

# Anju Shukla

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	Late Onset Subacute Profound Biotinidase Deficiency Caused by a Novel Homozygous Variant c.466-3T>G in the BTM Gene.. <i>Indian Journal of Pediatrics</i> , <b>2022</b> , 1	3	
100	C18orf32 loss-of-function is associated with a neurodevelopmental disorder with hypotonia and contractures.. <i>Human Genetics</i> , <b>2022</b> , 1	6.3	
99	Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II.. <i>European Journal of Medical Genetics</i> , <b>2022</b> , 104447	2.6	0
98	Second report of SHMT2 related neurodevelopmental disorder with cardiomyopathy, spasticity, and brain abnormalities.. <i>European Journal of Medical Genetics</i> , <b>2022</b> , 104481	2.6	0
97	Expanding the spectrum of syndromic PPP2R3C-related XY gonadal dysgenesis to XX gonadal dysgenesis. <i>Clinical Genetics</i> , <b>2021</b> ,	4	1
96	Novel Biallelic Variants in KIF21A Cause a Novel Phenotype of Fetal Akinesia with Neurodevelopmental Defects <b>2021</b> , 52,		
95	Further evidence of affected females with a heterozygous variant in FGF13 causing X-linked developmental and epileptic encephalopathy 90. <i>European Journal of Medical Genetics</i> , <b>2021</b> , 65, 104403 <sup>2.6</sup>	2.6	0
94	Hedgehog acyl-transferase-related multiple congenital anomalies: Report of an additional family and delineation of the syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2021</b> , 185, 2756-2765	2.5	1
93	A data set of variants derived from 1455 clinical and research exomes is efficient in variant prioritization for early-onset monogenic disorders in Indians. <i>Human Mutation</i> , <b>2021</b> , 42, e15-e61	4.7	3
92	Biallelic start loss variant, c.1A > G in GCSH is associated with variant nonketotic hyperglycinemia. <i>Clinical Genetics</i> , <b>2021</b> , 100, 201-205	4	0
91	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , <b>2021</b> , 108, 1138-1150	11	2
90	Pathogenic variants in PIDD1 lead to an autosomal recessive neurodevelopmental disorder with pachygyria and psychiatric features. <i>European Journal of Human Genetics</i> , <b>2021</b> , 29, 1226-1234	5.3	0
89	Clinical and genetic spectrum of 104 Indian families with central nervous system white matter abnormalities. <i>Clinical Genetics</i> , <b>2021</b> , 100, 542-550	4	1
88	Genetic disorders with central nervous system white matter abnormalities: An update. <i>Clinical Genetics</i> , <b>2021</b> , 99, 119-132	4	1
87	Biallelic mutations in the death domain of PIDD1 impair caspase-2 activation and are associated with intellectual disability. <i>Translational Psychiatry</i> , <b>2021</b> , 11, 1	8.6	66
86	Multilocus disease-causing genomic variations for Mendelian disorders: role of systematic phenotyping and implications on genetic counselling. <i>European Journal of Human Genetics</i> , <b>2021</b> , 29, 1774-1780	5.3	1
85	PPA2-associated sudden cardiac death: extending the clinical and allelic spectrum in 20 new families. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 2415-2425	8.1	0

84	NAD(P)HX dehydratase (NAXD) deficiency due to a novel biallelic missense variant and review of literature. <i>European Journal of Medical Genetics</i> , <b>2021</b> , 64, 104266	2.6	2
83	Expanding the electro-clinical phenotype of associated neuroregression. <i>Epilepsy and Behavior Reports</i> , <b>2021</b> , 16, 100485	1.3	0
82	Bi-allelic missense variant, p.Ser35Leu in EXOSC1 is associated with pontocerebellar hypoplasia. <i>Clinical Genetics</i> , <b>2021</b> , 99, 594-600	4	3
81	Clinically relevant variants in a large cohort of Indian patients with Marfan syndrome and related disorders identified by next-generation sequencing. <i>Scientific Reports</i> , <b>2021</b> , 11, 764	4.9	1
80	Burn-McKeown syndrome with biallelic promoter type 2 deletion in TXNL4A in two siblings. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 1313-1315	2.5	3
79	Recurrent bi-allelic splicing variant c.454+3A>G in TRAPPC4 is associated with progressive encephalopathy and muscle involvement. <i>Brain</i> , <b>2020</b> , 143, e29	11.2	4
78	Novel variant p.(Ala102Thr) in SDHB causes mitochondrial complex II deficiency: Case report and review of the literature. <i>Annals of Human Genetics</i> , <b>2020</b> , 84, 345-351	2.2	4
77	Spastic Paraplegia Type 56 in a Young Child. <i>Indian Journal of Pediatrics</i> , <b>2020</b> , 87, 650-651	3	2
76	Biallelic c.1263dupC in DOK7 results in fetal akinesia deformation sequence. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 804-807	2.5	1
75	Novel splice site and nonsense variants in INVS cause infantile nephronophthisis. <i>Gene</i> , <b>2020</b> , 729, 1442298	3.9	0
74	Bain type of X-linked syndromic mental retardation in a male with a pathogenic variant in HNRNPH2. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 183-188	2.5	8
73	Biallelic variants p.Arg1133Cys and p.Arg1379Cys in COL2A1: Further delineation of phenotypic spectrum of recessive Type 2 collagenopathies. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 338-347	2.5	3
72	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. <i>Brain</i> , <b>2020</b> , 143, 2929-2944	11.2	12
71	Phenotypic diversity and genetic complexity of PAX3-related Waardenburg syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 2951-2958	2.5	2
70	Identification and characterization of 30 novel pathogenic variations in 69 unrelated Indian patients with Mucopolidosis Type II and Type III. <i>Journal of Human Genetics</i> , <b>2020</b> , 65, 971-984	4.3	1
69	Response to Hall et al. <i>American Journal of Human Genetics</i> , <b>2020</b> , 107, 1188-1189	11	
68	Recurrent 1q21.1 deletion syndrome: report on variable expression, nonpenetrance and review of literature. <i>Clinical Dysmorphology</i> , <b>2020</b> , 29, 127-131	0.9	2
67	Mutations in MYLFP Cause a Novel Segmental Amyoplasia that Manifests as Distal Arthrogryposis. <i>American Journal of Human Genetics</i> , <b>2020</b> , 107, 293-310	11	6

66	Digital clubbing as the predominant manifestation of hypertrophic osteoarthropathy caused by pathogenic variants in HPGD in three Indian families. <i>Clinical Dysmorphology</i> , <b>2020</b> , 29, 123-126	0.9	
65	Bosley-Salih-Alorainy syndrome in patients from India. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 2699-2703	2.5	0
64	Trichothiodystrophy type 4 in an Indian family. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 2226-2229	2.5	1
63	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. <i>Journal of Human Genetics</i> , <b>2019</b> , 64, 1173-1186	4.3	20
62	Confirmation of a Rare Genetic Leukoencephalopathy due to a Novel Bi-allelic Variant in RPIA. <i>European Journal of Medical Genetics</i> , <b>2019</b> , 62, 103708	2.6	7
61	Human IFT52 mutations uncover a novel role for the protein in microtubule dynamics and centrosome cohesion. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 2720-2737	5.6	7
60	Variants in the transcriptional corepressor BCORL1 are associated with an X-linked disorder of intellectual disability, dysmorphic features, and behavioral abnormalities. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 870-874	2.5	3
59	Gaucher disease: single gene molecular characterization of one-hundred Indian patients reveals novel variants and the most prevalent mutation. <i>BMC Medical Genetics</i> , <b>2019</b> , 20, 31	2.1	17
58	Further evidence for causation of ischiopspinal dysostosis by a pathogenic variant in BMPER and expansion of the phenotype. <i>Congenital Anomalies (discontinued)</i> , <b>2019</b> , 59, 26-27	1.1	3
57	Bi-allelic c.181_183delTGT in BTB domain of KLHL7 is associated with overlapping phenotypes of Crisponi/CISS1-like and Bohring-Opitz like syndrome. <i>European Journal of Medical Genetics</i> , <b>2019</b> , 62, 103528	2.6	4
56	Meckel syndrome: Clinical and mutation profile in six fetuses. <i>Clinical Genetics</i> , <b>2019</b> , 96, 560-565	4	9
55	An emerging ribosomopathy affecting the skeleton due to biallelic variations in NEPRO. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 1709-1717	2.5	1
54	A novel bi-allelic loss-of-function variant in MYOD1: Further evidence for gene-disease association and phenotypic variability in MYOD1-related myopathy. <i>Clinical Genetics</i> , <b>2019</b> , 96, 276-277	4	4
53	The Genomics of Arthrogyrosis, a Complex Trait: Candidate Genes and Further Evidence for Oligogenic Inheritance. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 132-150	11	50
52	Homozygous variant, p.(Arg643Trp) in VAC14 causes striatonigral degeneration: report of a novel variant and review of VAC14-related disorders. <i>Journal of Human Genetics</i> , <b>2019</b> , 64, 1237-1242	4.3	5
51	Identification of a novel homozygous variant confirms ITPA as a developmental and epileptic encephalopathy gene. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 857-861	2.5	5
50	Hypomorphic Mutations in TONSL Cause SPONASTRIME Dysplasia. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 439-453	11	7
49	GATAD2B-related intellectual disability due to parental mosaicism and review of literature. <i>Clinical Dysmorphology</i> , <b>2019</b> , 28, 190-194	0.9	3

48	NAD(P)HX dehydratase (NAXD) deficiency: a novel neurodegenerative disorder exacerbated by febrile illnesses. <i>Brain</i> , <b>2019</b> , 142, 50-58	11.2	31
47	Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. <i>Bone</i> , <b>2019</b> , 120, 204-211	4.7	6
46	Locus and allelic heterogeneity and phenotypic variability in Waardenburg syndrome. <i>Clinical Genetics</i> , <b>2019</b> , 95, 398-402	4	13
45	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , <b>2019</b> , 179, 150-158	2.5	25
44	Locus and allelic heterogeneity in five families with hereditary spastic paraplegia. <i>Journal of Human Genetics</i> , <b>2019</b> , 64, 17-21	4.3	4
43	The homozygous variant c.797G>A/p.(Cys266Tyr) in PISD is associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. <i>Human Mutation</i> , <b>2019</b> , 40, 299-309	4.7	33
42	Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. <i>European Journal of Human Genetics</i> , <b>2018</b> , 26, 695-708	5.3	14
41	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. <i>Bone</i> , <b>2018</b> , 110, 368-377	4.7	21
40	Williams-Beuren syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1128-1136	2.5	31
39	A neurodegenerative mitochondrial disease phenotype due to biallelic loss-of-function variants in PNPLA8 encoding calcium-independent phospholipase A2. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 1232-1237	2.5	7
38	A biallelic 36-bp insertion in PIBF1 is associated with Joubert syndrome. <i>Journal of Human Genetics</i> , <b>2018</b> , 63, 935-939	4.3	7
37	Pycnodysostosis: Novel Variants in and Occurrence of Giant Cell Tumor. <i>Journal of Pediatric Genetics</i> , <b>2018</b> , 7, 9-13	0.7	3
36	Report of four novel variants in ASNS causing asparagine synthetase deficiency and review of literature. <i>Congenital Anomalies (discontinued)</i> , <b>2018</b> , 58, 181-182	1.1	10
35	p.Arg69Trp in RNASEH2C is a founder variant in three Indian families with Aicardi-Goutières syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2018</b> , 176, 156-160	2.5	4
34	Homozygosity for a nonsense variant in AIMP2 is associated with a progressive neurodevelopmental disorder with microcephaly, seizures, and spastic quadriplegia. <i>Journal of Human Genetics</i> , <b>2018</b> , 63, 19-25	4.3	21
33	Report of the Third Family with Multiple Mitochondrial Dysfunctions Syndrome 5 Caused by the Founder Variant p.(Glu87Lys) in. <i>Journal of Pediatric Genetics</i> , <b>2018</b> , 7, 130-133	0.7	5
32	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. <i>American Journal of Human Genetics</i> , <b>2018</b> , 103, 948-967 <sup>11</sup>		8
31	RSPO2 inhibition of RNF43 and ZNRF3 governs limb development independently of LGR4/5/6. <i>Nature</i> , <b>2018</b> , 557, 564-569	50.4	97

30	Genetic diversity of NDUFV1-dependent mitochondrial complex I deficiency. <i>European Journal of Human Genetics</i> , <b>2018</b> , 26, 1582-1587	5.3	8
29	A novel sequence variant in SFRP4 causing Pyle disease. <i>Journal of Human Genetics</i> , <b>2017</b> , 62, 575-576	4.3	19
28	Additional three patients with Smith-McCort dysplasia due to novel RAB33B mutations. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 588-595	2.5	12
27	Autosomal recessive spinocerebellar ataxia 20: Report of a new patient and review of literature. <i>European Journal of Medical Genetics</i> , <b>2017</b> , 60, 118-123	2.6	26
26	A reply to a commentary on homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. <i>Journal of Human Genetics</i> , <b>2017</b> , 62, 867	4.3	3
25	Homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. <i>Journal of Human Genetics</i> , <b>2017</b> , 62, 723-727	4.3	45
24	Intrafamilial variability in syndromic microphthalmia type 5 caused by a novel variation in OTX2. <i>Ophthalmic Genetics</i> , <b>2017</b> , 38, 533-536	1.2	4
23	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 117-127	11	39
22	Complexities in Genotype-Phenotype Correlation and Genetic Counseling in Collagen VI - Related Myopathy. <i>Indian Journal of Pediatrics</i> , <b>2017</b> , 84, 330-331	3	1
21	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. <i>Molecular Medicine Reports</i> , <b>2017</b> , 15, 3879-3884	2.9	5
20	Severe Form of Brachydactyly Type A1 in a Child with a c.298G > A Mutation in Gene. <i>Journal of Pediatric Genetics</i> , <b>2017</b> , 6, 177-180	0.7	4
19	Homozygous c.359del variant in MGME1 is associated with early onset cerebellar ataxia. <i>European Journal of Medical Genetics</i> , <b>2017</b> , 60, 533-535	2.6	6
18	Noonan syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 2323-2334	2.3	43
17	Hyperphosphatasia with Mental Retardation Syndrome Due to a Novel Mutation in. <i>Journal of Pediatric Genetics</i> , <b>2017</b> , 6, 191-193	0.7	6
16	Clinical Variability in Familial X-Linked Ohdo Syndrome-Maat-Kievit-Brunner Type with Mutation. <i>Journal of Pediatric Genetics</i> , <b>2017</b> , 6, 198-204	0.7	3
15	Occurrence of Synpolydactyly and Omphalocele in a Fetus with a Mutation. <i>Journal of Pediatric Genetics</i> , <b>2017</b> , 6, 194-197	0.7	5
14	India Allele Finder: a web-based annotation tool for identifying common alleles in next-generation sequencing data of Indian origin. <i>BMC Research Notes</i> , <b>2017</b> , 10, 233	2.3	2
13	Identification of a novel LRRK1 mutation in a family with osteosclerotic metaphyseal dysplasia. <i>Journal of Human Genetics</i> , <b>2017</b> , 62, 437-441	4.3	21

12	Variable presentation of Fraser syndrome in two fetuses and a novel mutation in FRAS1. <i>Congenital Anomalies (discontinued)</i> , <b>2017</b> , 57, 83-85	1.1	7
11	Biallelic Loss of Proprioception-Related PIEZO2 Causes Muscular Atrophy with Perinatal Respiratory Distress, Arthrogyrosis, and Scoliosis. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 1206-1216	1.1	4 <sup>1</sup>
10	Novel ALOX12B Mutation Identified in Parents following Single Nucleotide Polymorphism Microarray Testing of Banked DNA from a Fatal Case of Congenital Ichthyosis. <i>Indian Journal of Dermatology</i> , <b>2016</b> , 61, 122	0.9	
9	Homozygous deletion of exons 2 and 3 of NPC2 associated with Niemann-Pick disease type C. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170, 2486-9	2.5	4
8	Congenital omphalocele and cleft palate in two fetuses. <i>Congenital Anomalies (discontinued)</i> , <b>2016</b> , 56, 190-1	1.1	2
7	Spectrum of urorectal septum malformation sequence. <i>Congenital Anomalies (discontinued)</i> , <b>2016</b> , 56, 119-26	1.1	3
6	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170A, 410-417	2.5	18
5	Phenotypic variability in patients with interstitial 6q21-q22 microdeletion and Acro-Cardio-Facial syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170, 2998-3003	2.5	3
4	Recurrent and novel GLB1 mutations in India. <i>Gene</i> , <b>2015</b> , 567, 173-81	3.8	17
3	Clinical utility of fetal autopsy and its impact on genetic counseling. <i>Prenatal Diagnosis</i> , <b>2015</b> , 35, 685-91	3.2	25
2	Novel and recurrent mutations in WISP3 and an atypical phenotype. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167A, 2481-4	2.5	17
1	Co-occurrence of a de novo Williams and 22q11.2 microdeletion syndromes. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167A, 1927-31	2.5	3