Millan S Patel

List of Publications by Year
in descending order

60	6,178	201674	138484
papers	citations	27 h-index	58 g-index

63	63	63	9974
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Linkage analysis identifies an isolated strabismus locus at 14q12 overlapping with FOXG1 syndrome region. Journal of Medical Genetics, 2022, 59, 46-55.	3.2	2
2	Prevalence of ocular anomalies is increased in women with polycystic ovary syndromeâ€"exploration of association with PAX6 genotype. Ophthalmic Genetics, 2022, 43, 340-343.	1.2	2
3	Comprehensive human amniotic fluid metagenomics supports the sterile womb hypothesis. Scientific Reports, 2022, 12, 6875.	3.3	11
4	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. Human Mutation, 2021, 42, 862-876.	2.5	16
5	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	2.4	5
6	Somatic mosaicism detected by genome-wide sequencing in 500 parentâ€"child trios with suspected genetic disease: clinical and genetic counseling implications. Journal of Physical Education and Sports Management, 2021, 7, a006125.	1.2	8
7	Pathologic Skull Fracture in a Near-Term Neonate with Arthrochalasia Type Ehlers-Danlos Syndrome: A Case Report. Fetal and Pediatric Pathology, 2020, , 1-6.	0.7	2
8	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
9	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unitâ€"successes and challenges. European Journal of Pediatrics, 2019, 178, 1207-1218.	2.7	59
10	Congenital lactic acidosis, cerebral cysts and pulmonary hypertension in an infant with FOXRED1 related complex 1 deficiency. Molecular Genetics and Metabolism Reports, 2019, 19, 100472.	1.1	1
11	Congenital lactic acidosis, cerebral cysts and pulmonary hypertension in an infant with FOXRED1 related complex I deficiency. Molecular Genetics and Metabolism Reports, 2019, 18, 32-38.	1.1	8
12	Expanding the FANCO/RAD51C associated phenotype: Cleft lip and palate and lobar holoprosencephaly, two rare findings in Fanconi anemia. European Journal of Medical Genetics, 2018, 61, 257-261.	1.3	11
13	Competing Factors Link to Bone Health in Polycystic Ovary Syndrome: Chronic Low-Grade Inflammation Takes a Toll. Scientific Reports, 2017, 7, 3432.	3.3	34
14	HMMR acts in the PLK1-dependent spindle positioning pathway and supports neural development. ELife, 2017, 6, .	6.0	41
15	Paternal uniparental disomy 11p15.5 in the pancreatic nodule of an infant with Costello syndrome: Shared mechanism for hyperinsulinemic hypoglycemia in neonates with Costello and Beckwithâ€"Wiedemann syndrome and somatic loss of heterozygosity in Costello syndrome driving clonal expansion. American Journal of Medical Genetics, Part A, 2016, 170, 559-564.	1.2	11
16	Diagnosis of Van den Endeâ€"Gupta syndrome: Approach to the Mardenâ€"Walkerâ€like spectrum of disorders. American Journal of Medical Genetics, Part A, 2016, 170, 2310-2321.	1.2	9
17	GeneYenta: A PhenotypeÂBased Rare Disease Case Matching Tool Based on Online Dating Algorithms for the Acceleration of Exome Interpretation. Human Mutation, 2015, 36, 432-438.	2.5	16
18	Alternating hypoglycemia and hyperglycemia in a toddler with a homozygous p.R1419H ABCC8 mutation: an unusual clinical picture. Journal of Pediatric Endocrinology and Metabolism, 2015, 28, 345-51.	0.9	10

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19	Fatal Congenital Hypertrophic Cardiomyopathy and a Pancreatic Nodule Morphologically Identical to Focal Lesion of Congenital Hyperinsulinism in an Infant with Costello Syndrome: Case Report and Review of the Literature. Pediatric and Developmental Pathology, 2015, 18, 237-244.	1.0	14
20	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	6.2	73
21	A genome-wide copy number association study of osteoporotic fractures points to the 6p25.1 locus. Journal of Medical Genetics, 2014, 51, 122-131.	3.2	36
22	A cryptic familial rearrangement of 11p15.5, involving both imprinting centers, in a family with a history of short stature. American Journal of Medical Genetics, Part A, 2014, 164, 1587-1594.	1.2	10
23	Strabismus genetics across a spectrum of eye misalignment disorders. Clinical Genetics, 2014, 86, 103-111.	2.0	35
24	Prenatal and postnatal findings in serpentine fibula polycystic kidney syndrome and a review of the NOTCH2 spectrum disorders. American Journal of Medical Genetics, Part A, 2014, 164, 2490-2495.	1.2	6
25	Anterolateral diaphragmatic hernia with body wall defect understood in relation to the abaxial domain. American Journal of Medical Genetics, Part A, 2014, 164, 1860-1862.	1.2	1
26	Diffuse angiopathy in Adamsâ€Oliver syndrome associated with truncating DOCK6 mutations. American Journal of Medical Genetics, Part A, 2014, 164, 2656-2662.	1.2	32
27	Combined immunodeficiency associated with homozygous MALT1 mutations. Journal of Allergy and Clinical Immunology, 2014, 133, 1458-1462.e7.	2.9	103
28	Mutations in NOTCH1 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2014, 95, 275-284.	6.2	150
29	Evidence of ancillary trigeminal innervation of levator palpebrae in the general population. Journal of Clinical Neuroscience, 2014, 21, 301-304.	1.5	16
30	Calcium and Vitamin D Intake and Mortality: Results from the Canadian Multicentre Osteoporosis Study (CaMos). Journal of Clinical Endocrinology and Metabolism, 2013, 98, 3010-3018.	3.6	49
31	A variant of unknown significance in the GLA gene causing diagnostic uncertainty in a young female with isolated hypertrophic cardiomyopathy. Gene, 2012, 497, 320-322.	2.2	6
32	Genome-wide meta-analysis identifies 56 bone mineral density loci and reveals 14 loci associated with risk of fracture. Nature Genetics, 2012, 44, 491-501.	21.4	1,100
33	Assessment of gene-by-sex interaction effect on bone mineral density. Journal of Bone and Mineral Research, 2012, 27, 2051-2064.	2.8	47
34	Pontocerebellar Hypoplasia: Review of Classification and Genetics, and Exclusion of Several Genes Known to Be Important for Cerebellar Development. Journal of Child Neurology, 2011, 26, 288-294.	1.4	20
35	Outcome of prenatally diagnosed isolated clubfoot. Ultrasound in Obstetrics and Gynecology, 2010, 35, 708-714.	1.7	43
36	Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. BMC Genomics, 2009, 10, 526.	2.8	30

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37	Sudden death in spondylo-meta-epiphyseal dysplasia, short limb-abnormal calcification type. Clinical Dysmorphology, 2009, 18, 25-29.	0.3	12
38	Childhood-onset hemiatrophy caused by unilateral morphea. Clinical Dysmorphology, 2009, 18, 213-214.	0.3	6
39	Submicroscopic deletions of 11q24-25 in individuals without Jacobsen syndrome: re-examination of the critical region by high-resolution array-CGH. Molecular Cytogenetics, 2008, 1, 23.	0.9	25
40	Schinzelâ€“Giedion syndrome: Report of splenopancreatic fusion and proposed diagnostic criteria. American Journal of Medical Genetics, Part A, 2008, 146A, 1299-1306.	1.2	45
41	22q11.2 Distal Deletion: A Recurrent Genomic Disorder Distinct from DiGeorge Syndrome and Velocardiofacial Syndrome. American Journal of Human Genetics, 2008, 82, 214-221.	6.2	182
42	Preaxial polydactyly in neurofibromatosis 1. Clinical Dysmorphology, 2007, 16, 193-194.	0.3	6
43	The New Field of Neuroskeletal Biology. Calcified Tissue International, 2007, 80, 337-347.	3.1	83
44	Severe, fetalâ€onset form of olivopontocerebellar hypoplasia in three sibs: PCH type 5?. American Journal of Medical Genetics, Part A, 2006, 140A, 594-603.	1.2	61
45	The circadian modulation of leptin-controlled bone formation. Progress in Brain Research, 2006, 153, 177-188.	1.4	22
46	Neural control of hematopoietic stem cell mobilization via osteoblasts. BoneKEy Osteovision, 2006, 3, 39-41.	0.6	0
47	WNT7b mediates macrophage-induced programmed cell death in patterning of the vasculature. Nature, 2005, 437, 417-421.	27.8	383
48	The Molecular Clock Mediates Leptin-Regulated Bone Formation. Cell, 2005, 122, 803-815.	28.9	522
49	Canonical Wnt Signaling in Differentiated Osteoblasts Controls Osteoclast Differentiation. Developmental Cell, 2005, 8, 751-764.	7.0	1,402
50	Abnormal pericyte recruitment as a cause for pulmonary hypertension in Adams-Oliver syndrome. American Journal of Medical Genetics Part A, 2004, 129A, 294-299.	2.4	63
51	A New Insight into the Formation of Osteolytic Lesions in Multiple Myeloma. New England Journal of Medicine, 2003, 349, 2479-2480.	27.0	30
52	Cbfa1-independent decrease in osteoblast proliferation, osteopenia, and persistent embryonic eye vascularization in mice deficient in Lrp5, a Wnt coreceptor. Journal of Cell Biology, 2002, 157, 303-314.	5.2	1,032
53	Regulation of Bone Formation and Vision byLRP5. New England Journal of Medicine, 2002, 346, 1572-1574.	27.0	89
54	Genetic determinants of bone mass acquisition and risk for osteoporosis. Drug Development Research, 2000, 49, 216-226.	2.9	4

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55	Alleles of the Estrogen Receptor Î±-Gene and an Estrogen Receptor Cotranscriptional Activator Gene, Amplified in Breast Cancer-1 (AIB1), Are Associated with Quantitative Calcaneal Ultrasound. Journal of Bone and Mineral Research, 2000, 15, 2231-2239.	2.8	33
56	Associations of the Collagen Type IÎ±1 Sp1 Polymorphism with Five-Year Rates of Bone Loss in Older Adults. Calcified Tissue International, 2000, 66, 268-271.	3.1	58
57	Genetic determinants of osteoporosis. , 2000, , 131-146.		1
58	Three novel SALL1 mutations extend the mutational spectrum in Townes-Brocks syndrome. Journal of Medical Genetics, 2000, 37, 303-307.	3.2	26
59	Early-infantile galactosialidosis: Prenatal presentation and postnatal follow-up. American Journal of Medical Genetics Part A, 1999, 85, 38-47.	2.4	44
60	Identification of a sequence motif upstream of the Drosophila Dopa decarboxylase gene that enhances heterologous gene expression. Genome, 1994, 37, 526-534.	2.0	3