Katrin Hinderhofer

List of Publications by Year
in descending order

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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	Gene panel diagnostics reveals new pathogenic variants in pulmonary arterial hypertension. Respiratory Research, 2022, 23, 74.	3.6	18
2	Expression of FMRpolyG in Peripheral Blood Mononuclear Cells of Women with Fragile X Mental Retardation 1 Gene Premutation. Genes, 2022, 13, 451.	2.4	1
3	Activation of AKT/mammalian target of rapamycin signaling in the peripheral blood of women with premature ovarian insufficiency and its correlation with FMR1 expression. Reproductive Biology and Endocrinology, 2022, 20, 44.	3.3	10
4	Reduction of BMPR2 mRNA Expression in Peripheral Blood of Pulmonary Arterial Hypertension Patients: A Marker for Disease Severity?. Genes, 2022, 13, 759.	2.4	2
5	Lysozyme amyloidosisâ€"a report on a large German cohort and the characterisation of a novel amyloidogenic lysozyme gene variant. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2022, 29, 245-254.	3.0	5
6	A boy with <scp>Silver</scp>â€"<scp>Russell</scp> syndrome and Sotos syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 549-554.	1.2	0
7	Real-world outcomes in non-endemic hereditary transthyretin amyloidosis with polyneuropathy: a 20-year German single-referral centre experience. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2021, 28, 91-99.	3.0	8
8	Quantitative retrospective natural history modeling for orphan drug development. Journal of Inherited Metabolic Disease, 2021, 44, 99-109.	3.6	16
9	Germ cell mosaicism for AUTS2 exon 6 deletion. American Journal of Medical Genetics, Part A, 2021, 185, 1261-1265.	1.2	2
10	Evidence That Non-Syndromic Familial Tall Stature Has an Oligogenic Origin Including Ciliary Genes. Frontiers in Endocrinology, 2021, 12, 660731.	3.5	5
11	BMPR2 Promoter Variants Effect Gene Expression in Pulmonary Arterial Hypertension Patients. Genes, 2020, 11, 1168.	2.4	3
12	Myeloproliferative Diseases as Possible Risk Factor for Development of Chronic Thromboembolic Pulmonary Hypertensionâ€"A Genetic Study. International Journal of Molecular Sciences, 2020, 21, 3339.	4.1	13
13	NADPH oxidase subunit NOXO1 is a target for emphysema treatment in COPD. Nature Metabolism, 2020, 2, 532-546.	11.9	23
14	Genetic Predisposition to High-Altitude Pulmonary Edema. High Altitude Medicine and Biology, 2020, 21, 28-36.	0.9	21
15	New sequence variants in patients affected by amyloidosis show transthyretin instability by isoelectric focusing. Amyloid: the International Journal of Experimental and Clinical Investigation: the Official Journal of the International Society of Amyloidosis, 2019, 26, 85-93.	3.0	6
16	The Frog Xenopus as a Model to Study Joubert Syndrome: The Case of a Human Patient With Compound Heterozygous Variants in PIBF1. Frontiers in Physiology, 2019, 10, 134.	2.8	13
17	Mutually reinforcing effects of genetic variants and interferonâ€Î²2 1a therapy for pulmonary arterial hypertension development in multiple sclerosis patients. Pulmonary Circulation, 2019, 9, 1-6.	1.7	9
18	FMR1 expression in human granulosa cells increases with exon 1 CGG repeat length depending on ovarian reserve. Reproductive Biology and Endocrinology, 2018, 16, 65.	3.3	16

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19	First identification of KrÃ¼ppel-like factor 2 mutation in heritable pulmonary arterial hypertension. Clinical Science, 2017, 131, 689-698.	4.3	38
20	Diagnosis of CoPAN by whole exome sequencing: Waking up a sleeping tiger's eye. American Journal of Medical Genetics, Part A, 2017, 173, 1878-1886.	1.2	40
21	Expanding Phenotype of De Novo Mutations in GNAO1: Four New Cases and Review of Literature. Neuropediatrics, 2017, 48, 371-377.	0.6	33
22	DDX3X mutations in two girls with a phenotype overlapping Toriello–Carey syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 1369-1373.	1.2	41
23	Critical appraisal of genotype assessment in molybdenum cofactor deficiency. Journal of Inherited Metabolic Disease, 2017, 40, 801-811.	3.6	13
24	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
25	FMR1 and AKT/mTOR signalling pathways: potential functional interactions controlling folliculogenesis in human granulosa cells. Reproductive BioMedicine Online, 2017, 35, 485-493.	2.4	18
26	BMPR2 mutations and survival in pulmonary arterial hypertension: an individual participant data meta-analysis. Lancet Respiratory Medicine,the, 2016, 4, 129-137.	10.7	307
27	Identification of genetic defects in pulmonary arterial hypertension by a new gene panel diagnostic tool. Clinical Science, 2016, 130, 2043-2052.	4.3	25
28	EIF2AK4 mutation as â€œsecond hitâ€•in hereditary pulmonary arterial hypertension. Respiratory Research, 2016, 17, 141.	3.6	33
29	Exome sequencing reveals a novel CWF19L1 mutation associated with intellectual disability and cerebellar atrophy. American Journal of Medical Genetics, Part A, 2016, 170, 1502-1509.	1.2	13
30	Ornithine transcarbamylase deficiency of a male newborn with fatal outcome. International Journal of Legal Medicine, 2016, 130, 783-785.	2.2	3
31	Pulmonary Arterial Hypertension: A Current Perspective on Established and Emerging Molecular Genetic Defects. Human Mutation, 2015, 36, 1113-1127.	2.5	185
32	KIT Mutation and Loss of 14q May Be Sufficient for the Development of Clinically Symptomatic Very Low-Risk GIST. PLoS ONE, 2015, 10, e0130149.	2.5	6
33	Clinical Research in Vulnerable Populations: Variability and Focus of Institutional Review Boardsâ€™ Responses. PLoS ONE, 2015, 10, e0135997.	2.5	9
34	SIPA1L3 identified by linkage analysis and whole-exome sequencing as a novel gene for autosomal recessive congenital cataract. European Journal of Human Genetics, 2015, 23, 1627-1633.	2.8	15
35	Disruption of SOX6 Is Associated With a Rapid-Onset Dopa-Responsive Movement Disorder, Delayed Development, andÂDysmorphic Features. Pediatric Neurology, 2015, 52, 115-118.	2.1	12
36	Characterization of the first intragenic SATB2 duplication in a girl with intellectual disability, nearly absent speech and suspected hypodontia. European Journal of Human Genetics, 2015, 23, 704-707.	2.8	10

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37	Mutation in BMPR2 Promoter: A â€˜Second Hitâ€™ for Manifestation of Pulmonary Arterial Hypertension?. PLoS ONE, 2015, 10, e0133042.	2.5	26
38	Noninvasive Risk Stratification of Patients With Transthyretin Amyloidosis. JACC: Cardiovascular Imaging, 2014, 7, 502-510.	5.3	54