Sven F F Garbade

List of Publications by Year
in descending order

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82 papers	3,203 citations	218381 26 h-index	174990 52 g-index
87 all docs	87 docs citations	87 times ranked	3355 citing authors

#	Article	IF	Citations
1	Gene replacement therapy with onasemnogene abeparvovec in children with spinal muscular atrophy aged 24 months or younger and bodyweight up to 15 kg: an observational cohort study. The Lancet Child and Adolescent Health, 2022, 6, 17-27.	2.7	57
2	Quantitative retrospective natural history modeling of WDR45-related developmental and epileptic encephalopathy â€“ a systematic cross-sectional analysis of 160 published cases. Autophagy, 2022, 18, 1715-1727.	4.3	5
3	Opportunities and challenges in machine learningâ€based newborn screeningâ€”A systematic literature review. JIMD Reports, 2022, 63, 250-261.	0.7	13
4	Sudden neonatal death in individuals with medium-chain acyl-coenzyme A dehydrogenase deficiency: limit of newborn screening. European Journal of Pediatrics, 2022, 181, 2415-2422.	1.3	3
5	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	1.7	5
6	How longitudinal observational studies can guide screening strategy for rare diseases. Journal of Inherited Metabolic Disease, 2022, 45, 889-901.	1.7	5
7	Unmet Needs of Parents of Children with Urea Cycle Disorders. Children, 2022, 9, 712.	0.6	4
8	Integrative Approach to Predict Severity in Nonketotic Hyperglycinemia. Annals of Neurology, 2022, 92, 292-303.	2.8	3
9	Delineating the clinical spectrum of isolated methylmalonic acidurias: <scp>cblA</scp> and mut. Journal of Inherited Metabolic Disease, 2021, 44, 193-214.	1.7	25
10	Impact of newborn screening and quality of therapy on the neurological outcome in glutaric aciduria type 1: a meta-analysis. Genetics in Medicine, 2021, 23, 13-21.	1.1	30
11	Cross-sectional quantitative analysis of the natural history of TUBA1A and TUBB2B tubulinopathies. Genetics in Medicine, 2021, 23, 516-523.	1.1	8
12	Impact of interventional and nonâ€interventional variables on anthropometric longâ€term development in glutaric aciduria type 1: A national prospective multiâ€centre study. Journal of Inherited Metabolic Disease, 2021, 44, 629-638.	1.7	13
13	Quantitative retrospective natural history modeling for orphan drug development. Journal of Inherited Metabolic Disease, 2021, 44, 99-109.	1.7	16
14	Brain <scp>MR</scp> patterns in inherited disorders of monoamine neurotransmitters: An analysis of 70 patients. Journal of Inherited Metabolic Disease, 2021, 44, 1070-1082.	1.7	13
15	Newborn screening and disease variants predict neurological outcome in isovaleric aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 857-870.	1.7	18
16	Patterns of extreme temperature-related catastrophic events in Europe including the Russian Federation: a cross-sectional analysis of the Emergency Events Database. BMJ Open, 2021, 11, e046359.	0.8	9
17	Prevalence of SARS-CoV-2 Infection in Children and Their Parents in Southwest Germany. JAMA Pediatrics, 2021, 175, 586.	3.3	124
18	Assessment of intellectual impairment, healthâ€related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the <scp>iNTD</scp> registry. Journal of Inherited Metabolic Disease, 2021, 44, 1489-1502.	1.7	7

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19	Health Outcomes of Infants with Vitamin B12 Deficiency Identified by Newborn Screening and Early Treated. Journal of Pediatrics, 2021, 235, 42-48.	0.9	17
20	Impact of glycogen storage disease type I on adult daily life: a survey. Orphanet Journal of Rare Diseases, 2021, 16, 371.	1.2	12
21	Subdural hematoma in glutaric aciduria type 1: High excreters are prone to incidental <scp>SDH</scp> despite newborn screening. Journal of Inherited Metabolic Disease, 2021, 44, 1343-1352.	1.7	6
22	The biochemical subtype is a predictor for cognitive function in glutaric aciduria type 1: a national prospective follow-up study. Scientific Reports, 2021, 11, 19300.	1.6	9
23	Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nature Communications, 2021, 12, 5529.	5.8	21
24	Differences of Phenylalanine Concentrations in Dried Blood Spots and in Plasma: Erythrocytes as a Neglected Component for This Observation. Metabolites, 2021, 11, 680.	1.3	3
25	Transmission of Severe Acute Respiratory Syndrome Coronavirus 2 in Households with Children, Southwest Germany, Mayâ€“August 2020. Emerging Infectious Diseases, 2021, 27, 3009-3019.	2.0	25
26	The Biochemical High Excreter Phenotype Is the Major Risk Factor for Cognitive Impairment in Early Diagnosed Individuals with Glutaric Aciduria Type 1. Neuropediatrics, 2021, 52, .	0.3	0
27	High throughput newborn screening for aromatic ÊŸâ€aminoâ€acid decarboxylase deficiency by analysis of concentrations of 3â€Oâ€methyldopa from dried blood spots. Journal of Inherited Metabolic Disease, 2020, 43, 602-610.	1.7	26
28	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	1.1	46
29	Long-term Outcomes of Individuals With Metabolic Diseases Identified Through Newborn Screening. Pediatrics, 2020, 146, .	1.0	37
30	Long-term effects of medical management on growth and weight in individuals with urea cycle disorders. Scientific Reports, 2020, 10, 11948.	1.6	11
31	A Global Cndp1-Knock-Out Selectively Increases Renal Carnosine and Anserine Concentrations in an Age- and Gender-Specific Manner in Mice. International Journal of Molecular Sciences, 2020, 21, 4887.	1.8	11
32	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	2.6	138
33	Semi-quantitative detection of a vanillactic acid/vanillylmandelic acid ratio in urine is a reliable diagnostic marker for aromatic L-amino acid decarboxylase deficiency. Molecular Genetics and Metabolism, 2020, 131, 163-170.	0.5	8
34	Cardiac phenotype in propionic acidemia â€“ Results of an observational monocentric study. Molecular Genetics and Metabolism, 2020, 130, 41-48.	0.5	14
35	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	1.1	14
36	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. PLoS ONE, 2020, 15, e0230898.	1.1	17

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37	Severity-adjusted evaluation of newborn screening on the metabolic disease course in individuals with cytosolic urea cycle disorders. Molecular Genetics and Metabolism, 2020, 131, 390-397.	0.5	14
38	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
39	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
40	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
41	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
42	A cross-sectional quantitative analysis of the natural history of free sialic acid storage diseaseâ€”an ultra-orphan multisystemic lysosomal storage disorder. Genetics in Medicine, 2019, 21, 347-352.	1.1	14
43	Allelic phenotype values: a model for genotype-based phenotype prediction in phenylketonuria. Genetics in Medicine, 2019, 21, 580-590.	1.1	48
44	Ultraâ€orphan lysosomal storage diseases: A crossâ€sectional quantitative analysis of the natural history of alphaâ€mannosidosis. Journal of Inherited Metabolic Disease, 2019, 42, 975-983.	1.7	26
45	Disasters in Germany and France: An Analysis of the Emergency Events Database From a Pediatric Perspective. Disaster Medicine and Public Health Preparedness, 2019, 13, 958-965.	0.7	11
46	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	1.7	26
47	Quantitative natural history characterization in a cohort of 142 published cases of patients with galactosialidosisâ€”A crossâ€sectional study. Journal of Inherited Metabolic Disease, 2019, 42, 295-302.	1.7	21
48	Impairment of cognitive function in ornithine transcarbamylase deficiency is global rather than domainâ€specific and is associated with disease onset, sex, maximum ammonium, and number of hyperammonemic events. Journal of Inherited Metabolic Disease, 2019, 42, 243-253.	1.7	15
49	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	2.8	42
50	Clinical characteristics of 248 patients with Krabbe disease: quantitative natural history modeling based on published cases. Genetics in Medicine, 2019, 21, 2208-2215.	1.1	33
51	Extended diagnosis of purine and pyrimidine disorders from urine: LC MS/MS assay development and clinical validation. PLoS ONE, 2019, 14, e0212458.	1.1	25
52	Patterns, evolution, and severity of striatal injury in insidiousâ€vs acuteâ€onset glutaric aciduria type 1. Journal of Inherited Metabolic Disease, 2019, 42, 117-127.	1.7	34
53	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	1.7	35
54	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”a successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93.	1.7	4

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55	Newborn screening: A diseaseâ€changing intervention for glutaric aciduria type 1. Annals of Neurology, 2018, 83, 970-979.	2.8	65
56	Carnosine Catalyzes the Formation of the Oligo/Polymeric Products of Methylglyoxal. Cellular Physiology and Biochemistry, 2018, 46, 713-726.	1.1	22
57	Patterns, evolution, and severity of striatal injury in insidious- versus acute-onset glutaric aciduria type 1. Journal of Inherited Metabolic Disease, 2018, , .	1.7	11
58	A cross-sectional quantitative analysis of the natural history of Farber disease: an ultra-orphan condition with rheumatologic and neurological cardinal disease features. Genetics in Medicine, 2018, 20, 524-530.	1.1	24
59	Age-Related Changes and Reference Values of Bicaudate Ratio and Sagittal Brainstem Diameters on MRI. Neuropediatrics, 2018, 49, 269-275.	0.3	14
60	Quantitative clinical characteristics of 53 patients with MPS VII: a cross-sectional analysis. Genetics in Medicine, 2017, 19, 983-988.	1.1	42
61	Issues with European guidelines for phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 681-683.	5.5	26
62	Incidence, disease onset and short-term outcome in urea cycle disorders â€“cross-border surveillance in Germany, Austria and Switzerland. Orphanet Journal of Rare Diseases, 2017, 12, 111.	1.2	43
63	Relationships between transformational leadership and health: The mediating role of perceived job demands and occupational self-efficacy.. International Journal of Stress Management, 2017, 24, 34-61.	0.9	40
64	Blood Trimethylamine-N-Oxide Originates from Microbiota Mediated Breakdown of Phosphatidylcholine and Absorption from Small Intestine. PLoS ONE, 2017, 12, e0170742.	1.1	40
65	Clinical relevance of the effects of reach-to-grasp training using trunk restraint in individuals with hemiparesis poststroke: A systematic review. Journal of Rehabilitation Medicine, 2016, 48, 405-416.	0.8	22
66	A cross-sectional controlled developmental study of neuropsychological functions in patients with glutaric aciduria type I. Orphanet Journal of Rare Diseases, 2015, 10, 163.	1.2	10
67	The Relationship Between Self-Compassion and Well-Being: A Meta-Analysis. Applied Psychology: Health and Well-Being, 2015, 7, 340-364.	1.6	635
68	Unravelling the complex MRI pattern in glutaric aciduria type I using statistical modelsâ€”a cohort study in 180 patients. Journal of Inherited Metabolic Disease, 2014, 37, 763-773.	1.7	30
69	Dynamic changes of striatal and extrastriatal abnormalities in glutaric aciduria type I. Brain, 2009, 132, 1764-1782.	3.7	160
70	Pharmacokinetics of tetrahydrobiopterin following oral loadings with three single dosages in patients with phenylketonuria. Journal of Inherited Metabolic Disease, 2009, 32, 52-57.	1.7	13
71	Blood phenylalanine concentrations in patients with PAHâ€deficient hyperphenylalaninaemia off diet without and with three different single oral doses of tetrahydrobiopterin: Assessing responsiveness in a model of statistical process control. Journal of Inherited Metabolic Disease, 2009, 32, 514-522.	1.7	9
72	Prediction of outcome in isolated methylmalonic acidurias: combined use of clinical and biochemical parameters. Journal of Inherited Metabolic Disease, 2009, 32, 630-639.	1.7	65

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73	Neuropsychological speed tests and blood phenylalanine levels in patients with phenylketonuria: A meta-analysis. Neuroscience and Biobehavioral Reviews, 2009, 33, 414-421.	2.9	91
74	Qualitative urinary organic acid analysis: Methodological approaches and performance. Journal of Inherited Metabolic Disease, 2008, 31, 690-696.	1.7	14
75	Decline of Acute Encephalopathic Crises in Children with Glutaryl-CoA Dehydrogenase Deficiency Identified by Newborn Screening in Germany. Pediatric Research, 2007, 62, 357-363.	1.1	102
76	Response. Pediatric Research, 2007, 61, 134-135.	1.1	0
77	Abnormal sterol metabolism in holoprosencephaly: studies in cultured lymphoblasts. Journal of Medical Genetics, 2007, 44, 298-305.	1.5	25
78	Long-Term Outcome in Methylmalonic Acidurias Is Influenced by the Underlying Defect (mut0, mutâ˚,) Tj ETQq0 0 0 rgBT /Overlock 10 T	1.1	210
79	Effects of cholesterol and simvastatin treatment in patients with Smithâ€“Lemliâ€“Opitz syndrome (SLOS). Journal of Inherited Metabolic Disease, 2007, 30, 375-387.	1.7	62
80	Effects and clinical significance of tetrahydrobiopterin supplementation in phenylalanine hydroxylaseâ€deficient hyperphenylalaninaemia. Journal of Inherited Metabolic Disease, 2007, 30, 556-562.	1.7	11
81	Comprehensive Detection of Disorders of Purine and Pyrimidine Metabolism by HPLC with Electrospray Ionization Tandem Mass Spectrometry. Clinical Chemistry, 2006, 52, 1127-1137.	1.5	65
82	Natural History, Outcome, and Treatment Efficacy in Children and Adults with Glutaryl-CoA Dehydrogenase Deficiency. Pediatric Research, 2006, 59, 840-847.	1.1	224