Meena Balasubramanian

List of Publications by Year
in descending order

116	4,920	279798 23	114465 63
papers	citations	h-index	g-index

124	124	124	10082
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Prevalence and architecture of de novo mutations in developmental disorders. Nature, 2017, 542, 433-438.	27.8	1,211
2	Large-scale discovery of novel genetic causes of developmental disorders. Nature, 2015, 519, 223-228.	27.8	998
3	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
4	Clinical delineation and natural history of the PIK3CAâ€related overgrowth spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 1713-1733.	1.2	249
5	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
6	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
7	Genotype-Phenotype Correlation in NF1: Evidence for a More Severe Phenotype Associated with Missense Mutations Affecting NF1 Codons 844â€“848. American Journal of Human Genetics, 2018, 102, 69-87.	6.2	144
8	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	21.4	133
9	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	2.4	99
10	Case series: 2q33.1 microdeletion syndrome--further delineation of the phenotype. Journal of Medical Genetics, 2011, 48, 290-298.	3.2	59
11	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	1.2	57
12	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
13	Compound heterozygous variants in NBAS as a cause of atypical osteogenesis imperfecta. Bone, 2017, 94, 65-74.	2.9	54
14	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	6.2	45
15	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	7.6	43
16	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
17	Delineating the phenotypic spectrum of Bainbridge-Ropers syndrome: 12 new patients withde novo, heterozygous, loss-of-function mutations inASXL3and review of published literature. Journal of Medical Genetics, 2017, 54, 537-543.	3.2	39
18	De novo mutations in HNRNPU result in a neurodevelopmental syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3003-3012.	1.2	38

#	Article	IF	Citations
19	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	2.5	38
20	Genotypeâ€“phenotype study in type V osteogenesis imperfecta. Clinical Dysmorphology, 2013, 22, 93-101.	0.3	34
21	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	12.8	33
22	Phenotypic variability in patients with osteogenesis imperfecta caused by BMP1 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3150-3156.	1.2	32
23	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. Genetics in Medicine, 2020, 22, 1498-1506.	2.4	31
24	Intronic ITGA3 Mutation Impacts Splicing Regulation and Causes Interstitial Lung Disease, Nephrotic Syndrome, and Epidermolysis Bullosa. Journal of Investigative Dermatology, 2016, 136, 1056-1059.	0.7	29
25	Novel PLS3 variants in Xâ€linked osteoporosis: Exploring bone material properties. American Journal of Medical Genetics, Part A, 2018, 176, 1578-1586.	1.2	29
26	Phenotypic Spectrum in Osteogenesis Imperfecta Due to Mutations in TMEM38B: Unraveling a Complex Cellular Defect. Journal of Clinical Endocrinology and Metabolism, 2017, 102, 2019-2028.	3.6	27
27	SLC35A2-related congenital disorder of glycosylation: Defining the phenotype. European Journal of Paediatric Neurology, 2018, 22, 1095-1102.	1.6	27
28	IMAGe syndrome: Case report with a previously unreported feature and review of published literature. American Journal of Medical Genetics, Part A, 2010, 152A, 3138-3142.	1.2	24
29	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
30	P4HB recurrent missense mutation causing Cole-Carpenter syndrome. Journal of Medical Genetics, 2018, 55, 158-165.	3.2	20
31	Deep phenotyping of 14 new patients with IQSEC2 variants, including monozygotic twins of discordant phenotype. Clinical Genetics, 2019, 95, 496-506.	2.0	20
32	ZMYND11 â€related syndromic intellectual disability: 16 patients delineating and expanding the phenotypic spectrum. Human Mutation, 2020, 41, 1042-1050.	2.5	20
33	CRTAP mutation in a patient with Coleâ€Carpenter syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 587-591.	1.2	19
34	Clinical findings of 21 previously unreported probands with HNRNPUâ€related syndrome and comprehensive literature review. American Journal of Medical Genetics, Part A, 2020, 182, 1637-1654.	1.2	19
35	Chitayat syndrome: hyperphalangism, characteristic facies, hallux valgus and bronchomalacia results from a recurrent c.266A>G p.(Tyr89Cys) variant in theERFgene. Journal of Medical Genetics, 2017, 54, 157-165.	3.2	18
36	De novo SETD5 lossâ€ofâ€function variant as a cause for intellectual disability in a 10â€year old boy with an aberrant blind ending bronchus. American Journal of Medical Genetics, Part A, 2017, 173, 3165-3171.	1.2	18

#	Article	IF	Citations
37	Aortic aneurysm/dissection and osteogenesis imperfecta: Four new families and review of the literature. Bone, 2019, 121, 191-195.	2.9	18
38	Zimmermannâ€“Laband syndrome in a child previously described with brachydactyly, extrahepatic biliary atresia, patent ductus arteriosus and seizures. Clinical Dysmorphology, 2010, 19, 48-50.	0.3	17
39	Clinical and biochemical characteristics of adults with hypophosphatasia attending a metabolic bone clinic. Bone, 2021, 144, 115795.	2.9	17
40	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
41	Pancreatic hypoplasia presenting with neonatal diabetes mellitus in association with congenital heart defect and developmental delay. American Journal of Medical Genetics, Part A, 2010, 152A, 340-346.	1.2	16
42	An emerging, recognizable facial phenotype in association with mutations in GLIâ€similar 3 (GLIS3). American Journal of Medical Genetics, Part A, 2016, 170, 1918-1923.	1.2	16
43	A novel homozygous variant in SERPINH1 associated with a severe, lethal presentation of osteogenesis imperfecta with hydranencephaly. Gene, 2016, 595, 49-52.	2.2	16
44	Inverted duplication of 1q32.1 to 1q44 characterized by array CGH and review of distal 1q partial trisomy. American Journal of Medical Genetics, Part A, 2009, 149A, 793-797.	1.2	15
45	Clinical report: An interstitial deletion of 16p13.11 detected by array CGH in a patient with infantile spasms. European Journal of Medical Genetics, 2011, 54, 314-318.	1.3	15
46	Aplasia cutis congenita, terminal limb defects and periventricular leukomalacia in one sibling with minor findings in the other-probable autosomal recessive Adams-Oliver Syndrome. European Journal of Medical Genetics, 2009, 52, 234-238.	1.3	13
47	Ultrastructural and histological findings on examination of skin in osteogenesis imperfecta. Clinical Dysmorphology, 2015, 24, 45-54.	0.3	13
48	Expanding the molecular basis and phenotypic spectrum of ZDHHC9â€associated Xâ€linked intellectual disability. American Journal of Medical Genetics, Part A, 2018, 176, 1238-1244.	1.2	13
49	MAN1B-CDG: Novel variants with a distinct phenotype and review of literature. European Journal of Medical Genetics, 2019, 62, 109-114.	1.3	13
50	Expanding the genotypeâ€“phenotype correlation of de novo heterozygous missense variants in YWHAG as a cause of developmental and epileptic encephalopathy. American Journal of Medical Genetics, Part A, 2020, 182, 713-720.	1.2	13
51	Growth, development, and phenotypic spectrum of individuals with deletions of 2q33.1 involving SATB2. Clinical Genetics, 2021, 99, 547-557.	2.0	13
52	Osteogenesis imperfecta: Ultrastructural and histological findings on examination of skin revealing novel insights into genotype-phenotype correlation. Ultrastructural Pathology, 2016, 40, 71-76.	0.9	12
53	Childhood-onset generalized epilepsy in Bainbridge-Ropers syndrome. Epilepsy Research, 2018, 140, 166-170.	1.6	12
54	Expanding the phenotype of <scp>ASXL3</scp>â€related syndrome: A comprehensive description of 45 unpublished individuals with inherited and de novo pathogenic variants in <scp>ASXL3</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 3446-3458.	1.2	12

#	Article	IF	Citations
55	Type 1 collagenopathy presenting with a Russellâ€“Silver phenotype. American Journal of Medical Genetics, Part A, 2011, 155, 1414-1418.	1.2	11
56	Mosaic trisomy 11 in a fetus with bilateral renal agenesis. Clinical Dysmorphology, 2011, 20, 47-49.	0.3	11
57	Inherited duplication of the short arm of chromosome 18p11.32â€“p11.31 associated with developmental delay/intellectual disability. Clinical Dysmorphology, 2016, 25, 19-22.	0.3	11
58	SHANK3 variant as a cause of nonsyndromal autism in an 11-year-old boy and a review of published literature. Clinical Dysmorphology, 2018, 27, 113-115.	0.3	11
59	Recurrent rhabdomyolysis caused by carnitine palmitoyltransferase II deficiency, common but under-recognised: Lessons to be learnt. Molecular Genetics and Metabolism Reports, 2018, 15, 69-70.	1.1	11
60	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. Journal of Clinical Immunology, 2021, 41, 1781-1793.	3.8	10
61	Mosaicism in ASXL3-related syndrome: Description of five patients from three families. European Journal of Medical Genetics, 2020, 63, 103925.	1.3	9
62	Autism and heritable bone fragility: A true association?. Bone Reports, 2018, 8, 156-162.	0.4	8
63	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
64	Exploring the association between SRPX2 variants and neurodevelopment: How causal is it?. Gene, 2019, 685, 50-54.	2.2	8
65	Expanding the phenotype of SPARC-related osteogenesis imperfecta: clinical findings in two patients with pathogenic variants in SPARC and literature review. Journal of Medical Genetics, 2022, 59, 810-816.	3.2	8
66	Patientsâ€™ priorities and expectations on an EU registry for rare bone and mineral conditions. Orphanet Journal of Rare Diseases, 2021, 16, 463.	2.7	7
67	A novel de novo 20q13.32â€“q13.33 deletion in a 2-year-old child with poor growth, feeding difficulties and low bone mass. Journal of Human Genetics, 2015, 60, 313-317.	2.3	6
68	Short case report: Xq23 deletion involving PAK3 as a novel cause of developmental delay in a 6-year-old boy. Clinical Dysmorphology, 2017, 26, 38-40.	0.3	6
69	Functional mRNA analysis reveals aberrant splicing caused by novel intronic mutation in WDR45 in NBIA patient. American Journal of Medical Genetics, Part A, 2018, 176, 1049-1054.	1.2	6
70	Expanding the phenotype of <scp>HNRNPU</scp>â€related neurodevelopmental disorder with emphasis on seizure phenotype and review of literature. American Journal of Medical Genetics, Part A, 2022, 188, 1497-1514.	1.2	6
71	A Novel (Paternally Inherited) Duplication 13q31.3q32.3 in a 12-Year-Old Patient with Facial Dysmorphism and Developmental Delay. Molecular Syndromology, 2014, 5, 245-250.	0.8	5
72	Atypical, milder presentation in a child with CC2D2A and KIDINS220 variants. Clinical Dysmorphology, 2020, 29, 10-16.	0.3	5

#	Article	IF	Citations
73	<scp>ZMYND11</scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	2.0	5
74	Cerebral cavernous malformation: Clinical report of two families with variable phenotype associated with KRIT1 mutation. European Journal of Paediatric Neurology, 2013, 17, 661-665.	1.6	4
75	Copy number variants in association with type 1 collagenopathy: Atypical osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2016, 170, 476-481.	1.2	4
76	Atypical osteogenesis imperfecta caused by a 17q21.33 deletion involving COL1A1. Clinical Dysmorphology, 2017, 26, 228-230.	0.3	4
77	Expanding the phenotype of <scp>SETD5</scp>â€related disorder and presenting a novel association with bone fragility. Clinical Genetics, 2021, 100, 352-354.	2.0	4
78	Pneumothorax from subpleural blebsâ€"A new association of sotos syndrome?. American Journal of Medical Genetics, Part A, 2014, 164, 1222-1226.	1.2	3
79	Clinical report. Clinical Dysmorphology, 2015, 24, 151-155.	0.3	3
80	Tigroid pattern of cerebral white matter involvement in chromosome 6p25 deletion syndrome with concomitant 5p15 duplication. Journal of Pediatric Genetics, 2015, 01, 247-252.	0.7	3
81	Compound heterozygous variants in IFT140 as a cause of nonsyndromic retinitis pigmentosa. Ophthalmic Genetics, 2018, 39, 286-287.	1.2	3
82	Osteogenesis imperfecta type I: The role of deep phenotyping in a patient with a ruptured uterus. European Journal of Medical Genetics, 2020, 63, 104095.	1.3	3
83	Further delineation of phenotypic spectrum of <scp>SCN2A</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 867-877.	1.2	3
84	Diagnostic conundrums in antenatal presentation of a skeletal dysplasia with description of a heterozygous Câ€propeptide mutation in COL1A1 associated with a severe presentation of osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2016, 170, 3303-3307.	1.2	2
85	Clinical and molecular characterization of the first familial report of 1p32 microdeletion. Clinical Dysmorphology, 2018, 27, 36-41.	0.3	2
86	Developing pathways to clarify pathogenicity of unclassified variants in Osteogenesis Imperfecta genetic analysis. Molecular Genetics & Genomic Medicine, 2019, 7, e912.	1.2	2
87	Non-collagen pathogenic variants resulting in the osteogenesis imperfecta phenotype in children: a single-country observational cohort study. Archives of Disease in Childhood, 2022, 107, 486-490.	1.9	2
88	Pattern of Clinical Genetics Referral Following Perinatal Postmortems. Pediatric and Developmental Pathology, 2012, 15, 478-486.	1.0	1
89	Short Sternum: Feature of Trisomy Chromosome 7 and a New Association?. Pediatric and Developmental Pathology, 2014, 17, 70-72.	1.0	1
90	Congenital myotonic dystrophy. Clinical Dysmorphology, 2014, 23, 127-129.	0.3	1

#	Article	IF	Citations
91	Biallelic variants in <scp>GLE1</scp> with survival beyond neonatal period. Clinical Genetics, 2020, 98, 622-625.	2.0	1
92	Rough endoplasmic reticulum expansion: a consistent finding in a patient cohort with vascular Ehlers-Danlos Syndrome and Osteogenesis Imperfecta. Ultrastructural Pathology, 2021, 45, 1-7.	0.9	1
93	Report of two children with global developmental delay in association with de novo TLK2 variant and literature review. American Journal of Medical Genetics, Part A, 2021, , .	1.2	1
94	Further evidence for attenuated phenotype with variants in the BMPER gene causing DSD: Case report and literature review. European Journal of Medical Genetics, 2022, 65, 104470.	1.3	1
95	A novel homozygous keratin 10 mutation in siblings with autosomal recessive epidermolytic ichthyosis. Expert Review of Dermatology, 2010, 5, 519-523.	0.3	0
96	Is this a syndrome? Patterns in genetic conditions. , 0, , 105-119.		0
97	Clinical and Molecular Heterogeneity of Osteogenesis Imperfecta. Colloquium Series on Genomic and Molecular Medicine, 2017, 6, 1-63.	0.2	0
98	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
99	Clinical report follow up: Type 1 Collagenopathy presenting with a Russellâ€“Silver phenotype. American Journal of Medical Genetics, Part A, 2018, 179, 139-140.	1.2	0
100	Response to Finsterer: CPT-II deficiency needs to be detected in army personnel. Molecular Genetics and Metabolism Reports, 2018, 16, 12.	1.1	0
101	Dual diagnosis causing severe phenotype in a patient with Angelman syndrome. Clinical Dysmorphology, 2019, 28, 158-161.	0.3	0
102	Osteogenesis imperfecta type 1 with an incidental finding of bilateral radioulnar synostosis. Clinical Dysmorphology, 2020, 29, 155-157.	0.3	0
103	Phenotype-genotype correlation and role of ancillary investigations in atypical and rare forms of osteogenesis imperfecta. Bone Abstracts, 0, , .	0.0	0
104	Skeletal and bone material phenotype in recessive osteogenesis imperfecta due to a novel homozygous point mutation in TMEM38B. Bone Abstracts, 0, , .	0.0	0
105	Cole-Carpenter syndrome. Bone Abstracts, 0, , .	0.0	0
106	In-depth phenotyping including analyses of skin connective tissue in osteogenesis imperfecta. Bone Abstracts, 0, , .	0.0	0
107	Bone histomorphometry in patients withTMEM38Bmutations suggests a novel patho-mechanism leading to increased bone fragility. Endocrine Abstracts, 0, , .	0.0	0
108	Osteogenesis imperfecta type VI presenting as suspected physical abuse -- a report of two cases. Bone Abstracts, 0, , .	0.0	0

#	Article	IF	Citations
109	NBAS variants causing a novel form of inherited bone fragility. Bone Abstracts, 0, , .	0.0	0
110	P4HB recurrent missense mutation causing Cole-Carpenter syndrome: exploring the underlying mechanism. Bone Abstracts, 0, , .	0.0	0
111	Dual diagnosis of autism and osteogenesis imperfecta: Case examples to illustrate the implications of dual diagnosis for enhanced outcomes for child and family. Bone Abstracts, 0, , .	0.0	0
112	Radiographic evidence of zoledronic acid given during pregnancy - a case report. Bone Abstracts, 0, , .	0.0	0
113	Identifying the role of NBAS in bone fragility using zebrafish and exploring therapeutic targets to reverse NBAS activity. Bone Abstracts, 0, , .	0.0	0
114	An Emerging Link between Brain and Bone: Patient Report with a Pathogenic SCN9A Variant. SSRN Electronic Journal, 0, , .	0.4	0
115	Uniparental disomy as a mechanism for X-linked chondrodysplasia punctata. Clinical Dysmorphology, 2022, Publish Ahead of Print, .	0.3	0
116	First-line genome sequencing is here to stay, but how crucial is clinical phenotyping going to be?. BMJ Case Reports, 2022, 15, e247238.	0.5	0