Siddharth Banka

List of Publications by Citations

99 papers	2,972 citations	31 h-index	53 g-index
114 ext. papers	4,394 ext. citations	7.9 avg, IF	4.29 L-index

#	Paper	IF	Citations
99	Distinct genetic architectures for syndromic and nonsyndromic congenital heart defects identified by exome sequencing. *Nature Genetics*, **2016**, 48, 1060-5	36.3	200
98	Dominant Mutations in the Autoimmune Regulator AIRE Are Associated with Common Organ-Specific Autoimmune Diseases. *Immunity*, **2015**, 42, 1185-96	32.3	156
97	Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*, **2020**, 583, 96-102	50.4	139
96	Spectrum of MLL2 (ALR) mutations in 110 cases of Kabuki syndrome. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1511-6	2.5	132
95	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, 381-8	5.3	113
94	Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. *Oncotarget*, **2011**, 2, 1127-33	3.3	110
93	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
92	Evidence for 28 genetic disorders discovered by combining healthcare and research data. *Nature*, **2020**, 586, 757-762	50.4	103
91	The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurology, The*, **2014**, 13, 44-58	24.1	96
90	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	11	84
89	Novel KDM6A (UTX) mutations and a clinical and molecular review of the X-linked Kabuki syndrome (KS2). *Clinical Genetics*, **2015**, 87, 252-8	4	80
88	Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 2972-80	2.5	80
87	Kabuki syndrome: international consensus diagnostic criteria. *Journal of Medical Genetics*, **2019**, 56, 89-95	5.8	80
86	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. *Neurology*, **2016**, 87, 77-85	6.5	75
85	RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes. *American Journal of Human Genetics*, **2017**, 101, 466-477	11	73
84	Identification and characterization of an inborn error of metabolism caused by dihydrofolate reductase deficiency. *American Journal of Human Genetics*, **2011**, 88, 216-25	11	68
83	Corneal confocal microscopy detects small-fiber neuropathy in Charcot-Marie-Tooth disease type 1A patients. *Muscle and Nerve*, **2012**, 46, 698-704	3.4	67

82	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	3.6	60
81	A clinical and molecular review of ubiquitous glucose-6-phosphatase deficiency caused by G6PC3 mutations. *Orphanet Journal of Rare Diseases*, **2013**, 8, 84	4.2	53
80	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	11	53
79	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. *American Journal of Human Genetics*, **2017**, 101, 1021-1033	11	50
78	Exome sequencing identifies ATP4A gene as responsible of an atypical familial type I gastric neuroendocrine tumour. *Human Molecular Genetics*, **2015**, 24, 2914-22	5.6	49
77	RAP1-mediated MEK/ERK pathway defects in Kabuki syndrome. *Journal of Clinical Investigation*, **2015**, 125, 3585-99	15.9	48
76	Further delineation of the KAT6B molecular and phenotypic spectrum. *European Journal of Human Genetics*, **2015**, 23, 1165-70	5.3	45
75	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	11	44
74	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. *Brain*, **2019**, 142, 3382-3397	11.2	40
73	Expanding the clinical and molecular spectrum of thiamine pyrophosphokinase deficiency: a treatable neurological disorder caused by TPK1 mutations. *Molecular Genetics and Metabolism*, **2014**, 113, 301-6	3.7	40
72	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	5.3	40
71	MLL2 mosaic mutations and intragenic deletion-duplications in patients with Kabuki syndrome. *Clinical Genetics*, **2013**, 83, 467-71	4	34
70	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *New England Journal of Medicine*, **2021**, 385, 1868-1880	59.2	34
69	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
68	Mutations in the G6PC3 gene cause Dursun syndrome. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 2609-11	2.5	31
67	Pernicious anemia - genetic insights. *Autoimmunity Reviews*, **2011**, 10, 455-9	13.6	29
66	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. *American Journal of Human Genetics*, **2016**, 98, 363-72	11	26
65	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	5.3	25

64	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	3.8	25
63	Diagnosing childhood-onset inborn errors of metabolism by next-generation sequencing. *Archives of Disease in Childhood*, **2017**, 102, 1019-1029	2.2	24
62	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. *American Journal of Human Genetics*, **2020**, 106, 234-245	11	22
61	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	11	21
60	The clinical spectrum of the congenital myasthenic syndrome resulting from COL13A1 mutations. *Brain*, **2019**, 142, 1547-1560	11.2	20
59	The clinical presentation caused by truncating CHD8 variants. *Clinical Genetics*, **2019**, 96, 72-84	4	20
58	Leri's pleonosteosis, a congenital rheumatic disease, results from microduplication at 8q22.1 encompassing GDF6 and SDC2 and provides insight into systemic sclerosis pathogenesis. *Annals of the Rheumatic Diseases*, **2015**, 74, 1249-56	2.4	19
57	Kabuki syndrome: expanding the phenotype to include microphthalmia and anophthalmia. *Clinical Dysmorphology*, **2015**, 24, 135-9	0.9	18
56	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. *American Journal of Human Genetics*, **2019**, 104, 948-956	11	17
55	A restricted spectrum of missense KMT2D variants cause a multiple malformations disorder distinct from Kabuki syndrome. *Genetics in Medicine*, **2020**, 22, 867-877	8.1	17
54	G6PC3 mutations cause non-syndromic severe congenital neutropenia. *Molecular Genetics and Metabolism*, **2013**, 108, 138-41	3.7	16
53	Variability of bone marrow morphology in G6PC3 mutations: is there a genotype-phenotype correlation or age-dependent relationship?. *American Journal of Hematology*, **2011**, 86, 235-7	7.1	16
52	A comparative analysis of KMT2D missense variants in Kabuki syndrome, cancers and the general population. *Journal of Human Genetics*, **2019**, 64, 161-170	4.3	16
51	Genotype-phenotype specificity in Menke-Hennekam syndrome caused by missense variants in exon 30 or 31 of CREBBP. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1058-1062	2.5	14
50	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	4.2	14
49	Early diagnosis and treatment of cobalamin deficiency of infancy owing to occult maternal pernicious anemia. *Journal of Pediatric Hematology/Oncology*, **2010**, 32, 319-22	1.2	13
48	Impaired eIF5A function causes a Mendelian disorder that is partially rescued in model systems by spermidine. *Nature Communications*, **2021**, 12, 833	17.4	13
47	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	11	12

46	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. *Genetics in Medicine*, **2019**, 21, 1058-1064	8.1	12
45	HIST1H1E heterozygous protein-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	2.5	10
44	Trisomy 18 mosaicism: report of two cases. *World Journal of Pediatrics*, **2013**, 9, 179-81	4.6	10
43	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	8.1	9
42	Expanding the genetic and phenotypic spectrum of branched-chain amino acid transferase 2 deficiency. *Journal of Inherited Metabolic Disease*, **2019**, 42, 809-817	5.4	8
41	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	11	8
40	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	1.2	7
39	Growth hormone deficiency as a cause for short stature in Wiedemann-Steiner Syndrome. *Endocrinology, Diabetes and Metabolism Case Reports*, **2018**, 2018,	1.4	7
38	Mutation-specific pathophysiological mechanisms define different neurodevelopmental disorders associated with SATB1 dysfunction. *American Journal of Human Genetics*, **2021**, 108, 346-356	11	7
37	Exome Sequencing Identifies a Dominant TNNT3 Mutation in a Large Family with Distal Arthrogryposis. *Molecular Syndromology*, **2014**, 5, 218-28	1.5	6
36	First report of occurrence of choroid plexus papilloma and medulloblastoma in the same patient. *Childs Nervous System*, **2007**, 23, 587-9	1.7	6
35	Profound vitamin D deficiency in four siblings with Imerslund-Grasbeck syndrome with homozygous CUBN mutation. *JIMD Reports*, **2019**, 49, 43-47	1.9	5
34	Expanding Clinical Presentations Due to Variations in THOC2 mRNA Nuclear Export Factor. *Frontiers in Molecular Neuroscience*, **2020**, 13, 12	6.1	4
33	Bi-allelic loss-of-function variants in BCAS3 cause a syndromic neurodevelopmental disorder. *American Journal of Human Genetics*, **2021**, 108, 1069-1082	11	4
32	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. *European Journal of Human Genetics*, **2021**, 29, 1337-1347	5.3	4
31	Post-translational formation of hypusine in eIF5A: implications in human neurodevelopment. *Amino Acids*, **2021**, 1	3.5	4
30	A basement membrane discovery pipeline uncovers network complexity, regulators, and human disease associations.. *Science Advances*, **2022**, 8, eabn2265	14.3	4
29	A novel 800 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**, 155A, 1453-7	2.5	3

28	Liver cyst caused by the peritoneal catheter of a CSF shunt. *Pediatric Neurosurgery*, **2007**, 43, 444-5	0.9	3
27	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability.. *Human Genetics and Genomics Advances*, **2022**, 3, 100074	0.8	3
26	Clinical delineation, sex differences, and genotype-phenotype correlation in pathogenic KDM6A variants causing X-linked Kabuki syndrome type 2. *Genetics in Medicine*, **2021**, 23, 1202-1210	8.1	3
25	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. *Genetics in Medicine*, **2021**, 23, 2122-2137	8.1	3
24	Presence of pathogenic copy number variants (CNVs) is correlated with socioeconomic status. *Journal of Medical Genetics*, **2020**, 57, 70-72	5.8	2
23	Mutations in PRDM5 in Brittle Cornea Syndrome Identify a Pathway Regulating Extracellular Matrix Development and Maintenance. *American Journal of Human Genetics*, **2011**, 89, 346	11	2
22	Liver cyst caused by the peritoneal catheter of a cerebrospinal fluid shunt. *Pediatric Neurosurgery*, **2007**, 43, 343-4	0.9	2
21	Personalised virtual gene panels reduce interpretation workload and maintain diagnostic rates of proband-only clinical exome sequencing for rare disorders. *Journal of Medical Genetics*, **2021**,	5.8	2
20	Genotype-phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 90	14.4	2
19	Recurrent KCNT2 missense variants affecting p.Arg190 result in a recognizable phenotype. *American Journal of Medical Genetics, Part A*, **2021**, 185, 3083-3091	2.5	2
18	Neutrophil dysfunction triggers inflammatory bowel disease in G6PC3 deficiency. *Journal of Leukocyte Biology*, **2021**, 109, 1147-1154	6.5	2
17	Haploinsufficiency of ATP6V0C possibly underlies 16p13.3 deletions that cause microcephaly, seizures, and neurodevelopmental disorder. *American Journal of Medical Genetics, Part A*, **2020**,	2.5	2
16	ERBB4 exonic deletions on chromosome 2q34 in patients with intellectual disability or epilepsy. *European Journal of Human Genetics*, **2021**, 29, 1377-1383	5.3	2
15	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders.. *Human Genetics and Genomics Advances*, **2022**, 3, 100075	0.8	1
14	Biallelic TMEM260 variants cause truncus arteriosus, with or without renal defects. *Clinical Genetics*, **2022**, 101, 127-133	4	1
13	A basement membrane discovery pipeline uncovers network complexity, new regulators, and human disease associations		1
12	A homozygous GRIN1 null variant causes a more severe phenotype of early infantile epileptic encephalopathy. *American Journal of Medical Genetics, Part A*, **2021**,	2.5	1
11	Delineation of the First Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency		1

10	Haploinsufficiency of ARFGEF1 is associated with developmental delay, intellectual disability, and epilepsy with variable expressivity. *Genetics in Medicine*, **2021**, 23, 1901-1911	8.1	1
9	Biallelic variants in PCDHGC4 cause a novel neurodevelopmental syndrome with progressive microcephaly, seizures, and joint anomalies. *Genetics in Medicine*, **2021**, 23, 2138-2149	8.1	1
8	Response to: Mutation in MMP2 gene may result in scleroderma-like skin thickening by Bader-Meunier et al. *Annals of the Rheumatic Diseases*, **2016**, 75, e2	2.4	1
7	A maternally inherited frameshift CDKL5 variant in a male with global developmental delay and late-onset generalized epilepsy. *American Journal of Medical Genetics, Part A*, **2019**, 179, 507-511	2.5	1
6	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. *Npj Genomic Medicine*, **2021**, 6, 92	6.2	0
5	Reply: Expanding the clinical and genetic spectrum of PCYT2-related disorders. *Brain*, **2020**, 143, e77	11.2	0
4	Solving unsolved rare neurological diseases-a Solve-RD viewpoint. *European Journal of Human Genetics*, **2021**, 29, 1332-1336	5.3	0
3	The diagnostic utility of clinical exome sequencing in 60 patients with hearing loss disorders: A single-institution experience. *Clinical Otolaryngology*, **2021**, 46, 1257-1262	1.8	0
2	De-novo duplication of 5(q13.3q21.1) in a child with vitreo-retinal dysplasia and learning disability. *Clinical Dysmorphology*, **2010**, 19, 73-75	0.9	
1	Refining the clinical phenotype associated with missense variants in exons 38 and 39 of KMT2D.. *American Journal of Medical Genetics, Part A*, **2022**,	2.5	