Saskia B Wortmann

List of Publications by Year
in descending order

118	4,587	76326	128289
papers	citations	40 h-index	60 g-index

130	130	130	6994
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. Human Mutation, 2013, 34, 1721-1726.	2.5	303
2	Whole exome sequencing of suspected mitochondrial patients in clinical practice. Journal of Inherited Metabolic Disease, 2015, 38, 437-443.	3.6	186
3	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. Nature Genetics, 2012, 44, 797-802.	21.4	175
4	Nextâ€generation metabolic screening: targeted and untargeted metabolomics for the diagnosis of inborn errors of metabolism in individual patients. Journal of Inherited Metabolic Disease, 2018, 41, 337-353.	3.6	145
5	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
6	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. American Journal of Human Genetics, 2015, 96, 245-257.	6.2	111
7	CADmutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	106
8	Biochemical and genetic analysis of 3-methylglutaconic aciduria type IV: a diagnostic strategy. Brain, 2009, 132, 136-146.	7.6	90
9	Mutation in mitochondrial ribosomal protein MRPS22 leads to Cornelia de Lange-like phenotype, brain abnormalities and hypertrophic cardiomyopathy. European Journal of Human Genetics, 2011, 19, 394-399.	2.8	90
10	Treating neutropenia and neutrophil dysfunction in glycogen storage disease type Ib with an SGLT2 inhibitor. Blood, 2020, 136, 1033-1043.	1.4	90
11	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. Genome Medicine, 2022, 14, 38.	8.2	85
12	Inborn errors of metabolism with 3â€methylglutaconic aciduria as discriminative feature: proper classification and nomenclature. Journal of Inherited Metabolic Disease, 2013, 36, 923-928.	3.6	84
13	Molecular structural diversity of mitochondrial cardiolipins. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 4158-4163.	7.1	82
14	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. Brain, 2019, 142, 3382-3397.	7.6	76
15	Dimerization of the cytokine receptors gp130 and LIFR analysed in single cells. Journal of Cell Science, 2005, 118, 5129-5140.	2.0	74
16	3â€Methylglutaconic aciduriaâ€”lessons from 50 genes and 977 patients. Journal of Inherited Metabolic Disease, 2013, 36, 913-921.	3.6	74
17	Delineating MT-ATP6 -associated disease. Neurology: Genetics, 2020, 6, e393.	1.9	73
18	The 3â€methylglutaconic acidurias: what's new?. Journal of Inherited Metabolic Disease, 2012, 35, 13-22.	3.6	72

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19	SDHA mutations causing a multisystem mitochondrial disease: novel mutations and genetic overlap with hereditary tumors. European Journal of Human Genetics, 2015, 23, 202-209.	2.8	71
20	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Molecular Genetics and Metabolism, 2014, 111, 342-352.	1.1	65
21	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
22	Progressive deafnessâ€“dystonia due to SERAC1 mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
23	Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective?. Orphanet Journal of Rare Diseases, 2018, 13, 120.	2.7	61
24	A Guideline for the Diagnosis of Pediatric Mitochondrial Disease: The Value of Muscle and Skin Biopsies in the Genetics Era. Neuropediatrics, 2017, 48, 309-314.	0.6	60
25	Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. American Journal of Human Genetics, 2017, 101, 525-538.	6.2	58
26	Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain, 2017, 140, e11-e11.	7.6	57
27	Mitochondrial DNA mutation analysis from exome sequencingâ€”A more holistic approach in diagnostics of suspected mitochondrial disease. Journal of Inherited Metabolic Disease, 2019, 42, 909-917.	3.6	57
28	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	6.2	56
29	The role of the clinician in the multiâ€omics era: are you ready?. Journal of Inherited Metabolic Disease, 2018, 41, 571-582.	3.6	55
30	Defective metabolic programming impairs early neuronal morphogenesis in neural cultures and an organoid model of Leigh syndrome. Nature Communications, 2021, 12, 1929.	12.8	55
31	Treatable inherited metabolic disorders causing intellectual disability: 2021 review and digital app. Orphanet Journal of Rare Diseases, 2021, 16, 170.	2.7	52
32	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. Journal of Inherited Metabolic Disease, 2011, 34, 923-927.	3.6	50
33	Inborn errors of metabolism in the biosynthesis and remodelling of phospholipids. Journal of Inherited Metabolic Disease, 2015, 38, 99-110.	3.6	47
34	Long Term Association of the Cytokine Receptor gp130 and the Janus Kinase Jak1 Revealed by FRAP Analysis. Journal of Biological Chemistry, 2003, 278, 39205-39213.	3.4	46
35	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
36	A novel mutation in COQ2 leading to fatal infantile multisystem disease. Journal of the Neurological Sciences, 2013, 326, 24-28.	0.6	45

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37	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	3.6	45
38	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1ÂDeficiency. Journal of Pediatrics, 2016, 175, 130-136.e8.	1.8	43
39	Mitochondrial dysfunction and organic aciduria in five patients carrying mutations in the Ras-MAPK pathway. European Journal of Human Genetics, 2011, 19, 138-144.	2.8	42
40	Cholineâ€relatedâ€inherited metabolic diseasesâ€"A mini review. Journal of Inherited Metabolic Disease, 2019, 42, 237-242.	3.6	42
41	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. Fertility and Sterility, 2017, 108, 168-174.	1.0	42
42	Molecular and clinical spectra of FBXL4 deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
43	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	6.2	40
44	Muddâ€™s disease (MAT I/III deficiency): a survey of data for MAT1A homozygotes and compound heterozygotes. Orphanet Journal of Rare Diseases, 2015, 10, 99.	2.7	39
45	Pathogenic Bi-allelic Mutations in NDUFAF8 Cause Leigh Syndrome with an Isolated Complex I Deficiency. American Journal of Human Genetics, 2020, 106, 92-101.	6.2	39
46	â€œTranscriptomicsâ€: molecular diagnosis of inborn errors of metabolism via RNAâ€sequencing. Journal of Inherited Metabolic Disease, 2018, 41, 525-532.	3.6	38
47	Improved inflammatory bowel disease, wound healing and normal oxidative burst under treatment with empagliflozin in glycogen storage disease type Ib. Orphanet Journal of Rare Diseases, 2020, 15, 218.	2.7	37
48	Bi-Allelic UQCRFS1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	6.2	36
49	The role of the mitochondrial ribosome in human disease: searching for mutations in 12S mitochondrial rRNA with high disruptive potential. Human Molecular Genetics, 2014, 23, 949-967.	2.9	35
50	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. EBioMedicine, 2020, 54, 102730.	6.1	35
51	Developing outcome measures for pediatric mitochondrial disorders: Which complaints and limitations are most burdensome to patients and their parents?. Mitochondrion, 2013, 13, 15-24.	3.4	34
52	Mitochondrial Energy Production Correlates With the Age-Related BMI. Pediatric Research, 2009, 65, 103-108.	2.3	32
53	Brain imaging in classic nonketotic hyperglycinemia: Quantitative analysis and relation to phenotype. Journal of Inherited Metabolic Disease, 2019, 42, 438-450.	3.6	30
54	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	6.2	30

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55	Efficacy and safety of empagliflozin in glycogen storage disease type Ib: Data from an international questionnaire. Genetics in Medicine, 2022, 24, 1781-1788.	2.4	29
56	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. Journal of Medical Genetics, 2018, 55, 39-47.	3.2	28
57	Mitochondrial DNA m.3242GÂ>ÂA mutation, an under diagnosed cause of hypertrophic cardiomyopathy and renal tubular dysfunction?. European Journal of Medical Genetics, 2012, 55, 552-556.	1.3	27
58	A scoring system predicting the clinical course of CLPB defect based on the foetal and neonatal presentation of 31 patients. Journal of Inherited Metabolic Disease, 2017, 40, 853-860.	3.6	27
59	Ketogenic diet for mitochondrial disease: a systematic review on efficacy and safety. Orphanet Journal of Rare Diseases, 2021, 16, 295.	2.7	26
60	Biallelic variants inWARS2encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	2.5	24
61	The genotypic and phenotypic spectrum of MTO1 deficiency. Molecular Genetics and Metabolism, 2018, 123, 28-42.	1.1	24
62	Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. Brain, 2019, 142, 3876-3891.	7.6	23
63	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. Genetics in Medicine, 2021, 23, 1705-1714.	2.4	22
64	HTRA2 Defect: A Recognizable Inborn Error of Metabolism with 3-Methylglutaconic Aciduria as Discriminating Feature Characterized by Neonatal Movement Disorder and Epilepsyâ€”Report of 11 Patients. Neuropediatrics, 2018, 49, 373-378.	0.6	21
65	Long Survival in Leigh Syndrome: New Cases and Review of Literature. Neuropediatrics, 2014, 45, 346-353.	0.6	20
66	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
67	How to proceed after â€œnegativeâ€•exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. Journal of Inherited Metabolic Disease, 2022, 45, 663-681.	3.6	20
68	Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. Genetics in Medicine, 2020, 22, 908-916.	2.4	19
69	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	2.4	19
70	Expanding the clinical and genetic spectrum of CAD deficiency: an epileptic encephalopathy treatable with uridine supplementation. Genetics in Medicine, 2020, 22, 1589-1597.	2.4	19
71	Variants in NGLY1 lead to intellectual disability, myoclonus epilepsy, sensorimotor axonal polyneuropathy and mitochondrial dysfunction. Clinical Genetics, 2020, 97, 556-566.	2.0	19
72	Common mutation in the PHKA2 gene with variable phenotype in patients with liver phosphorylase b kinase deficiency. Molecular Genetics and Metabolism, 2011, 104, 691-694.	1.1	18

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73	The role of clinical response to treatment in determining pathogenicity of genomic variants. Genetics in Medicine, 2021, 23, 581-585.	2.4	18
74	Leucine Loading Test is Only Discriminative for 3-Methylglutaconic Aciduria Due to AUH Defect. JIMD Reports, 2014, 16, 1-6.	1.5	17
75	Fatal pitfalls in newborn screening for mitochondrial trifunctional protein (MTP)/long-chain 3-Hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency. Orphanet Journal of Rare Diseases, 2018, 13, 122.	2.7	17
76	CLPB (caseinolytic peptidase B homolog), the first mitochondrial protein refoldase associated with human disease. Biochimica Et Biophysica Acta - General Subjects, 2020, 1864, 129512.	2.4	17
77	Bi-allelic Variants in TKFC Encoding Triokinase/FMN Cyclase Are Associated with Cataracts and Multisystem Disease. American Journal of Human Genetics, 2020, 106, 256-263.	6.2	16
78	Heterozygous truncating variants in SUFU cause congenital ocular motor apraxia. Genetics in Medicine, 2021, 23, 341-351.	2.4	16
79	Functional interpretation of ATAD3A variants in neuro-mitochondrial phenotypes. Genome Medicine, 2021, 13, 55.	8.2	16
80	Mild orotic aciduria in UMPS heterozygotes: a metabolic finding without clinical consequences. Journal of Inherited Metabolic Disease, 2017, 40, 423-431.	3.6	14
81	3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. Clinica Chimica Acta, 2017, 471, 95-100.	1.1	14
82	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. Neuropediatrics, 2018, 49, 059-062.	0.6	14
83	Phosphoglucomutase-1 deficiency: Early presentation, metabolic management and detection in neonatal blood spots. Molecular Genetics and Metabolism, 2020, 131, 135-146.	1.1	14
84	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
85	Galactokinase deficiency: lessons from the GalNet registry. Genetics in Medicine, 2021, 23, 202-210.	2.4	14
86	A spoonful of Lâ€fucoseâ€"an efficient therapy for GFUSâ€CDG, a new glycosylation disorder. EMBO Molecular Medicine, 2021, 13, e14332.	6.9	13
87	Austrian study shows that delays in accessing acute paediatric health care outweighed the risks of COVIDâ€19. Acta Paediatrica, International Journal of Paediatrics, 2020, 109, 2309-2310.	1.5	12
88	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. Brain, 2021, 144, e30-e30.	7.6	12
89	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
90	Elevated Homocysteine after Elevated Propionylcarnitine or Low Methionine in Newborn Screening Is Highly Predictive for Low Vitamin B12 and Holo-Transcobalamin Levels in Newborns. Diagnostics, 2020, 10, 626.	2.6	11

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91	Expanding the clinical and genetic spectrum of FDXR deficiency by functional validation of variants of uncertain significance. Human Mutation, 2021, 42, 310-319.	2.5	11
92	Expanding the phenotypic spectrum of BCS1Lâ€related mitochondrial disease. Annals of Clinical and Translational Neurology, 2021, 8, 2155-2165.	3.7	11
93	Diagnosis and Management of Drooling in Children With Progressive Dystonia. Journal of Child Neurology, 2016, 31, 1220-1226.	1.4	10
94	A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscular Disorders, 2015, 25, 262-267.	0.6	9
95	Clinicoâ€radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. Human Mutation, 2022, 43, 403-419.	2.5	9
96	SUCLA2 Deficiency: A Deafness-Dystonia Syndrome with Distinctive Metabolic Findings (Report of a) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.5	8
97	Severe ichthyosis in MPDU1â€CDG. Journal of Inherited Metabolic Disease, 2018, 41, 1293-1294.	3.6	8
98	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. Genetics in Medicine, 2021, 23, 2415-2425.	2.4	8
99	Congenital disorders of glycosylation with defective fucosylation. Journal of Inherited Metabolic Disease, 2021, 44, 1441-1452.	3.6	8
100	Ketogenic diet for treating alopecia in BCS1lâ€related mitochondrial disease (Bjornstad syndrome). JIMD Reports, 2020, 53, 10-11.	1.5	7
101	Mutation of the WARS2 Gene as the Cause of a Severe Hyperkinetic Movement Disorder. Movement Disorders Clinical Practice, 2020, 7, 88-90.	1.5	6
102	Teaching NeuroImages: Neuroimaging Findings in Inosine Triphosphate Pyrophosphohydrolase Deficiency. Neurology, 2021, 97, e109-e110.	1.1	6
103	Thiamine Pyrophosphokinase Deficiency due to Mutations in the TPK1 Gene: A Rare, Treatable Neurodegenerative Disorder. Neuropediatrics, 2021, 52, 126-132.	0.6	6
104	Cytosolic Phosphoenolpyruvate Carboxykinase Deficiency: Cause of Hypoglycemia-Induced Seizure and Death. Neuropediatrics, 2021, 52, 398-402.	0.6	5
105	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	2.4	5
106	A retrospective study on disease management in children and adolescents with phenylketonuria during the Covid-19 pandemic lockdown in Austria. Orphanet Journal of Rare Diseases, 2021, 16, 367.	2.7	5
107	Pathogenic variants in MRPL44 cause infantile cardiomyopathy due to a mitochondrial translation defect. Molecular Genetics and Metabolism, 2021, 133, 362-371.	1.1	5
108	Prevalence and clinical prediction of mitochondrial disorders in a large neuropediatric cohort. Clinical Genetics, 2021, 100, 766-770.	2.0	5

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109	Muscular and Molecular Pathology Associated with SPATA5 Deficiency in a Child with EHLMRS. International Journal of Molecular Sciences, 2021, 22, 7835.	4.1	4
110	Mutations in <scp>HID1</scp> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. Annals of Neurology, 2021, 90, 143-158.	5.3	3
111	Mitochondrial Disease and Hearing Loss in Children: A Systematic Review. Laryngoscope, 2022, 132, 2459-2472.	2.0	3
112	Refractory severe intestinal vasculitis due to Henochâ€ŠschÃ¶nlein Purpura: successful treatment with plasmapheresis. Acta Paediatrica, International Journal of Paediatrics, 2006, 95, 622-623.	1.5	0
113	Response to Biesecker et al.. Genetics in Medicine, 2021, 23, 793-794.	2.4	0
114	P 233. Thiamine Pyrophosphokinase Deficiency due to Mutations in the TPK1 Gene: A Rare, Treatable Neurodegenerative Disorder. , 2018, 49, .		0
115	History of Repeated Bleeding from Intact Skin and Mucous Membranes: A Quiz. Acta Dermato-Venereologica, 2021, 101, adv00592.	1.3	0
116	ATP synthase deficiency due to m.8528T>C mutationÂâ€“ a novel cause of severe neonatal hyperammonemia requiring hemodialysis. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 389-393.	0.9	0
117	Mitochondrial Transporter Defects: Successful Treatment with Ketogenic Diet Therapy. Neuropediatrics, 2021, 52, .	0.6	0
118	The switch in the diagnosis of mitochondrial diseases from the classical 'function first' to the NGS-based 'genetics first' diagnostic era. Medycyna Wieku Rozwojowego, 2020, 24, 47-52.	0.2	0