Anju Shukla

List of Publications by Citations

101 papers	1,011 citations	18 h-index	28 g-index
112 ext. papers	1,466 ext. citations	4.2 avg, IF	4.2 L-index

#	Paper	IF	Citations
101	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. *Nature*, **2018**, 557, 564-569	50.4	97
100	Biallelic mutations in the death domain of PIDD1 impair caspase-2 activation and are associated with intellectual disability. *Translational Psychiatry*, **2021**, 11, 1	8.6	66
99	The Genomics of Arthrogryposis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. *American Journal of Human Genetics*, **2019**, 105, 132-150	11	50
98	Homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. *Journal of Human Genetics*, **2017**, 62, 723-727	4.3	45
97	Noonan syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 2323-2334	2.5	43
96	Biallelic Loss of Proprioception-Related PIEZO2 Causes Muscular Atrophy with Perinatal Respiratory Distress, Arthrogryposis, and Scoliosis. *American Journal of Human Genetics*, **2016**, 99, 1206-1216	11	41
95	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. *American Journal of Human Genetics*, **2017**, 100, 117-127	11	39
94	The homozygous variant c.797G>A/p.(Cys266Tyr) in PISD is associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. *Human Mutation*, **2019**, 40, 299-309	4.7	33
93	Williams-Beuren syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1128-1136	2.5	31
92	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. *Brain*, **2019**, 142, 50-58	11.2	31
91	Autosomal recessive spinocerebellar ataxia 20: Report of a new patient and review of literature. *European Journal of Medical Genetics*, **2017**, 60, 118-123	2.6	26
90	Clinical utility of fetal autopsy and its impact on genetic counseling. *Prenatal Diagnosis*, **2015**, 35, 685-91	3.2	25
89	Cornelia de Lange syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 150-158	2.5	25
88	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. *Bone*, **2018**, 110, 368-377	4.7	21
87	Identification of a novel LRRK1 mutation in a family with osteosclerotic metaphyseal dysplasia. *Journal of Human Genetics*, **2017**, 62, 437-441	4.3	21
86	Homozygosity for a nonsense variant in AIMP2 is associated with a progressive neurodevelopmental disorder with microcephaly, seizures, and spastic quadriparesis. *Journal of Human Genetics*, **2018**, 63, 19-25	4.3	21
85	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. *Journal of Human Genetics* , **2019**, 64, 1173-1186	4.3	20

84	A novel sequence variant in SFRP4 causing Pyle disease. *Journal of Human Genetics*, **2017**, 62, 575-576	4.3	19
83	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. *American Journal of Medical Genetics, Part A*, **2016**, 170A, 410-417	2.5	18
82	Gaucher disease: single gene molecular characterization of one-hundred Indian patients reveals novel variants and the most prevalent mutation. *BMC Medical Genetics*, **2019**, 20, 31	2.1	17
81	Recurrent and novel GLB1 mutations in India. *Gene*, **2015**, 567, 173-81	3.8	17
80	Novel and recurrent mutations in WISP3 and an atypical phenotype. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 2481-4	2.5	17
79	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. *European Journal of Human Genetics*, **2018**, 26, 695-708	5.3	14
78	Locus and allelic heterogeneity and phenotypic variability in Waardenburg syndrome. *Clinical Genetics*, **2019**, 95, 398-402	4	13
77	Additional three patients with Smith-McCort dysplasia due to novel RAB33B mutations. *American Journal of Medical Genetics, Part A*, **2017**, 173, 588-595	2.5	12
76	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. *Brain*, **2020**, 143, 2929-2944	11.2	12
75	Report of four novel variants in ASNS causing asparagine synthetase deficiency and review of literature. *Congenital Anomalies (discontinued)*, **2018**, 58, 181-182	1.1	10
74	Meckel syndrome: Clinical and mutation profile in six fetuses. *Clinical Genetics*, **2019**, 96, 560-565	4	9
73	Bain type of X-linked syndromic mental retardation in a male with a pathogenic variant in HNRNPH2. *American Journal of Medical Genetics, Part A*, **2020**, 182, 183-188	2.5	8
72	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	11	8
71	Genetic diversity of NDUFV1-dependent mitochondrial complex I deficiency. *European Journal of Human Genetics*, **2018**, 26, 1582-1587	5.3	8
70	Confirmation of a Rare Genetic Leukoencephalopathy due to a Novel Bi-allelic Variant in RPIA. *European Journal of Medical Genetics*, **2019**, 62, 103708	2.6	7
69	Human IFT52 mutations uncover a novel role for the protein in microtubule dynamics and centrosome cohesion. *Human Molecular Genetics*, **2019**, 28, 2720-2737	5.6	7
68	A neurodegenerative mitochondrial disease phenotype due to biallelic loss-of-function variants in PNPLA8 encoding calcium-independent phospholipase A2 *American Journal of Medical Genetics, Part A*, **2018**, 176, 1232-1237	2.5	7
67	A biallelic 36-bp insertion in PIBF1 is associated with Joubert syndrome. *Journal of Human Genetics*, **2018**, 63, 935-939	4.3	7

66	Variable presentation of Fraser syndrome in two fetuses and a novel mutation in FRAS1. *Congenital Anomalies (discontinued)*, **2017**, 57, 83-85	1.1	7
65	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. *American Journal of Human Genetics*, **2019**, 104, 439-453	11	7
64	Homozygous c.359del variant in MGME1 is associated with early onset cerebellar ataxia. *European Journal of Medical Genetics*, **2017**, 60, 533-535	2.6	6
63	Hyperphosphatasia with Mental Retardation Syndrome Due to a Novel Mutation in. *Journal of Pediatric Genetics*, **2017**, 6, 191-193	0.7	6
62	Mutations in MYLPF Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. *American Journal of Human Genetics*, **2020**, 107, 293-310	11	6
61	Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. *Bone*, **2019**, 120, 204-211	4.7	6
60	A novel mutation (c.121-13T>A) in the polypyrimidine tract of the splice acceptor site of intron 2 causes exon 3 skipping in mitochondrial acetoacetyl-CoA thiolase gene. *Molecular Medicine Reports*, **2017**, 15, 3879-3884	2.9	5
59	Homozygous variant, p.(Arg643Trp) in VAC14 causes striatonigral degeneration: report of a novel variant and review of VAC14-related disorders. *Journal of Human Genetics*, **2019**, 64, 1237-1242	4.3	5
58	Occurrence of Synpolydactyly and Omphalocele in a Fetus with a Mutation. *Journal of Pediatric Genetics*, **2017**, 6, 194-197	0.7	5
57	Identification of a novel homozygous variant confirms ITPA as a developmental and epileptic encephalopathy gene. *American Journal of Medical Genetics, Part A*, **2019**, 179, 857-861	2.5	5
56	Report of the Third Family with Multiple Mitochondrial Dysfunctions Syndrome 5 Caused by the Founder Variant p.(Glu87Lys) in. *Journal of Pediatric Genetics*, **2018**, 7, 130-133	0.7	5
55	Intrafamilial variability in syndromic microphthalmia type 5 caused by a novel variation in OTX2. *Ophthalmic Genetics*, **2017**, 38, 533-536	1.2	4
54	Recurrent bi-allelic splicing variant c.454+3A>G in TRAPPC4 is associated with progressive encephalopathy and muscle involvement. *Brain*, **2020**, 143, e29	11.2	4
53	Novel variant p.(Ala102Thr) in SDHB causes mitochondrial complex II deficiency: Case report and review of the literature. *Annals of Human Genetics*, **2020**, 84, 345-351	2.2	4
52	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	2.6	4
51	A novel bi-allelic loss-of-function variant in MYOD1: Further evidence for gene-disease association and phenotypic variability in MYOD1-related myopathy. *Clinical Genetics*, **2019**, 96, 276-277	4	4
50	Severe Form of Brachydactyly Type A1 in a Child with a c.298G > A Mutation in Gene. *Journal of Pediatric Genetics*, **2017**, 6, 177-180	0.7	4
49	Homozygous deletion of exons 2 and 3 of NPC2 associated with Niemann-Pick disease type C. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2486-9	2.5	4

48	Locus and allelic heterogeneity in five families with hereditary spastic paraplegia. *Journal of Human Genetics*, **2019**, 64, 17-21	4.3	4
47	p.Arg69Trp in RNASEH2C is a founder variant in three Indian families with Aicardi-Goutières syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 156-160	2.5	4
46	A reply to a commentary on homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. *Journal of Human Genetics*, **2017**, 62, 867	4.3	3
45	Variants in the transcriptional corepressor BCORL1 are associated with an X-linked disorder of intellectual disability, dysmorphic features, and behavioral abnormalities. *American Journal of Medical Genetics, Part A*, **2019**, 179, 870-874	2.5	3
44	Burn-McKeown syndrome with biallelic promoter type 2 deletion in TXNL4A in two siblings. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1313-1315	2.5	3
43	Pycnodysostosis: Novel Variants in and Occurrence of Giant Cell Tumor. *Journal of Pediatric Genetics*, **2018**, 7, 9-13	0.7	3
42	Further evidence for causation of ischiospinal dysostosis by a pathogenic variant in BMPER and expansion of the phenotype. *Congenital Anomalies (discontinued)*, **2019**, 59, 26-27	1.1	3
41	Clinical Variability in Familial X-Linked Ohdo Syndrome-Maat-Kievit-Brunner Type with Mutation. *Journal of Pediatric Genetics*, **2017**, 6, 198-204	0.7	3
40	Co-occurrence of a de novo Williams and 22q11.2 microdeletion syndromes. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 1927-31	2.5	3
39	Biallelic variants p.Arg1133Cys and p.Arg1379Cys in COL2A1: Further delineation of phenotypic spectrum of recessive Type 2 collagenopathies. *American Journal of Medical Genetics, Part A*, **2020**, 182, 338-347	2.5	3
38	A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for early-onset monogenic disorders in Indians. *Human Mutation*, **2021**, 42, e15-e61	4.7	3
37	Spectrum of urorectal septum malformation sequence. *Congenital Anomalies (discontinued)*, **2016**, 56, 119-26	1.1	3
36	Phenotypic variability in patients with interstitial 6q21-q22 microdeletion and Acro-Cardio-Facial syndrome. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2998-3003	2.5	3
35	GATAD2B-related intellectual disability due to parental mosaicism and review of literature. *Clinical Dysmorphology*, **2019**, 28, 190-194	0.9	3
34	Bi-allelic missense variant, p.Ser35Leu in EXOSC1 is associated with pontocerebellar hypoplasia. *Clinical Genetics*, **2021**, 99, 594-600	4	3
33	Spastic Paraplegia Type 56 in a Young Child. *Indian Journal of Pediatrics*, **2020**, 87, 650-651	3	2
32	India Allele Finder: a web-based annotation tool for identifying common alleles in next-generation sequencing data of Indian origin. *BMC Research Notes*, **2017**, 10, 233	2.3	2
31	Phenotypic diversity and genetic complexity of PAX3-related Waardenburg syndrome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2951-2958	2.5	2

30	Recurrent 1q21.1 deletion syndrome: report on variable expression, nonpenetrance and review of literature. *Clinical Dysmorphology*, **2020**, 29, 127-131	0.9	2
29	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. *American Journal of Human Genetics*, **2021**, 108, 1138-1150	11	2
28	Congenital omphalocele and cleft palate in two fetuses. *Congenital Anomalies (discontinued)*, **2016**, 56, 190-1	1.1	2
27	NAD(P)HX dehydratase (NAXD) deficiency due to a novel biallelic missense variant and review of literature. *European Journal of Medical Genetics*, **2021**, 64, 104266	2.6	2
26	Complexities in Genotype-Phenotype Correlation and Genetic Counseling in Collagen VI - Related Myopathy. *Indian Journal of Pediatrics*, **2017**, 84, 330-331	3	1
25	Biallelic c.1263dupC in DOK7 results in fetal akinesia deformation sequence. *American Journal of Medical Genetics, Part A*, **2020**, 182, 804-807	2.5	1
24	An emerging ribosomopathy affecting the skeleton due to biallelic variations in NEPRO. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1709-1717	2.5	1
23	Expanding the spectrum of syndromic PPP2R3C-related XY gonadal dysgenesis to XX gonadal dysgenesis. *Clinical Genetics*, **2021**,	4	1
22	Identification and characterization of 30 novel pathogenic variations in 69 unrelated Indian patients with Mucolipidosis Type II and Type III. *Journal of Human Genetics*, **2020**, 65, 971-984	4.3	1
21	Trichothiodystrophy type 4 in an Indian family. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2226-2229	2.5	1
20	Hedgehog acyl-transferase-related multiple congenital anomalies: Report of an additional family and delineation of the syndrome. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2756-2765	2.5	1
19	Clinical and genetic spectrum of 104 Indian families with central nervous system white matter abnormalities. *Clinical Genetics*, **2021**, 100, 542-550	4	1
18	Genetic disorders with central nervous system white matter abnormalities: An update. *Clinical Genetics*, **2021**, 99, 119-132	4	1
17	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	5.3	1
16	Clinically relevant variants in a large cohort of Indian patients with Marfan syndrome and related disorders identified by next-generation sequencing. *Scientific Reports*, **2021**, 11, 764	4.9	1
15	Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II.. *European Journal of Medical Genetics*, **2022**, 104447	2.6	0
14	Further evidence of affected females with a heterozygous variant in FGF13 causing X-linked developmental and epileptic encephalopathy 90. *European Journal of Medical Genetics*, **2021**, 65, 104403	2.6	0
13	Novel splice site and nonsense variants in INVS cause infantile nephronophthisis. *Gene*, **2020**, 729, 144229	3.8	0

12	Bosley-Salih-Alorainy syndrome in patients from India. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2699-2703	2.5	0
11	Biallelic start loss variant, c.1A> G in GCSH is associated with variant nonketotic hyperglycinemia. *Clinical Genetics*, **2021**, 100, 201-205	4	0
10	Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. *European Journal of Human Genetics*, **2021**, 29, 1226-1234	5.3	0
9	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. *Genetics in Medicine*, **2021**, 23, 2415-2425	8.1	0
8	Expanding the electro-clinical phenotype of associated neuroregression. *Epilepsy and Behavior Reports*, **2021**, 16, 100485	1.3	0
7	Second report of SHMT2 related neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities.. *European Journal of Medical Genetics*, **2022**, 104481	2.6	0
6	Late Onset Subacute Profound Biotinidase Deficiency Caused by a Novel Homozygous Variant c.466-3T>G in the BTD Gene.. *Indian Journal of Pediatrics*, **2022**, 1	3	
5	C18orf32 loss-of-function is associated with a neurodevelopmental disorder with hypotonia and contractures.. *Human Genetics*, **2022**, 1	6.3	
4	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects **2021**, 52,		
3	Novel ALOX12B Mutation Identified in Parents following Single Nucleotide Polymorphism Microarray Testing of Banked DNA from a Fatal Case of Congenital Ichthyosis. *Indian Journal of Dermatology*, **2016**, 61, 122	0.9	
2	Response to Hall et al. *American Journal of Human Genetics*, **2020**, 107, 1188-1189	11	
1	Digital clubbing as the predominant manifestation of hypertrophic osteoarthropathy caused by pathogenic variants in HPGD in three Indian families. *Clinical Dysmorphology*, **2020**, 29, 123-126	0.9	