Siddharth Banka

List of Publications by Year
in descending order

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101 papers	5,505 citations	101543 36 h-index	102487 66 g-index
114 all docs	114 docs citations	114 times ranked	9525 citing authors

#	Article	IF	Citations
1	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
2	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. Nature Genetics, 2016, 48, 1060-1065.	21.4	351
3	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
4	Whole-genome sequencing of patients with rare diseases in a national health system. Nature, 2020, 583, 96-102.	27.8	338
5	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. Immunity, 2015, 42, 1185-1196.	14.3	246
6	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
7	Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1511-1516.	1.2	160
8	Kabuki syndrome: international consensus diagnostic criteria. Journal of Medical Genetics, 2019, 56, 89-95.	3.2	146
9	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. Oncotarget, 2011, 2, 1127-1133.	1.8	145
10	How genetically heterogeneous is Kabuki syndrome?: MLL2 testing in 116 patients, review and analyses of mutation and phenotypic spectrum. European Journal of Human Genetics, 2012, 20, 381-388.	2.8	142
11	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. American Journal of Medical Genetics, Part A, 2013, 161, 2972-2980.	1.2	119
12	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. American Journal of Human Genetics, 2017, 101, 466-477.	6.2	119
13	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108
14	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. American Journal of Human Genetics, 2011, 88, 767-777.	6.2	106
15	Novel <scp>KDM6A</scp> (<scp>UTX</scp>) mutations and a clinical and molecular review of the Xâ€linked Kabuki syndrome (<scp>KS2</scp>). Clinical Genetics, 2015, 87, 252-258.	2.0	102
16	TBC1D24 genotypeâ€"phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
17	Identification and Characterization of an Inborn Error of Metabolism Caused by Dihydrofolate Reductase Deficiency. American Journal of Human Genetics, 2011, 88, 216-225.	6.2	90
18	Corneal confocal microscopy detects smallâ€fiber neuropathy in Charcotâ€"Marieâ€"Tooth disease type 1A patients. Muscle and Nerve, 2012, 46, 698-704.	2.2	89

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19	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
20	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
21	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. American Journal of Human Genetics, 2016, 98, 981-992.	6.2	81
22	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. Brain, 2019, 142, 3382-3397.	7.6	76
23	A basement membrane discovery pipeline uncovers network complexity, regulators, and human disease associations. Science Advances, 2022, 8, eabn2265.	10.3	76
24	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. Journal of Clinical Investigation, 2015, 125, 3585-3599.	8.2	69
25	Extended Spectrum of Human Glucose-6-Phosphatase Catalytic Subunit 3 Deficiency: Novel Genotypes and Phenotypic Variability in Severe Congenital Neutropenia. Journal of Pediatrics, 2012, 160, 679-683.e2.	1.8	67
26	A clinical and molecular review of ubiquitous glucose-6-phosphatase deficiency caused by G6PC3 mutations. Orphanet Journal of Rare Diseases, 2013, 8, 84.	2.7	67
27	Exome sequencing identifies ATP4A gene as responsible of an atypical familial type I gastric neuroendocrine tumour. Human Molecular Genetics, 2015, 24, 2914-2922.	2.9	60
28	Opposite Modulation of RAC1 by Mutations in TRIO Is Associated with Distinct, Domain-Specific Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 338-355.	6.2	58
29	Further delineation of the KAT6B molecular and phenotypic spectrum. European Journal of Human Genetics, 2015, 23, 1165-1170.	2.8	56
30	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
31	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	6.2	56
32	Further delineation of the phenotype of severe congenital neutropenia type 4 due to mutations in G6PC3. European Journal of Human Genetics, 2011, 19, 18-22.	2.8	50
33	Expanding the clinical and molecular spectrum of thiamine pyrophosphokinase deficiency: A treatable neurological disorder caused by TPK1 mutations. Molecular Genetics and Metabolism, 2014, 113, 301-306.	1.1	50
34	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
35	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
36	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45

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37	<scp>MLL2</scp> mosaic mutations and intragenic deletionâ€“duplications in patients with Kabuki syndrome. Clinical Genetics, 2013, 83, 467-471.	2.0	44
38	Diagnosing childhood-onset inborn errors of metabolism by next-generation sequencing. Archives of Disease in Childhood, 2017, 102, 1019-1029.	1.9	43
39	Non-coding region variants upstream of MEF2C cause severe developmental disorder through three distinct loss-of-function mechanisms. American Journal of Human Genetics, 2021, 108, 1083-1094.	6.2	42
40	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
41	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct fromKabuki syndrome. Genetics in Medicine, 2020, 22, 867-877.	2.4	41
42	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. Nature Communications, 2021, 12, 833.	12.8	41
43	Mutations in the G6PC3 gene cause Dursun syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 2609-2611.	1.2	37
44	Role of reverse phenotyping in interpretation of next generation sequencing data and a review of INPP5E related disorders. European Journal of Paediatric Neurology, 2016, 20, 286-295.	1.6	36
45	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. American Journal of Human Genetics, 2016, 98, 363-372.	6.2	36
46	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	2.8	34
47	Pernicious anemia â€“ Genetic insights. Autoimmunity Reviews, 2011, 10, 455-459.	5.8	33
48	The clinical presentation caused by truncating CHD8 variants. Clinical Genetics, 2019, 96, 72-84.	2.0	32
49	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
50	The clinical spectrum of the congenital myasthenic syndrome resulting from COL13A1 mutations. Brain, 2019, 142, 1547-1560.	7.6	30
51	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. American Journal of Human Genetics, 2021, 108, 346-356.	6.2	30
52	Clinical delineation, sex differences, and genotypeâ€“phenotype correlation in pathogenic KDM6A variants causing X-linked Kabuki syndrome type 2. Genetics in Medicine, 2021, 23, 1202-1210.	2.4	30
53	A comparative analysis of KMT2D missense variants in Kabuki syndrome, cancers and the general population. Journal of Human Genetics, 2019, 64, 161-170.	2.3	26
54	Genotypeâ€“phenotype specificity in Menkeâ€“Hennekam syndrome caused by missense variants in exon 30 or 31 of CREBBP. American Journal of Medical Genetics, Part A, 2019, 179, 1058-1062.	1.2	23

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55	Telemedicine strategy of the European Reference Network ITHACA for the diagnosis and management of patients with rare developmental disorders. Orphanet Journal of Rare Diseases, 2020, 15, 103.	2.7	23
56	Leriâ€™s pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassingGDF6andSDC2and provides insight into systemic sclerosis pathogenesis. Annals of the Rheumatic Diseases, 2015, 74, 1249-1256.	0.9	22
57	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Genetics in Medicine, 2019, 21, 1058-1064.	2.4	22
58	Kabuki syndrome. Clinical Dysmorphology, 2015, 24, 135-139.	0.3	21
59	Post-translational formation of hypusine in eIF5A: implications in human neurodevelopment. Amino Acids, 2022, 54, 485-499.	2.7	19
60	Variability of bone marrow morphology in G6PC3 mutations: Is there a genotypeâ€“phenotype correlation or ageâ€dependent relationship?. American Journal of Hematology, 2011, 86, 235-237.	4.1	18
61	Expanding the genetic and phenotypic spectrum of branchedâ€chain amino acid transferase 2 deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 809-817.	3.6	18
62	Early Diagnosis and Treatment of Cobalamin Deficiency of Infancy Owing to Occult Maternal Pernicious Anemia. Journal of Pediatric Hematology/Oncology, 2010, 32, 319-322.	0.6	17
63	G6PC3 mutations cause non-syndromic severe congenital neutropenia. Molecular Genetics and Metabolism, 2013, 108, 138-141.	1.1	16
64	HIST1H1E heterozygous proteinâ€truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the HIST1H1E syndrome phenotype in 30 individuals. American Journal of Medical Genetics, Part A, 2019, 179, 2049-2055.	1.2	16
65	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	8.2	16
66	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	2.4	16
67	Trisomy 18 mosaicism: report of two cases. World Journal of Pediatrics, 2013, 9, 179-181.	1.8	15
68	Neutrophil dysfunction triggers inflammatory bowel disease in G6PC3 deficiency. Journal of Leukocyte Biology, 2021, 109, 1147-1154.	3.3	14
69	Personalised virtual gene panels reduce interpretation workload and maintain diagnostic rates of proband-only clinical exome sequencing for rare disorders. Journal of Medical Genetics, 2022, 59, 393-398.	3.2	14
70	Growth hormone deficiency as a cause for short stature in Wiedemannâ€“Steiner Syndrome. Endocrinology, Diabetes and Metabolism Case Reports, 2018, 2018, .	0.5	14
71	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.7	14
72	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. Frontiers in Molecular Neuroscience, 2020, 13, 12.	2.9	12

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73	Exome Sequencing Identifies a Dominant <i>TNNT3</i> Mutation in a Large Family with Distal Arthrogryposis. Molecular Syndromology, 2014, 5, 218-228.	0.8	11
74	Profound vitamin D deficiency in four siblings with Imerslundâ€Grasbeck syndrome with homozygous CUBN mutation. JIMD Reports, 2019, 49, 43-47.	1.5	11
75	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. Genetics in Medicine, 2021, 23, 2138-2149.	2.4	11
76	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. Npj Genomic Medicine, 2021, 6, 92.	3.8	11
77	Biallelic <scp>TMEM260</scp> variants cause truncus arteriosus, with or without renal defects. Clinical Genetics, 2022, 101, 127-133.	2.0	10
78	Comparison of methylation episignatures in KMT2B- and KMT2D-related human disorders. Epigenomics, 2022, 14, 537-547.	2.1	10
79	Haploinsufficiency of <scp>ATP6V0C</scp> possibly underlies 16p13.3 deletions that cause microcephaly, seizures, and neurodevelopmental disorder. American Journal of Medical Genetics, Part A, 2021, 185, 196-202.	1.2	9
80	ERBB4 exonic deletions on chromosome 2q34 in patients with intellectual disability or epilepsy. European Journal of Human Genetics, 2021, 29, 1377-1383.	2.8	9
81	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. Genetics in Medicine, 2021, 23, 1901-1911.	2.4	9
82	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2021, 108, 1069-1082.	6.2	8
83	Array comparative genomic hybridisation-based identification of two imbalances of chromosome 1p in a 9-year-old girl with a monosomy 1p36 related phenotype and a family history of learning difficulties: a case report. Journal of Medical Case Reports, 2008, 2, 355.	0.8	7
84	Recurrent <scp>KCNT2</scp> missense variants affecting p.Arg190 result in a recognizable phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3083-3091.	1.2	7
85	A homozygous <scp>GRIN1</scp> null variant causes a more severe phenotype of early infantile epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2022, 188, 595-599.	1.2	7
86	First report of occurrence of choroid plexus papilloma and medulloblastoma in the same patient. Child's Nervous System, 2007, 23, 587-589.	1.1	6
87	Activating RAC1 variants in the switch II region cause a developmental syndrome and alter neuronal morphology. Brain, 2022, 145, 4232-4245.	7.6	6
88	Liver Cyst Caused by the Peritoneal Catheter of a CSF Shunt. Pediatric Neurosurgery, 2007, 43, 444-445.	0.7	5
89	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. American Journal of Human Genetics, 2011, 89, 346.	6.2	5
90	Solving unsolved rare neurological diseasesâ€”a Solve-RD viewpoint. European Journal of Human Genetics, 2021, 29, 1332-1336.	2.8	4

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91	A novel 800â€‰kb microduplication of chromosome 16q22.1 resulting in learning disability and epilepsy may explain phenotypic variability in a family with 15q13 microdeletion. American Journal of Medical Genetics, Part A, 2011, 155, 1453-1457.	1.2	3
92	Presence of pathogenic copy number variants (CNVs) is correlated with socioeconomic status. Journal of Medical Genetics, 2020, 57, 70-72.	3.2	3
93	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A singleâ€institution experience. Clinical Otolaryngology, 2021, 46, 1257-1262.	1.2	3
94	Liver Cyst Caused by the Peritoneal Catheter of a Cerebrospinal Fluid Shunt. Pediatric Neurosurgery, 2007, 43, 343-344.	0.7	2
95	A maternally inherited frameshift CDKL5 variant in a male with global developmental delay and lateâ€onset generalized epilepsy. American Journal of Medical Genetics, Part A, 2019, 179, 507-511.	1.2	2
96	Response to: â€˜Mutation in MMP2 gene may result in scleroderma-like skin thickeningâ€™ by Bader-Meunieret al. Annals of the Rheumatic Diseases, 2016, 75, e2-e2.	0.9	1
97	Reply: Expanding the clinical and genetic spectrum of PCYT2-related disorders. Brain, 2020, 143, e77-e77.	7.6	1
98	De-novo duplication of 5(q13.3q21.1) in a child with vitreo-retinal dysplasia and learning disability. Clinical Dysmorphology, 2010, 19, 73-75.	0.3	0
99	The relevance of deep genomic analyses in families with variably expressive CNVs in the era of personalized medicine. Molecular Genetics and Metabolism, 2021, 132, S69.	1.1	0
100	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0
101	Refining the clinical phenotype associated with missense variants in exons 38 and 39 of KMT2D. American Journal of Medical Genetics, Part A, 2022, , .	1.2	0