Katharina Steindl

List of Publications by Year
in descending order

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51 papers	1,481 citations	394421 19 h-index	377865 34 g-index
53 all docs	53 docs citations	53 times ranked	3578 citing authors

#	Article	IF	Citations
1	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
2	YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction. American Journal of Human Genetics, 2017, 100, 907-925.	6.2	125
3	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	3.2	72
4	Biallelic UFM1 and UFC1 mutations expand the essential role of ufmylation in brain development. Brain, 2018, 141, 1934-1945.	7.6	70
5	FOXP2variants in 14 individuals with developmental speech and language disorders broaden the mutational and clinical spectrum. Journal of Medical Genetics, 2017, 54, 64-72.	3.2	67
6	Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly. Genetics in Medicine, 2019, 21, 2043-2058.	2.4	57
7	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
8	The role of recessive inheritance in early-onset epileptic encephalopathies: a combined whole-exome sequencing and copy number study. European Journal of Human Genetics, 2019, 27, 408-421.	2.8	52
9	ZC4H2, an XLID gene, is required for the generation of a specific subset of CNS interneurons. Human Molecular Genetics, 2015, 24, 4848-4861.	2.9	48
10	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
11	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	6.2	48
12	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759.	2.8	47
13	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. American Journal of Human Genetics, 2017, 101, 985-994.	6.2	44
14	Characterization of SETD1A haploinsufficiency in humans and Drosophila defines a novel neurodevelopmental syndrome. Molecular Psychiatry, 2021, 26, 2013-2024.	7.9	43
15	Further corroboration of distinct functional features in SCN2A variants causing intellectual disability or epileptic phenotypes. Molecular Medicine, 2019, 25, 6.	4.4	42
16	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
17	A recurrent germline mutation in the PIGA gene causes Simpsonâ€Golabiâ€Behmel syndrome type 2. American Journal of Medical Genetics, Part A, 2016, 170, 392-402.	1.2	34
18	Genotype-phenotype evaluation of MED13L defects in the light of a novel truncating and a recurrent missense mutation. European Journal of Medical Genetics, 2017, 60, 451-464.	1.3	34

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19	New insights into the clinical and molecular spectrum of the novel CYFIP2-related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. Genetics in Medicine, 2021, 23, 543-554.	2.4	32
20	Mutations in XRCC4 cause primary microcephaly, short stature and increased genomic instability. Human Molecular Genetics, 2015, 24, 3708-17.	2.9	26
21	Loss-of-function and missense variants in NSD2 cause decreased methylation activity and are associated with a distinct developmental phenotype. Genetics in Medicine, 2021, 23, 1474-1483.	2.4	24
22	Clinical and experimental evidence suggest a link between KIF7 and C5orf42-related ciliopathies through Sonic Hedgehog signaling. European Journal of Human Genetics, 2018, 26, 197-209.	2.8	23
23	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	2.4	23
24	A severe congenital myasthenic syndrome with â€œdropped headâ€•caused by novel MUSK mutations. Muscle and Nerve, 2015, 52, 668-673.	2.2	21
25	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	6.2	18
26	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	3.8	18
27	Genetic Analysis in a Swiss Cohort of Bilateral Congenital Cataract. JAMA Ophthalmology, 2021, 139, 691.	2.5	18
28	CDK5RAP2 primary microcephaly is associated with hypothalamic, retinal and cochlear developmental defects. Journal of Medical Genetics, 2020, 57, 389-399.	3.2	17
29	Swiss newborn screening for severe T and B cell deficiency with a combined TREC/KREC assay â€“ management recommendations. Swiss Medical Weekly, 2020, 150, w20254.	1.6	17
30	Whole Exome Sequencing in Coloboma/Microphthalmia: Identification of Novel and Recurrent Variants in Seven Genes. Genes, 2021, 12, 65.	2.4	16
31	Noninvasive prenatal testing: more caution in counseling is needed in high risk pregnancies with ultrasound abnormalities. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2016, 200, 72-75.	1.1	15
32	Exome Sequencing in a Swiss Childhood Glaucoma Cohort Reveals CYP1B1 and FOXC1 Variants as Most Frequent Causes. Translational Vision Science and Technology, 2020, 9, 47.	2.2	15
33	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
34	Genotypeâ€“phenotype spectrum in isolated and syndromic nanophthalmos. Acta Ophthalmologica, 2021, 99, e594-e607.	1.1	13
35	CSNK2B: A broad spectrum of neurodevelopmental disability and epilepsy severity. Epilepsia, 2021, 62, e103-e109.	5.1	13
36	Confirmation of Ogden syndrome as an Xâ€linked recessive fatal disorder due to a recurrent <scp>NAA10</scp> variant and review of the literature. American Journal of Medical Genetics, Part A, 2021, 185, 2546-2560.	1.2	12

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37	Clinical and functional characterization of two novel ZBTB20 mutations causing Primrose syndrome. Human Mutation, 2018, 39, 959-964.	2.5	11
38	Bi-allelic Pathogenic Variants in HS2ST1 Cause a Syndrome Characterized by Developmental Delay and Corpus Callosum, Skeletal, and Renal Abnormalities. American Journal of Human Genetics, 2020, 107, 1044-1061.	6.2	11
39	Expanding the phenotype: Four new cases and hope for treatment in <scp>Bachmannâ€Bupp</scp> syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3485-3493.	1.2	10
40	Bi-allelic loss of function variants in SLC30A5 as cause of perinatal lethal cardiomyopathy. European Journal of Human Genetics, 2021, 29, 808-815.	2.8	9
41	Human COQ4 deficiency: delineating the clinical, metabolic and neuroimaging phenotypes. Journal of Medical Genetics, 2022, 59, 878-887.	3.2	9
42	Genome-wide non-invasive prenatal testing in single- and multiple-pregnancies at any risk: Identification of maternal polymorphisms to reduce the number of unnecessary invasive confirmation testing. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2020, 252, 19-29.	1.1	7
43	Insight Into the Ontogeny of GnRH Neurons From Patients Born Without a Nose. Journal of Clinical Endocrinology and Metabolism, 2020, 105, 1538-1551.	3.6	7
44	Rare variants in KDR, encoding VEGF Receptor 2, are associated with tetralogy of Fallot. Genetics in Medicine, 2021, 23, 1952-1960.	2.4	7
45	Further characterization of <scp>Borjesonâ€Forssmanâ€Lehmann</scp> syndrome in females due to de novo variants in <scp>PHF6</scp>. Clinical Genetics, 2022, 102, 182-190.	2.0	5
46	Sotos syndrome: a pitfall in the presurgical workup of temporal lobe epilepsy. Epileptic Disorders, 2021, 23, 506-510.	1.3	4
47	Cancer in children with biallelic BRCA1 variants and Fanconi anemiaâ€like features: Report of a malignant brain tumor in a young child. Pediatric Blood and Cancer, 2022, 69, e29680.	1.5	2
48	Periorbital pigmented skin tags and milia. Pediatric Dermatology, 2020, 37, 740-741.	0.9	1
49	Prevalence of genetic susceptibility for breast and ovarian cancer in a non-cancer related study population: secondary germline findings from a Swiss single centre cohort. Swiss Medical Weekly, 2019, 149, w20092.	1.6	1
50	Lissencephaly with Brainstem Hypoplasia and Dysplasia: Think MACF1. Neuropediatrics, 2021, 52, 227.	0.6	0
51	Response to Cueto-GonzÃ¡lez etÂal. Genetics in Medicine, 2022, 24, 757.	2.4	0