

Siddharth Banka

List of Publications by Year in descending order

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101
papers

5,505
citations

101384

36
h-index

102304

66
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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 – Preliminary Report. <i>New England Journal of Medicine</i> , 2021, 385, 1868-1880.	13.9	352
2	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. <i>Nature Genetics</i> , 2016, 48, 1060-1065.	9.4	351
3	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020, 586, 757-762.	13.7	343
4	Whole-genome sequencing of patients with rare diseases in a national health system. <i>Nature</i> , 2020, 583, 96-102.	13.7	338
5	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. <i>Immunity</i> , 2015, 42, 1185-1196.	6.6	246
6	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. <i>American Journal of Human Genetics</i> , 2018, 102, 175-187.	2.6	204
7	Spectrum of <i>MLL2</i> (<i>ALR</i>) mutations in 110 cases of Kabuki syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1511-1516.	0.7	160
8	Kabuki syndrome: international consensus diagnostic criteria. <i>Journal of Medical Genetics</i> , 2019, 56, 89-95.	1.5	146
9	Germline mutations in the oncogene <i>EZH2</i> cause Weaver syndrome and increased human height. <i>Oncotarget</i> , 2011, 2, 1127-1133.	0.8	145
10	How genetically heterogeneous is Kabuki syndrome?: <i>MLL2</i> testing in 116 patients, review and analyses of mutation and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2012, 20, 381-388.	1.4	142
11	Weaver syndrome and <i>EZH2</i> mutations: Clarifying the clinical phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2013, 161, 2972-2980.	0.7	119
12	<i>RAC1</i> Missense Mutations in Developmental Disorders with Diverse Phenotypes. <i>American Journal of Human Genetics</i> , 2017, 101, 466-477.	2.6	119
13	The genetic basis of DOORS syndrome: an exome-sequencing study. <i>Lancet Neurology</i> , The, 2014, 13, 44-58.	4.9	108
14	Mutations in <i>PRDM5</i> in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. <i>American Journal of Human Genetics</i> , 2011, 88, 767-777.	2.6	106
15	Novel <i>KDM6A</i> (<i>UTX</i>) mutations and a clinical and molecular review of the X-linked Kabuki syndrome (<i>KS2</i>). <i>Clinical Genetics</i> , 2015, 87, 252-258.	1.0	102
16	<i>TBC1D24</i> genotype-phenotype correlation. <i>Neurology</i> , 2016, 87, 77-85.	1.5	97
17	Identification and Characterization of an Inborn Error of Metabolism Caused by Dihydrofolate Reductase Deficiency. <i>American Journal of Human Genetics</i> , 2011, 88, 216-225.	2.6	90
18	Corneal confocal microscopy detects small fiber neuropathy in Charcot-Marie-Tooth disease type 1A patients. <i>Muscle and Nerve</i> , 2012, 46, 698-704.	1.0	89

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19	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	2.6	87
20	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 101, 1021-1033.	2.6	83
21	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. <i>American Journal of Human Genetics</i> , 2016, 98, 981-992.	2.6	81
22	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. <i>Brain</i> , 2019, 142, 3382-3397.	3.7	76
23	A basement membrane discovery pipeline uncovers network complexity, regulators, and human disease associations. <i>Science Advances</i> , 2022, 8, eabn2265.	4.7	76
24	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. <i>Journal of Clinical Investigation</i> , 2015, 125, 3585-3599.	3.9	69
25	Extended Spectrum of Human Glucose-6-Phosphatase Catalytic Subunit 3 Deficiency: Novel Genotypes and Phenotypic Variability in Severe Congenital Neutropenia. <i>Journal of Pediatrics</i> , 2012, 160, 679-683.e2.	0.9	67
26	A clinical and molecular review of ubiquitous glucose-6-phosphatase deficiency caused by G6PC3 mutations. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 84.	1.2	67
27	Exome sequencing identifies ATP4A gene as responsible of an atypical familial type I gastric neuroendocrine tumour. <i>Human Molecular Genetics</i> , 2015, 24, 2914-2922.	1.4	60
28	Opposite Modulation of RAC1 by Mutations in TRIO Is Associated with Distinct, Domain-Specific Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2020, 106, 338-355.	2.6	58
29	Further delineation of the KAT6B molecular and phenotypic spectrum. <i>European Journal of Human Genetics</i> , 2015, 23, 1165-1170.	1.4	56
30	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. <i>American Journal of Human Genetics</i> , 2019, 104, 1210-1222.	2.6	56
31	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. <i>American Journal of Human Genetics</i> , 2020, 106, 234-245.	2.6	56
32	Further delineation of the phenotype of severe congenital neutropenia type 4 due to mutations in G6PC3. <i>European Journal of Human Genetics</i> , 2011, 19, 18-22.	1.4	50
33	Expanding the clinical and molecular spectrum of thiamine pyrophosphokinase deficiency: A treatable neurological disorder caused by TPK1 mutations. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 301-306.	0.5	50
34	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismature of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021, 108, 502-516.	2.6	48
35	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. <i>European Journal of Human Genetics</i> , 2019, 27, 747-759.	1.4	47
36	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. <i>American Journal of Human Genetics</i> , 2019, 104, 948-956.	2.6	45

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37	<i>MLL2</i> mosaic mutations and intragenic deletion/duplications in patients with Kabuki syndrome. <i>Clinical Genetics</i> , 2013, 83, 467-471.	1.0	44
38	Diagnosing childhood-onset inborn errors of metabolism by next-generation sequencing. <i>Archives of Disease in Childhood</i> , 2017, 102, 1019-1029.	1.0	43
39	Non-coding region variants upstream of <i>MEF2C</i> cause severe developmental disorder through three distinct loss-of-function mechanisms. <i>American Journal of Human Genetics</i> , 2021, 108, 1083-1094.	2.6	42
40	Novel diagnostic DNA methylation epigenatures expand and refine the epigenetic landscapes of Mendelian disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100075.	1.0	42
41	A restricted spectrum of missense <i>KMT2D</i> variants cause a multiple malformations disorder distinct from Kabuki syndrome. <i>Genetics in Medicine</i> , 2020, 22, 867-877.	1.1	41
42	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. <i>Nature Communications</i> , 2021, 12, 833.	5.8	41
43	Mutations in the <i>G6PC3</i> gene cause Dursun syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2609-2611.	0.7	37
44	Role of reverse phenotyping in interpretation of next generation sequencing data and a review of INPP5E related disorders. <i>European Journal of Paediatric Neurology</i> , 2016, 20, 286-295.	0.7	36
45	Small 6q16.1 Deletions Encompassing <i>POU3F2</i> Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. <i>American Journal of Human Genetics</i> , 2016, 98, 363-372.	2.6	36
46	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. <i>European Journal of Human Genetics</i> , 2021, 29, 1337-1347.	1.4	34
47	Pernicious anemia – Genetic insights. <i>Autoimmunity Reviews</i> , 2011, 10, 455-459.	2.5	33
48	The clinical presentation caused by truncating <i>CHD8</i> variants. <i>Clinical Genetics</i> , 2019, 96, 72-84.	1.0	32
49	New insights into the clinical and molecular spectrum of the novel <i>CYFIP2</i> -related neurodevelopmental disorder and impairment of the WRC-mediated actin dynamics. <i>Genetics in Medicine</i> , 2021, 23, 543-554.	1.1	32
50	The clinical spectrum of the congenital myasthenic syndrome resulting from <i>COL13A1</i> mutations. <i>Brain</i> , 2019, 142, 1547-1560.	3.7	30
51	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with <i>SATB1</i> dysfunction. <i>American Journal of Human Genetics</i> , 2021, 108, 346-356.	2.6	30
52	Clinical delineation, sex differences, and genotype-phenotype correlation in pathogenic <i>KDM6A</i> variants causing X-linked Kabuki syndrome type 2. <i>Genetics in Medicine</i> , 2021, 23, 1202-1210.	1.1	30
53	A comparative analysis of <i>KMT2D</i> missense variants in Kabuki syndrome, cancers and the general population. <i>Journal of Human Genetics</i> , 2019, 64, 161-170.	1.1	26
54	Genotype-phenotype specificity in Menke-Hennekam syndrome caused by missense variants in exon 30 or 31 of <i>CREBBP</i> . <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1058-1062.	0.7	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. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 103.	1.2	23
56	Leri's pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassing <i>GDF6</i> and <i>SDC2</i> and provides insight into systemic sclerosis pathogenesis. <i>Annals of the Rheumatic Diseases</i> , 2015, 74, 1249-1256.	0.5	22
57	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. <i>Genetics in Medicine</i> , 2019, 21, 1058-1064.	1.1	22
58	Kabuki syndrome. <i>Clinical Dysmorphology</i> , 2015, 24, 135-139.	0.1	21
59	Post-translational formation of hypusine in eIF5A: implications in human neurodevelopment. <i>Amino Acids</i> , 2022, 54, 485-499.	1.2	19
60	Variability of bone marrow morphology in <i>G6PC3</i> mutations: Is there a genotype-phenotype correlation or age-dependent relationship?. <i>American Journal of Hematology</i> , 2011, 86, 235-237.	2.0	18
61	Expanding the genetic and phenotypic spectrum of branched-chain amino acid transferase 2 deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 809-817.	1.7	18
62	Early Diagnosis and Treatment of Cobalamin Deficiency of Infancy Owing to Occult Maternal Pernicious Anemia. <i>Journal of Pediatric Hematology/Oncology</i> , 2010, 32, 319-322.	0.3	17
63	<i>G6PC3</i> mutations cause non-syndromic severe congenital neutropenia. <i>Molecular Genetics and Metabolism</i> , 2013, 108, 138-141.	0.5	16
64	<i>HIST1H1E</i> heterozygous protein-truncating variants cause a recognizable syndrome with intellectual disability and distinctive facial gestalt: A study to clarify the <i>HIST1H1E</i> syndrome phenotype in 30 individuals. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2049-2055.	0.7	16
65	Genotype-phenotype correlations and novel molecular insights into the <i>DHX30</i> -associated neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 90.	3.6	16
66	Delineating the molecular and phenotypic spectrum of the <i>SETD1B</i> -related syndrome. <i>Genetics in Medicine</i> , 2021, 23, 2122-2137.	1.1	16
67	Trisomy 18 mosaicism: report of two cases. <i>World Journal of Pediatrics</i> , 2013, 9, 179-181.	0.8	15
68	Neutrophil dysfunction triggers inflammatory bowel disease in <i>G6PC3</i> deficiency. <i>Journal of Leukocyte Biology</i> , 2021, 109, 1147-1154.	1.5	14
69	Personalised virtual gene panels reduce interpretation workload and maintain diagnostic rates of proband-only clinical exome sequencing for rare disorders. <i>Journal of Medical Genetics</i> , 2022, 59, 393-398.	1.5	14
70	Growth hormone deficiency as a cause for short stature in Wiedemann-Steiner Syndrome. <i>Endocrinology, Diabetes and Metabolism Case Reports</i> , 2018, 2018, .	0.2	14
71	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100074.	1.0	14
72	Expanding Clinical Presentations Due to Variations in <i>THOC2</i> mRNA Nuclear Export Factor. <i>Frontiers in Molecular Neuroscience</i> , 2020, 13, 12.	1.4	12

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73	Exome Sequencing Identifies a Dominant <i>TNNT3</i> Mutation in a Large Family with Distal Arthrogyriposis. <i>Molecular Syndromology</i> , 2014, 5, 218-228.	0.3	11
74	Profound vitamin D deficiency in four siblings with Imlerslund-Grasbeck syndrome with homozygous CUBN mutation. <i>JIMD Reports</i> , 2019, 49, 43-47.	0.7	11
75	Biallelic variants in <i>PCDHGC4</i> cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. <i>Genetics in Medicine</i> , 2021, 23, 2138-2149.	1.1	11
76	Deficiency of TET3 leads to a genome-wide DNA hypermethylation epigenature in human whole blood. <i>Npj Genomic Medicine</i> , 2021, 6, 92.	1.7	11
77	Biallelic <i>TMEM260</i> variants cause truncus arteriosus, with or without renal defects. <i>Clinical Genetics</i> , 2022, 101, 127-133.	1.0	10
78	Comparison of methylation epigenatures in <i>KMT2B</i> - and <i>KMT2D</i> -related human disorders. <i>Epigenomics</i> , 2022, 14, 537-547.	1.0	10
79	Haploinsufficiency of <i>ATP6VOC</i> possibly underlies 16p13.3 deletions that cause microcephaly, seizures, and neurodevelopmental disorder. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 196-202.	0.7	9
80	<i>ERBB4</i> exonic deletions on chromosome 2q34 in patients with intellectual disability or epilepsy. <i>European Journal of Human Genetics</i> , 2021, 29, 1377-1383.	1.4	9
81	Haploinsufficiency of <i>ARFGEF1</i> is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. <i>Genetics in Medicine</i> , 2021, 23, 1901-1911.	1.1	9
82	Bi-allelic loss-of-function variants in <i>BCAS3</i> cause a syndromic neurodevelopmental disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 1069-1082.	2.6	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. <i>Journal of Medical Case Reports</i> , 2008, 2, 355.	0.4	7
84	Recurrent <i>KCNT2</i> missense variants affecting p.Arg190 result in a recognizable phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 3083-3091.	0.7	7
85	A homozygous <i>GRIN1</i> null variant causes a more severe phenotype of early infantile epileptic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 595-599.	0.7	7
86	First report of occurrence of choroid plexus papilloma and medulloblastoma in the same patient. <i>Child's Nervous System</i> , 2007, 23, 587-589.	0.6	6
87	Activating <i>RAC1</i> variants in the switch II region cause a developmental syndrome and alter neuronal morphology. <i>Brain</i> , 2022, 145, 4232-4245.	3.7	6
88	Liver Cyst Caused by the Peritoneal Catheter of a CSF Shunt. <i>Pediatric Neurosurgery</i> , 2007, 43, 444-445.	0.4	5
89	Mutations in <i>PRDM5</i> in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. <i>American Journal of Human Genetics</i> , 2011, 89, 346.	2.6	5
90	Solving unsolved rare neurological diseases—a Solve-RD viewpoint. <i>European Journal of Human Genetics</i> , 2021, 29, 1332-1336.	1.4	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.	0.7	3
92	Presence of pathogenic copy number variants (CNVs) is correlated with socioeconomic status. Journal of Medical Genetics, 2020, 57, 70-72.	1.5	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.	0.6	3
94	Liver Cyst Caused by the Peritoneal Catheter of a Cerebrospinal Fluid Shunt. Pediatric Neurosurgery, 2007, 43, 343-344.	0.4	2
95	A maternally inherited frameshift <i>CDKL5</i> variant in a male with global developmental delay and lateâ€onset generalized epilepsy. American Journal of Medical Genetics, Part A, 2019, 179, 507-511.	0.7	2
96	Response to: â€Mutation in MMP2 gene may result in scleroderma-like skin thickeningâ€™ by Bader-Meunier<i>et al</i>. Annals of the Rheumatic Diseases, 2016, 75, e2-e2.	0.5	1
97	Reply: Expanding the clinical and genetic spectrum of PCYT2-related disorders. Brain, 2020, 143, e77-e77.	3.7	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.	0.1	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.	0.5	0
100	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	1.1	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, , .	0.7	0