

Saskia B Wortmann

List of Publications by Year in descending order

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Version: 2024-02-01

118
papers

4,587
citations

76326

40
h-index

128289

60
g-index

130
all docs

130
docs citations

130
times ranked

6994
citing authors

#	ARTICLE	IF	CITATIONS
1	Variants in Mitochondrial ATP Synthase Cause Variable Neurologic Phenotypes. <i>Annals of Neurology</i> , 2022, 91, 225-237.	5.3	12
2	Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency. <i>Human Mutation</i> , 2022, 43, 403-419.	2.5	9
3	Mitochondrial Disease and Hearing Loss in Children: A Systematic Review. <i>Laryngoscope</i> , 2022, 132, 2459-2472.	2.0	3
4	Clinical implementation of RNA sequencing for Mendelian disease diagnostics. <i>Genome Medicine</i> , 2022, 14, 38.	8.2	85
5	How to proceed after "negative" exome: A review on genetic diagnostics, limitations, challenges, and emerging new multiomics techniques. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 663-681.	3.6	20
6	Efficacy and safety of empagliflozin in glycogen storage disease type Ib: Data from an international questionnaire. <i>Genetics in Medicine</i> , 2022, 24, 1781-1788.	2.4	29
7	The role of clinical response to treatment in determining pathogenicity of genomic variants. <i>Genetics in Medicine</i> , 2021, 23, 581-585.	2.4	18
8	Heterozygous truncating variants in <i>SUFU</i> cause congenital ocular motor apraxia. <i>Genetics in Medicine</i> , 2021, 23, 341-351.	2.4	16
9	De novo variants in <i>SNAP25</i> cause an early-onset developmental and epileptic encephalopathy. <i>Genetics in Medicine</i> , 2021, 23, 653-660.	2.4	20
10	Galactokinase deficiency: lessons from the GalNet registry. <i>Genetics in Medicine</i> , 2021, 23, 202-210.	2.4	14
11	Defective phosphatidylethanolamine biosynthesis leads to a broad ataxia-spasticity spectrum. <i>Brain</i> , 2021, 144, e30-e30.	7.6	12
12	Response to Biesecker et al.. <i>Genetics in Medicine</i> , 2021, 23, 793-794.	2.4	0
13	Cytosolic Phosphoenolpyruvate Carboxykinase Deficiency: Cause of Hypoglycemia-Induced Seizure and Death. <i>Neuropediatrics</i> , 2021, 52, 398-402.	0.6	5
14	Expanding the clinical and genetic spectrum of <i>FDXR</i> deficiency by functional validation of variants of uncertain significance. <i>Human Mutation</i> , 2021, 42, 310-319.	2.5	11
15	Teaching NeuroImages: Neuroimaging Findings in Inosine Triphosphate Pyrophosphohydrolase Deficiency. <i>Neurology</i> , 2021, 97, e109-e110.	1.1	6
16	Defective metabolic programming impairs early neuronal morphogenesis in neural cultures and an organoid model of Leigh syndrome. <i>Nature Communications</i> , 2021, 12, 1929.	12.8	55
17	Treatable inherited metabolic disorders causing intellectual disability: 2021 review and digital app. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 170.	2.7	52
18	Functional interpretation of <i>ATAD3A</i> variants in neuro-mitochondrial phenotypes. <i>Genome Medicine</i> , 2021, 13, 55.	8.2	16

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19	Mutations in <i>HID1</i> Cause Syndromic Infantile Encephalopathy and Hypopituitarism. <i>Annals of Neurology</i> , 2021, 90, 143-158.	5.3	3
20	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. <i>Genetics in Medicine</i> , 2021, 23, 1873-1881.	2.4	5
21	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. <i>Genetics in Medicine</i> , 2021, 23, 1705-1714.	2.4	22
22	Muscular and Molecular Pathology Associated with SPATA5 Deficiency in a Child with EHLMRS. <i>International Journal of Molecular Sciences</i> , 2021, 22, 7835.	4.1	4
23	Ketogenic diet for mitochondrial disease: a systematic review on efficacy and safety. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 295.	2.7	26
24	A retrospective study on disease management in children and adolescents with phenylketonuria during the Covid-19 pandemic lockdown in Austria. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 367.	2.7	5
25	Pathogenic variants in MRPL44 cause infantile cardiomyopathy due to a mitochondrial translation defect. <i>Molecular Genetics and Metabolism</i> , 2021, 133, 362-371.	1.1	5
26	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. <i>Genetics in Medicine</i> , 2021, 23, 2415-2425.	2.4	8
27	Congenital disorders of glycosylation with defective fucosylation. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1441-1452.	3.6	8
28	A spoonful of L-fucose" an efficient therapy for GFUS"CDG, a new glycosylation disorder. <i>EMBO Molecular Medicine</i> , 2021, 13, e14332.	6.9	13
29	Prevalence and clinical prediction of mitochondrial disorders in a large neuropediatric cohort. <i>Clinical Genetics</i> , 2021, 100, 766-770.	2.0	5
30	Thiamine Pyrophosphokinase Deficiency due to Mutations in the TPK1 Gene: A Rare, Treatable Neurodegenerative Disorder. <i>Neuropediatrics</i> , 2021, 52, 126-132.	0.6	6
31	Expanding the phenotypic spectrum of <i>BCS1L</i> -related mitochondrial disease. <i>Annals of Clinical and Translational Neurology</i> , 2021, 8, 2155-2165.	3.7	11
32	History of Repeated Bleeding from Intact Skin and Mucous Membranes: A Quiz. <i>Acta Dermato-Venereologica</i> , 2021, 101, adv00592.	1.3	0
33	ATP synthase deficiency due to m.8528T>C mutation" a novel cause of severe neonatal hyperammonemia requiring hemodialysis. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2021, 34, 389-393.	0.9	0
34	Mitochondrial Transporter Defects: Successful Treatment with Ketogenic Diet Therapy. <i>Neuropediatrics</i> , 2021, 52, .	0.6	0
35	CLPB (caseinolytic peptidase B homolog), the first mitochondrial protein refoldase associated with human disease. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2020, 1864, 129512.	2.4	17
36	Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. <i>Genetics in Medicine</i> , 2020, 22, 908-916.	2.4	19

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37	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
38	Bi-Allelic UQCRCF1 Variants Are Associated with Mitochondrial Complex III Deficiency, Cardiomyopathy, and Alopecia Totalis. American Journal of Human Genetics, 2020, 106, 102-111.	6.2	36
39	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
40	Defining clinical subgroups and genotype-phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	2.4	46
41	Monogenic variants in dystonia: an exome-wide sequencing study. Lancet Neurology, The, 2020, 19, 908-918.	10.2	139
42	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
43	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
44	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
45	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
46	Austrian study shows that delays in accessing acute paediatric health care outweighed the risks of COVID-19. Acta Paediatrica, International Journal of Paediatrics, 2020, 109, 2309-2310.	1.5	12
47	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
48	Phosphoglucomutase-1 deficiency: Early presentation, metabolic management and detection in neonatal blood spots. Molecular Genetics and Metabolism, 2020, 131, 135-146.	1.1	14
49	Ketogenic diet for treating alopecia in BCS1L-related mitochondrial disease (Bjornstad syndrome). JIMD Reports, 2020, 53, 10-11.	1.5	7
50	Treating neutropenia and neutrophil dysfunction in glycogen storage disease type Ib with an SGLT2 inhibitor. Blood, 2020, 136, 1033-1043.	1.4	90
51	Delineating <i>MT-ATP6</i> -associated disease. Neurology: Genetics, 2020, 6, e393.	1.9	73
52	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
53	Variants in <i>NGLY1</i> lead to intellectual disability, myoclonus epilepsy, sensorimotor axonal polyneuropathy and mitochondrial dysfunction. Clinical Genetics, 2020, 97, 556-566.	2.0	19
54	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

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55	Lifetime risk of autosomal recessive mitochondrial disorders calculated from genetic databases. <i>EBioMedicine</i> , 2020, 54, 102730.	6.1	35
56	The switch in the diagnosis of mitochondrial diseases from the classical 'function first' to the NGS-based 'genetics first' diagnostic era. <i>Medycyna Wieku Rozwojowego</i> , 2020, 24, 47-52.	0.2	0
57	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. <i>American Journal of Human Genetics</i> , 2019, 105, 302-316.	6.2	56
58	Biallelic DMXL2 mutations impair autophagy and cause Ohtahara syndrome with progressive course. <i>Brain</i> , 2019, 142, 3876-3891.	7.6	23
59	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. <i>Brain</i> , 2019, 142, 3382-3397.	7.6	76
60	Choline-related inherited metabolic diseases – A mini review. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 237-242.	3.6	42
61	Mitochondrial DNA mutation analysis from exome sequencing – A more holistic approach in diagnostics of suspected mitochondrial disease. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 909-917.	3.6	57
62	Brain imaging in classic nonketotic hyperglycinemia: Quantitative analysis and relation to phenotype. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 438-450.	3.6	30
63	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. <i>Journal of Clinical Investigation</i> , 2019, 130, 108-125.	8.2	65
64	Molecular structural diversity of mitochondrial cardiolipins. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 4158-4163.	7.1	82
65	Next-generation metabolic screening: targeted and untargeted metabolomics for the diagnosis of inborn errors of metabolism in individual patients. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 337-353.	3.6	145
66	Transcriptomics: molecular diagnosis of inborn errors of metabolism via RNA sequencing. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 525-532.	3.6	38
67	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. <i>American Journal of Human Genetics</i> , 2018, 102, 460-467.	6.2	40
68	The role of the clinician in the multi-omics era: are you ready?. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 571-582.	3.6	55
69	Severe ichthyosis in MPDU1 – CDG. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 1293-1294.	3.6	8
70	Biallelic Mutations in SLC1A2; an Additional Mode of Inheritance for SLC1A2-Related Epilepsy. <i>Neuropediatrics</i> , 2018, 49, 059-062.	0.6	14
71	SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. <i>Journal of Medical Genetics</i> , 2018, 55, 39-47.	3.2	28
72	The genotypic and phenotypic spectrum of MTO1 deficiency. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 28-42.	1.1	24

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73	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
74	HTRA2 Defect: A Recognizable Inborn Error of Metabolism with 3-Methylglutaconic Aciduria as Discriminating Feature Characterized by Neonatal Movement Disorder and Epilepsy—Report of 11 Patients. Neuropediatrics, 2018, 49, 373-378.	0.6	21
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	P 233. Thiamine Pyrophosphokinase Deficiency due to Mutations in the TPK1 Gene: A Rare, Treatable Neurodegenerative Disorder. , 2018, 49, .		0
77	Mild orotic aciduria in <i>UMPS</i> heterozygotes: a metabolic finding without clinical consequences. Journal of Inherited Metabolic Disease, 2017, 40, 423-431.	3.6	14
78	3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. Clinica Chimica Acta, 2017, 471, 95-100.	1.1	14
79	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
80	<i>CAD</i> mutations and uridine-responsive epileptic encephalopathy. Brain, 2017, 140, 279-286.	7.6	106
81	Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain, 2017, 140, e11-e11.	7.6	57
82	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
83	Biallelic variants in <i>WARS2</i> encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Human Mutation, 2017, 38, 1786-1795.	2.5	24
84	Molecular and clinical spectra of <i>FBXL4</i> deficiency. Human Mutation, 2017, 38, 1649-1659.	2.5	41
85	Progressive deafness—dystonia due to <i>SERAC1</i> mutations: A study of 67 cases. Annals of Neurology, 2017, 82, 1004-1015.	5.3	63
86	A scoring system predicting the clinical course of <i>CLPB</i> defect based on the foetal and neonatal presentation of 31 patients. Journal of Inherited Metabolic Disease, 2017, 40, 853-860.	3.6	27
87	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. Fertility and Sterility, 2017, 108, 168-174.	1.0	42
88	Diagnosis and Management of Drooling in Children With Progressive Dystonia. Journal of Child Neurology, 2016, 31, 1220-1226.	1.4	10
89	Defining the Phenotype and Assessing Severity in Phosphoglucomutase-1 Deficiency. Journal of Pediatrics, 2016, 175, 130-136.e8.	1.8	43
90	Mudd's disease (MAT I/III deficiency): a survey of data for <i>MAT1A</i> homozygotes and compound heterozygotes. Orphanet Journal of Rare Diseases, 2015, 10, 99.	2.7	39

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91	SDHA mutations causing a multisystem mitochondrial disease: novel mutations and genetic overlap with hereditary tumors. <i>European Journal of Human Genetics</i> , 2015, 23, 202-209.	2.8	71
92	A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. <i>Neuromuscular Disorders</i> , 2015, 25, 262-267.	0.6	9
93	Inborn errors of metabolism in the biosynthesis and remodelling of phospholipids. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 99-110.	3.6	47
94	CLPB Mutations Cause 3-Methylglutaconic Aciduria, Progressive Brain Atrophy, Intellectual Disability, Congenital Neutropenia, Cataracts, Movement Disorder. <i>American Journal of Human Genetics</i> , 2015, 96, 245-257.	6.2	111
95	Whole exome sequencing of suspected mitochondrial patients in clinical practice. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 437-443.	3.6	186
96	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to <i>FBXL4</i> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 905-914.	3.6	45
97	SUCLA2 Deficiency: A Deafness-Dystonia Syndrome with Distinctive Metabolic Findings (Report of a Tj ETQq1 1 0.784314 rgBT /Over	1.5	8
98	Long Survival in Leigh Syndrome: New Cases and Review of Literature. <i>Neuropediatrics</i> , 2014, 45, 346-353.	0.6	20
99	The role of the mitochondrial ribosome in human disease: searching for mutations in 12S mitochondrial rRNA with high disruptive potential. <i>Human Molecular Genetics</i> , 2014, 23, 949-967.	2.9	35
100	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 342-352.	1.1	65
101	Leucine Loading Test is Only Discriminative for 3-Methylglutaconic Aciduria Due to AUH Defect. <i>JIMD Reports</i> , 2014, 16, 1-6.	1.5	17
102	3-Methylglutaconic aciduria lessons from 50 genes and 977 patients. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 913-921.	3.6	74
103	Inborn errors of metabolism with 3-methylglutaconic aciduria as discriminative feature: proper classification and nomenclature. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 923-928.	3.6	84
104	A Post-Hoc Comparison of the Utility of Sanger Sequencing and Exome Sequencing for the Diagnosis of Heterogeneous Diseases. <i>Human Mutation</i> , 2013, 34, 1721-1726.	2.5	303
105	Developing outcome measures for pediatric mitochondrial disorders: Which complaints and limitations are most burdensome to patients and their parents?. <i>Mitochondrion</i> , 2013, 13, 15-24.	3.4	34
106	A novel mutation in COQ2 leading to fatal infantile multisystem disease. <i>Journal of the Neurological Sciences</i> , 2013, 326, 24-28.	0.6	45
107	Mutations in the phospholipid remodeling gene SERAC1 impair mitochondrial function and intracellular cholesterol trafficking and cause dystonia and deafness. <i>Nature Genetics</i> , 2012, 44, 797-802.	21.4	175
108	Mitochondrial DNA m.3242G>A mutation, an under diagnosed cause of hypertrophic cardiomyopathy and renal tubular dysfunction?. <i>European Journal of Medical Genetics</i> , 2012, 55, 552-556.	1.3	27

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109	The 3-methylglutaconic acidurias: what's new?. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 13-22.	3.6	72
110	Common mutation in the PHKA2 gene with variable phenotype in patients with liver phosphorylase b kinase deficiency. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 691-694.	1.1	18
111	Mitochondrial dysfunction and organic aciduria in five patients carrying mutations in the Ras-MAPK pathway. <i>European Journal of Human Genetics</i> , 2011, 19, 138-144.	2.8	42
112	Mutation in mitochondrial ribosomal protein MRPS22 leads to Cornelia de Lange-like phenotype, brain abnormalities and hypertrophic cardiomyopathy. <i>European Journal of Human Genetics</i> , 2011, 19, 394-399.	2.8	90
113	Nijmegen paediatric CDG rating scale: a novel tool to assess disease progression. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 923-927.	3.6	50
114	Biochemical and genetic analysis of 3-methylglutaconic aciduria type IV: a diagnostic strategy. <i>Brain</i> , 2009, 132, 136-146.	7.6	90
115	Mitochondrial Energy Production Correlates With the Age-Related BMI. <i>Pediatric Research</i> , 2009, 65, 103-108.	2.3	32
116	Refractory severe intestinal vasculitis due to Henoch-Schönlein Purpura: successful treatment with plasmapheresis. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2006, 95, 622-623.	1.5	0
117	Dimerization of the cytokine receptors gp130 and LIFR analysed in single cells. <i>Journal of Cell Science</i> , 2005, 118, 5129-5140.	2.0	74
118	Long Term Association of the Cytokine Receptor gp130 and the Janus Kinase Jak1 Revealed by FRAP Analysis. <i>Journal of Biological Chemistry</i> , 2003, 278, 39205-39213.	3.4	46