

Katrin Hinderhofer

List of Publications by Year in descending order

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Version: 2024-02-01

38
papers

1,188
citations

567281

15
h-index

395702

33
g-index

39
all docs

39
docs citations

39
times ranked

2593
citing authors

#	ARTICLE	IF	CITATIONS
1	BMPR2 mutations and survival in pulmonary arterial hypertension: an individual participant data meta-analysis. <i>Lancet Respiratory Medicine</i> , 2016, 4, 129-137.	10.7	307
2	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. <i>Human Mutation</i> , 2015, 36, 1113-1127.	2.5	185
3	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. <i>American Journal of Human Genetics</i> , 2017, 101, 768-788.	6.2	136
4	Noninvasive Risk Stratification of Patients With Transthyretin Amyloidosis. <i>JACC: Cardiovascular Imaging</i> , 2014, 7, 502-510.	5.3	54
5	<i>DDX3X</i> mutations in two girls with a phenotype overlapping Toriello's "Carey syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1369-1373.	1.2	41
6	Diagnosis of CoPAN by whole exome sequencing: Waking up a sleeping tiger's eye. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 1878-1886.	1.2	40
7	First identification of <i>APP</i> -like factor 2 mutation in heritable pulmonary arterial hypertension. <i>Clinical Science</i> , 2017, 131, 689-698.	4.3	38
8	EIF2AK4 mutation as a "second hit" in hereditary pulmonary arterial hypertension. <i>Respiratory Research</i> , 2016, 17, 141.	3.6	33
9	Expanding Phenotype of De Novo Mutations in GNAO1: Four New Cases and Review of Literature. <i>Neuropediatrics</i> , 2017, 48, 371-377.	0.6	33
10	Mutation in BMPR2 Promoter: A "Second Hit"™ for Manifestation of Pulmonary Arterial Hypertension?. <i>PLoS ONE</i> , 2015, 10, e0133042.	2.5	26
11	Identification of genetic defects in pulmonary arterial hypertension by a new gene panel diagnostic tool. <i>Clinical Science</i> , 2016, 130, 2043-2052.	4.3	25
12	NADPH oxidase subunit NOXO1 is a target for emphysema treatment in COPD. <i>Nature Metabolism</i> , 2020, 2, 532-546.	11.9	23
13	Genetic Predisposition to High-Altitude Pulmonary Edema. <i>High Altitude Medicine and Biology</i> , 2020, 21, 28-36.	0.9	21
14	FMR1 and AKT/mTOR signalling pathways: potential functional interactions controlling folliculogenesis in human granulosa cells. <i>Reproductive BioMedicine Online</i> , 2017, 35, 485-493.	2.4	18
15	Gene panel diagnostics reveals new pathogenic variants in pulmonary arterial hypertension. <i>Respiratory Research</i> , 2022, 23, 74.	3.6	18
16	FMR1 expression in human granulosa cells increases with exon 1 CGG repeat length depending on ovarian reserve. <i>Reproductive Biology and Endocrinology</i> , 2018, 16, 65.	3.3	16
17	Quantitative retrospective natural history modeling for orphan drug development. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 99-109.	3.6	16
18	SIPA1L3 identified by linkage analysis and whole-exome sequencing as a novel gene for autosomal recessive congenital cataract. <i>European Journal of Human Genetics</i> , 2015, 23, 1627-1633.	2.8	15

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19	Exome sequencing reveals a novel <i>CWF19L1</i> mutation associated with intellectual disability and cerebellar atrophy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1502-1509.	1.2	13
20	Critical appraisal of genotype assessment in molybdenum cofactor deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 801-811.	3.6	13
21	The Frog <i>Xenopus</i> as a Model to Study Joubert Syndrome: The Case of a Human Patient With Compound Heterozygous Variants in <i>PIBF1</i> . <i>Frontiers in Physiology</i> , 2019, 10, 134.	2.8	13
22	Myeloproliferative Diseases as Possible Risk Factor for Development of Chronic Thromboembolic Pulmonary Hypertension—A Genetic Study. <i>International Journal of Molecular Sciences</i> , 2020, 21, 3339.	4.1	13
23	Disruption of <i>SOX6</i> Is Associated With a Rapid-Onset Dopa-Responsive Movement Disorder, Delayed Development, and Dysmorphic Features. <i>Pediatric Neurology</i> , 2015, 52, 115-118.	2.1	12
24	Characterization of the first intragenic <i>SATB2</i> duplication in a girl with intellectual disability, nearly absent speech and suspected hypodontia. <i>European Journal of Human Genetics</i> , 2015, 23, 704-707.	2.8	10
25	Activation of AKT/mammalian target of rapamycin signaling in the peripheral blood of women with premature ovarian insufficiency and its correlation with <i>FMR1</i> expression. <i>Reproductive Biology and Endocrinology</i> , 2022, 20, 44.	3.3	10
26	Clinical Research in Vulnerable Populations: Variability and Focus of Institutional Review Boards™ Responses. <i>PLoS ONE</i> , 2015, 10, e0135997.	2.5	9
27	Mutually reinforcing effects of genetic variants and interferon γ 1a therapy for pulmonary arterial hypertension development in multiple sclerosis patients. <i>Pulmonary Circulation</i> , 2019, 9, 1-6.	1.7	9
28	Real-world outcomes in non-endemic hereditary transthyretin amyloidosis with polyneuropathy: a 20-year German single-referral centre experience. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2021, 28, 91-99.	3.0	8
29	<i>KIT</i> Mutation and Loss of 14q May Be Sufficient for the Development of Clinically Symptomatic Very Low-Risk GIST. <i>PLoS ONE</i> , 2015, 10, e0130149.	2.5	6
30	New sequence variants in patients affected by amyloidosis show transthyretin instability by isoelectric focusing. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2019, 26, 85-93.	3.0	6
31	Evidence That Non-Syndromic Familial Tall Stature Has an Oligogenic Origin Including Ciliary Genes. <i>Frontiers in Endocrinology</i> , 2021, 12, 660731.	3.5	5
32	Lysozyme amyloidosis—a report on a large German cohort and the characterisation of a novel amyloidogenic lysozyme gene variant. <i>Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis</i> , 2022, 29, 245-254.	3.0	5
33	Ornithine transcarbamylase deficiency of a male newborn with fatal outcome. <i>International Journal of Legal Medicine</i> , 2016, 130, 783-785.	2.2	3
34	<i>BMPR2</i> Promoter Variants Effect Gene Expression in Pulmonary Arterial Hypertension Patients. <i>Genes</i> , 2020, 11, 1168.	2.4	3
35	Germ cell mosaicism for <i>AUTS2</i> exon 6 deletion. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1261-1265.	1.2	2
36	Reduction of <i>BMPR2</i> mRNA Expression in Peripheral Blood of Pulmonary Arterial Hypertension Patients: A Marker for Disease Severity?. <i>Genes</i> , 2022, 13, 759.	2.4	2

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37	Expression of FMRpolyG in Peripheral Blood Mononuclear Cells of Women with Fragile X Mental Retardation 1 Gene Premutation. <i>Genes</i> , 2022, 13, 451.	2.4	1
38	A boy with <scp>Silver</scp>â€“<scp>Russell</scp> syndrome and Sotos syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 549-554.	1.2	0