2020, 21, 273. | 2.6 | 5 |
| 48 | PatientMatcher: A customizable Pythonâ€based openâ€source tool for matching undiagnosed rare disease patients via the Matchmaker Exchange network. Human Mutation, 2022, , . | 2.5 | 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. | 5.1 | 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. | 2.4 | 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. | 3.6 | 3 |
| 52 | Novel Mutation m.10372A>G in MT-ND3 Causing Sensorimotor Axonal Polyneuropathy. Neurology: Genetics, 2021, 7, e566. | 1.9 | 3 |
| 53 | Introduction to the ECR special issue on rare diseases. Experimental Cell Research, 2014, 325, 1. | 2.6 | 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. | 1.9 | 0 |