Stephanie L Bielas

List of Publications by Year
in descending order

46	2,945	304602	233338
papers	citations	22 h-index	45 g-index

48	48	48	5524
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Homozygous variant p.(Arg163Trp) in PIGH causes glycosylphosphatidylinositol biosynthesis defect with epileptic encephalopathy and delayed myelination. Clinical Dysmorphology, 2022, Publish Ahead of Print, .	0.1	0
2	Molecular and functional heterogeneity in dorsal and ventral oligodendrocyte progenitor cells of the mouse forebrain in response to DNA damage. Nature Communications, 2022, 13, 2331.	5.8	5
3	A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for earlyâ€onset monogenic disorders in Indians. Human Mutation, 2021, 42, e15-e61.	1.1	25
4	Myofibrillar Structural Variability Underlies Contractile Function in Stem Cell-Derived Cardiomyocytes. Stem Cell Reports, 2021, 16, 470-477.	2.3	7
5	Clinical and genetic spectrum of 104 Indian families with central nervous system white matter abnormalities. Clinical Genetics, 2021, 100, 542-550.	1.0	12
6	Multilocus disease-causing genomic variations for Mendelian disorders: role of systematic phenotyping and implications on genetic counselling. European Journal of Human Genetics, 2021, 29, 1774-1780.	1.4	7
7	Biâ€allelic missense variant, p.<scp>Ser35Leu</scp> in <scp>EXOSC1</scp> is associated with pontocerebellar hypoplasia. Clinical Genetics, 2021, 99, 594-600.	1.0	16
8	Genotypeâ€“phenotype analysis of 523 patients by genetics evaluation and clinical exome sequencing. Pediatric Research, 2020, 87, 735-739.	1.1	28
9	Bain type of Xâ€linked syndromic mental retardation in a male with a pathogenic variant in HNRNPH2. American Journal of Medical Genetics, Part A, 2020, 182, 183-188.	0.7	14
10	CHD7 promotes neural progenitor differentiation in embryonic stem cells via altered chromatin accessibility and nascent gene expression. Scientific Reports, 2020, 10, 17445.	1.6	23
11	Recurrent 1q21.1 deletion syndrome: report on variable expression, nonpenetrance and review of literature. Clinical Dysmorphology, 2020, 29, 127-131.	0.1	5
12	Modeling Bainbridge-Ropers Syndrome in Xenopus laevis Embryos. Frontiers in Physiology, 2020, 11, 75.	1.3	14
13	Histone Acetyltransferase MOF Blocks Acquisition of Quiescence in Ground-State ESCs through Activating Fatty Acid Oxidation. Cell Stem Cell, 2020, 27, 441-458.e10.	5.2	37
14	Bi-allelic c.181_183delTGT in BTB domain of KLHL7 is associated with overlapping phenotypes of Crisponi/CISS1-like and Bohring-Opitz like syndrome. European Journal of Medical Genetics, 2019, 62, 103528.	0.7	7
15	Locus and allelic heterogeneity in five families with hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 17-21.	1.1	7
16	A biallelic 36-bp insertion in PIBF1 is associated with Joubert syndrome. Journal of Human Genetics, 2018, 63, 935-939.	1.1	14
17	Genotype-phenotype correlations in individuals with pathogenicREREvariants. Human Mutation, 2018, 39, 666-675.	1.1	34
18	Report of four novel variants in ASNS causing asparagine synthetase deficiency and review of literature. Congenital Anomalies (discontinued), 2018, 58, 181-182.	0.3	15

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19	Dysregulation of cotranscriptional alternative splicing underlies CHARGE syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, E620-E629.	3.3	28
20	Genetic analysis of CHARGE syndrome identifies overlapping molecular biology. Genetics in Medicine, 2018, 20, 1022-1029.	1.1	43
21	Genetic diversity of NDUFV1-dependent mitochondrial complex I deficiency. European Journal of Human Genetics, 2018, 26, 1582-1587.	1.4	15
22	Photosensitivity and type I IFN responses in cutaneous lupus are driven by epidermal-derived interferon kappa. Annals of the Rheumatic Diseases, 2018, 77, 1653-1664.	0.5	162
23	Homozygous mutation inNUP107leads to microcephaly with steroid-resistant nephrotic condition similar to Galloway-Mowat syndrome. Journal of Medical Genetics, 2017, 54, 399-403.	1.5	62
24	Autosomal recessive spinocerebellar ataxia 20: Report of a new patient and review of literature. European Journal of Medical Genetics, 2017, 60, 118-123.	0.7	29
25	Homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. Journal of Human Genetics, 2017, 62, 723-727.	1.1	53
26	Homozygous c.359del variant in MGME1 is associated with early onset cerebellar ataxia. European Journal of Medical Genetics, 2017, 60, 533-535.	0.7	8
27	Hyperphosphatasia with Mental Retardation Syndrome Due to a Novel Mutation in PGAP3. Journal of Pediatric Genetics, 2017, 06, 191-193.	0.3	10
28	Histone H2A Monoubiquitination in Neurodevelopmental Disorders. Trends in Genetics, 2017, 33, 566-578.	2.9	19
29	Homozygous deletion of exons 2 and 3 of NPC2 associated with Niemannâ€“Pick disease type C. American Journal of Medical Genetics, Part A, 2016, 170, 2486-2489.	0.7	5
30	Mutations in Citron Kinase Cause Recessive Microlissencephaly with Multinucleated Neurons. American Journal of Human Genetics, 2016, 99, 511-520.	2.6	59
31	Biallelic Mutations in Citron Kinase Link Mitotic Cytokinesis to Human Primary Microcephaly. American Journal of Human Genetics, 2016, 99, 501-510.	2.6	70
32	De novodominantASXL3mutations alter H2A deubiquitination and transcription in Bainbridgeâ€“Ropers syndrome. Human Molecular Genetics, 2016, 25, 597-608.	1.4	56
33	Off-Target Effect of doublecortin Family shRNA on Neuronal Migration Associated with Endogenous MicroRNA Dysregulation. Neuron, 2014, 82, 1255-1262.	3.8	79
34	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	6.0	84
35	Exome Sequencing Can Improve Diagnosis and Alter Patient Management. Science Translational Medicine, 2012, 4, 138ra78.	5.8	226
36	CEP41 is mutated in Joubert syndrome and is required for tubulin glutamylation at the cilium. Nature Genetics, 2012, 44, 193-199.	9.4	157

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37	Expanding the clinical spectrum of SPG11 gene mutations in recessive hereditary spastic paraplegia with thin corpus callosum. European Journal of Medical Genetics, 2011, 54, 82-85.	0.7	9
38	SRD5A3 Is Required for Converting Polyprenol to Dolichol and Is Mutated in a Congenital Glycosylation Disorder. Cell, 2010, 142, 203-217.	13.5	253
39	Mutations in INPP5E, encoding inositol polyphosphate-5-phosphatase E, link phosphatidyl inositol signaling to the ciliopathies. Nature Genetics, 2009, 41, 1032-1036.	9.4	383
40	Mutations in the Cilia Gene ARL13B Lead to the Classical Form of Joubert Syndrome. American Journal of Human Genetics, 2008, 83, 170-179.	2.6	352
41	Spinophilin Facilitates Dephosphorylation of Doublecortin by PP1 to Mediate Microtubule Bundling at the Axonal Wrist. Cell, 2007, 129, 579-591.	13.5	133
42	Spinophilin Facilitates Dephosphorylation of Doublecortin by PP1 to Mediate Microtubule Bundling at the Axonal Wrist. Cell, 2007, 129, 1227-1228.	13.5	2
43	CEP290 Mutations Are Frequently Identified in the Oculo-Renal Form of Joubert Syndromeâ€“Related Disorders. American Journal of Human Genetics, 2007, 81, 104-113.	2.6	137
44	Transgenic Mouse Line with Green-fluorescent Protein-labeled Centrin 2 allows Visualization of the Centrosome in Living Cells. Transgenic Research, 2004, 13, 155-164.	1.3	97
45	Cytoskeletal-associated proteins in the migration of cortical neurons. Journal of Neurobiology, 2004, 58, 149-159.	3.7	24
46	CORTICAL NEURONAL MIGRATION MUTANTS SUGGEST SEPARATE BUT INTERSECTING PATHWAYS. Annual Review of Cell and Developmental Biology, 2004, 20, 593-618.	4.0	118