Meena Balasubramanian

List of Publications by Year
in descending order

116	4,920	279487 23	114278 63
papers	citations	h-index	g-index

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

#	Article	IF	Citations
1	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.	1.5	8
2	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.	1.0	2
3	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.	0.7	6
4	Uniparental disomy as a mechanism for X-linked chondrodysplasia punctata. Clinical Dysmorphology, 2022, Publish Ahead of Print, .	0.1	0
5	First-line genome sequencing is here to stay, but how crucial is clinical phenotyping going to be?. BMJ Case Reports, 2022, 15, e247238.	0.2	0
6	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.	0.7	1
7	Further delineation of phenotypic spectrum of <scp>SCN2A</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 867-877.	0.7	3
8	Clinical and biochemical characteristics of adults with hypophosphatasia attending a metabolic bone clinic. Bone, 2021, 144, 115795.	1.4	17
9	Growth, development, and phenotypic spectrum of individuals with deletions of 2q33.1 involving SATB2. Clinical Genetics, 2021, 99, 547-557.	1.0	13
10	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.	1.4	17
11	Expanding the phenotype of <scp>SETD5</scp>â€related disorder and presenting a novel association with bone fragility. Clinical Genetics, 2021, 100, 352-354.	1.0	4
12	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.	2.6	42
13	<scp>ZMYND11</scp> variants are a novel cause of centrotemporal and generalised epilepsies with neurodevelopmental disorder. Clinical Genetics, 2021, 100, 412-429.	1.0	5
14	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.	0.7	12
15	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. Journal of Clinical Immunology, 2021, 41, 1781-1793.	2.0	10
16	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.	0.4	1
17	The contribution of X-linked coding variation to severe developmental disorders. Nature Communications, 2021, 12, 627.	5.8	33
18	Patientsâ€™ priorities and expectations on an EU registry for rare bone and mineral conditions. Orphanet Journal of Rare Diseases, 2021, 16, 463.	1.2	7

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19	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, , .	0.7	1
20	Atypical, milder presentation in a child with CC2D2A and KIDINS220 variants. Clinical Dysmorphology, 2020, 29, 10-16.	0.1	5
21	Osteogenesis imperfecta type 1 with an incidental finding of bilateral radioulnar synostosis. Clinical Dysmorphology, 2020, 29, 155-157.	0.1	0
22	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	13.7	343
23	Osteogenesis imperfecta type I: The role of deep phenotyping in a patient with a ruptured uterus. European Journal of Medical Genetics, 2020, 63, 104095.	0.7	3
24	Biallelic variants in <scp>GLE1</scp> with survival beyond neonatal period. Clinical Genetics, 2020, 98, 622-625.	1.0	1
25	SMAD6 variants in craniosynostosis: genotype and phenotype evaluation. Genetics in Medicine, 2020, 22, 1498-1506.	1.1	31
26	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	1.8	24
27	Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genetics in Medicine, 2020, 22, 1005-1014.	1.1	99
28	ZMYND11 â€related syndromic intellectual disability: 16 patients delineating and expanding the phenotypic spectrum. Human Mutation, 2020, 41, 1042-1050.	1.1	20
29	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.	0.7	13
30	Mosaicism in ASXL3-related syndrome: Description of five patients from three families. European Journal of Medical Genetics, 2020, 63, 103925.	0.7	9
31	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.	0.7	19
32	MAN1B-CDG: Novel variants with a distinct phenotype and review of literature. European Journal of Medical Genetics, 2019, 62, 109-114.	0.7	13
33	Cohesin complex-associated holoprosencephaly. Brain, 2019, 142, 2631-2643.	3.7	43
34	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	2.6	8
35	Developing pathways to clarify pathogenicity of unclassified variants in Osteogenesis Imperfecta genetic analysis. Molecular Genetics & Genomic Medicine, 2019, 7, e912.	0.6	2
36	Aortic aneurysm/dissection and osteogenesis imperfecta: Four new families and review of the literature. Bone, 2019, 121, 191-195.	1.4	18

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37	Deep phenotyping of 14 new patients with IQSEC2 variants, including monozygotic twins of discordant phenotype. Clinical Genetics, 2019, 95, 496-506.	1.0	20
38	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	2.6	56
39	Mutation update for the SATB2 gene. Human Mutation, 2019, 40, 1013-1029.	1.1	38
40	Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. American Journal of Human Genetics, 2019, 104, 948-956.	2.6	45
41	Dual diagnosis causing severe phenotype in a patient with Angelman syndrome. Clinical Dysmorphology, 2019, 28, 158-161.	0.1	0
42	Exploring the association between SRPX2 variants and neurodevelopment: How causal is it?. Gene, 2019, 685, 50-54.	1.0	8
43	Childhood-onset generalized epilepsy in Bainbridge-Ropers syndrome. Epilepsy Research, 2018, 140, 166-170.	0.8	12
44	Autism and heritable bone fragility: A true association?. Bone Reports, 2018, 8, 156-162.	0.2	8
45	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.	0.7	6
46	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.	0.7	13
47	Natural history and genotypeâ€phenotype correlations in 72 individuals with SATB2â€associated syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 925-935.	0.7	57
48	P4HB recurrent missense mutation causing Cole-Carpenter syndrome. Journal of Medical Genetics, 2018, 55, 158-165.	1.5	20
49	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.	2.6	144
50	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
51	Cover Image, Volume 176A, Number 4, April 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	0.7	0
52	Clinical and molecular characterization of the first familial report of 1p32 microdeletion. Clinical Dysmorphology, 2018, 27, 36-41.	0.1	2
53	Compound heterozygous variants in IFT140 as a cause of nonsyndromic retinitis pigmentosa. Ophthalmic Genetics, 2018, 39, 286-287.	0.5	3
54	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.	0.1	11

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55	Clinical report follow up: Type 1 Collagenopathy presenting with a Russellâ€“Silver phenotype. American Journal of Medical Genetics, Part A, 2018, 179, 139-140.	0.7	0
56	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	6.0	158
57	SLC35A2-related congenital disorder of glycosylation: Defining the phenotype. European Journal of Paediatric Neurology, 2018, 22, 1095-1102.	0.7	27
58	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.	0.4	11
59	Response to Finsterer: CPT-II deficiency needs to be detected in army personnel. Molecular Genetics and Metabolism Reports, 2018, 16, 12.	0.4	0
60	Novel PLS3 variants in Xâ€linked osteoporosis: Exploring bone material properties. American Journal of Medical Genetics, Part A, 2018, 176, 1578-1586.	0.7	29
61	Prevalence and architecture of de novo mutations in developmental disorders. Nature, 2017, 542, 433-438.	13.7	1,211
62	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.	1.5	18
63	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.	1.5	39
64	Clinical and Molecular Heterogeneity of Osteogenesis Imperfecta. Colloquium Series on Genomic and Molecular Medicine, 2017, 6, 1-63.	0.2	0
65	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.	1.8	27
66	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.	0.7	18
67	De novo mutations in HNRNPU result in a neurodevelopmental syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 3003-3012.	0.7	38
68	Atypical osteogenesis imperfecta caused by a 17q21.33 deletion involving COL1A1. Clinical Dysmorphology, 2017, 26, 228-230.	0.1	4
69	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.	0.1	6
70	Compound heterozygous variants in NBAS as a cause of atypical osteogenesis imperfecta. Bone, 2017, 94, 65-74.	1.4	54
71	Inherited duplication of the short arm of chromosome 18p11.32â€“p11.31 associated with developmental delay/intellectual disability. Clinical Dysmorphology, 2016, 25, 19-22.	0.1	11
72	Copy number variants in association with type 1 collagenopathy: Atypical osteogenesis imperfecta. American Journal of Medical Genetics, Part A, 2016, 170, 476-481.	0.7	4

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73	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.	0.7	16
74	Intronic ITGA3 Mutation Impacts Splicing Regulation and Causes Interstitial Lung Disease, Nephrotic Syndrome, and Epidermolysis Bullosa. Journal of Investigative Dermatology, 2016, 136, 1056-1059.	0.3	29
75	Phenotypic variability in patients with osteogenesis imperfecta caused by BMP1 mutations. American Journal of Medical Genetics, Part A, 2016, 170, 3150-3156.	0.7	32
76	A novel homozygous variant in SERPINH1 associated with a severe, lethal presentation of osteogenesis imperfecta with hydranencephaly. Gene, 2016, 595, 49-52.	1.0	16
77	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.	0.7	2
78	Osteogenesis imperfecta: Ultrastructural and histological findings on examination of skin revealing novel insights into genotype-phenotype correlation. Ultrastructural Pathology, 2016, 40, 71-76.	0.4	12
79	CRTAP mutation in a patient with Coleâ€Carpenter syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 587-591.	0.7	19
80	Clinical report. Clinical Dysmorphology, 2015, 24, 151-155.	0.1	3
81	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.	1.1	6
82	Ultrastructural and histological findings on examination of skin in osteogenesis imperfecta. Clinical Dysmorphology, 2015, 24, 45-54.	0.1	13
83	Tigroid pattern of cerebral white matter involvement in chromosome 6p25 deletion syndrome with concomitant 5p15 duplication. Journal of Pediatric Genetics, 2015, 01, 247-252.	0.3	3
84	Discovery of four recessive developmental disorders using probabilistic genotype and phenotype matching among 4,125 families. Nature Genetics, 2015, 47, 1363-1369.	9.4	133
85	Large-scale discovery of novel genetic causes of developmental disorders. Nature, 2015, 519, 223-228.	13.7	998
86	Short Sternum: Feature of Trisomy Chromosome 7 and a New Association?. Pediatric and Developmental Pathology, 2014, 17, 70-72.	0.5	1
87	Pneumothorax from subpleural blebsâ€”A new association of sotos syndrome?. American Journal of Medical Genetics, Part A, 2014, 164, 1222-1226.	0.7	3
88	Congenital myotonic dystrophy. Clinical Dysmorphology, 2014, 23, 127-129.	0.1	1
89	Clinical delineation and natural history of the PIK3CAâ€related overgrowth spectrum. American Journal of Medical Genetics, Part A, 2014, 164, 1713-1733.	0.7	249
90	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.	0.3	5

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91	Cerebral cavernous malformation: Clinical report of two families with variable phenotype associated with KRIT1 mutation. European Journal of Paediatric Neurology, 2013, 17, 661-665.	0.7	4
92	Genotypeâ€“phenotype study in type V osteogenesis imperfecta. Clinical Dysmorphology, 2013, 22, 93-101.	0.1	34
93	Pattern of Clinical Genetics Referral Following Perinatal Postmortems. Pediatric and Developmental Pathology, 2012, 15, 478-486.	0.5	1
94	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.	0.7	15
95	Type 1 collagenopathy presenting with a Russellâ€“Silver phenotype. American Journal of Medical Genetics, Part A, 2011, 155, 1414-1418.	0.7	11
96	Case series: 2q33.1 microdeletion syndrome–further delineation of the phenotype. Journal of Medical Genetics, 2011, 48, 290-298.	1.5	59
97	Mosaic trisomy 11 in a fetus with bilateral renal agenesis. Clinical Dysmorphology, 2011, 20, 47-49.	0.1	11
98	Zimmermannâ€“Laband syndrome in a child previously described with brachydactyly, extrahepatic biliary atresia, patent ductus arteriosus and seizures. Clinical Dysmorphology, 2010, 19, 48-50.	0.1	17
99	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.	0.7	16
100	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.	0.7	24
101	A novel homozygous keratin 10 mutation in siblings with autosomal recessive epidermolytic ichthyosis. Expert Review of Dermatology, 2010, 5, 519-523.	0.3	0
102	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.	0.7	15
103	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.	0.7	13
104	Is this a syndrome? Patterns in genetic conditions. , 0, , 105-119.		0
105	Phenotype-genotype correlation and role of ancillary investigations in atypical and rare forms of osteogenesis imperfecta. Bone Abstracts, 0, , .	0.0	0
106	Skeletal and bone material phenotype in recessive osteogenesis imperfecta due to a novel homozygous point mutation in TMEM38B. Bone Abstracts, 0, , .	0.0	0
107	Cole-Carpenter syndrome. Bone Abstracts, 0, , .	0.0	0
108	In-depth phenotyping including analyses of skin connective tissue in osteogenesis imperfecta. Bone Abstracts, 0, , .	0.0	0

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109	Bone histomorphometry in patients withTMEM38Bmutations suggests a novel patho-mechanism leading to increased bone fragility. Endocrine Abstracts, 0, , .	0.0	0
110	Osteogenesis imperfecta type VI presenting as suspected physical abuse -- a report of two cases. Bone Abstracts, 0, , .	0.0	0
111	NBAS variants causing a novel form of inherited bone fragility. Bone Abstracts, 0, , .	0.0	0
112	P4HB recurrent missense mutation causing Cole-Carpenter syndrome: exploring the underlying mechanism. Bone Abstracts, 0, , .	0.0	0
113	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
114	Radiographic evidence of zoledronic acid given during pregnancy - a case report. Bone Abstracts, 0, , .	0.0	0
115	Identifying the role of NBAS in bone fragility using zebrafish and exploring therapeutic targets to reverse NBAS activity. Bone Abstracts, 0, , .	0.0	0
116	An Emerging Link between Brain and Bone: Patient Report with a Pathogenic SCN9A Variant. SSRN Electronic Journal, 0, , .	0.4	0