Roberto Mendoza-Londono

List of Publications by Year
in descending order

87 papers	5,966 citations	109321 35 h-index	76900 74 g-index
88 all docs	88 docs citations	88 times ranked	10160 citing authors

#	Article	IF	Citations
1	The utility of <scp>DNA</scp> methylation signatures in directing genome sequencing workflow: Kabuki syndrome and <scp>CDK13</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 1368-1375.	1.2	5
2	Diagnostic outcomes for molecular genetic testing in children with suspected Ehlersâ€“Danlos syndrome. American Journal of Medical Genetics, Part A, 2022, , .	1.2	1
3	A Chromosomal Duplication Encompassing Interleukin-33 Causes a Novel Hyper IgE Phenotype Characterized by Eosinophilic Esophagitis and Generalized Autoimmunity. Gastroenterology, 2022, 163, 510-513.e3.	1.3	8
4	Progressive decline of T and B cell numbers and function in a patient with CDC42 deficiency. Immunologic Research, 2021, 69, 53-58.	2.9	5
5	<scp>The pointâ€ofâ€care</scp> use of a facial phenotyping tool in the genetics clinic: Enhancing diagnosis and education with machine learning. American Journal of Medical Genetics, Part A, 2021, 185, 1151-1158.	1.2	18
6	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. Human Mutation, 2021, 42, 862-876.	2.5	16
7	Diagnostic utility of next-generation sequence genetic panel testing in children presenting with a clinically significant fracture history. Archives of Osteoporosis, 2021, 16, 88.	2.4	3
8	The phenotypic spectrum of AMER1 â€related osteopathia striata with cranial sclerosis: The first Canadian cohort. American Journal of Medical Genetics, Part A, 2021, 185, 3793-3803.	1.2	2
9	A rare unbalanced translocation (trisomy 5q33.3â€qter, monosomy 13q34â€qter) results in growth hormone deficiency and brain anomalies. Molecular Genetics & Genomic Medicine, 2021, 9, e1821.	1.2	1
10	Clinical and molecular features of 66 patients with musculocontractural Ehlersâ^'Danlos syndrome caused by pathogenic variants in CHST14 (mcEDS-CHST14). Journal of Medical Genetics, 2021, , jmedgenet-2020-107623.	3.2	18
11	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
12	Blood pressure in adults with short stature skeletal dysplasias. American Journal of Medical Genetics, Part A, 2020, 182, 150-161.	1.2	14
13	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109.	5.9	47
14	COG5 variants lead to complex early onset retinal degeneration, upregulation of PERK and DNA damage. Scientific Reports, 2020, 10, 21269.	3.3	1
15	Clinical utility of genomic sequencing: a measurement toolkit. Npj Genomic Medicine, 2020, 5, 56.	3.8	37
16	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
17	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	1.5	25
18	25 Application of the 2017 Hypermobile Ehlers Danlos Syndrome Diagnostic Criteria in a Paediatric Population. Paediatrics and Child Health, 2019, 24, e10-e11.	0.6	0

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19	16q22.1 microdeletion and anticipatory guidance. American Journal of Medical Genetics, Part A, 2019, 179, 1287-1292.	1.2	1
20	Novel c.G630A TCIRG1 mutation causes aberrant splicing resulting in an unusually mild form of autosomal recessive osteopetrosis. Journal of Cellular Biochemistry, 2019, 120, 17180-17193.	2.6	12
21	Disruption of the PTHLH regulatory landscape results in features consistent with hyperparathyroid disease. American Journal of Medical Genetics, Part A, 2019, 179, 663-667.	1.2	2
22	ATP6AP2 variant impairs CNS development and neuronal survival to cause fulminant neurodegeneration. Journal of Clinical Investigation, 2019, 129, 2145-2162.	8.2	37
23	Severe Neonatal Cholestasis as an Early Presentation of McCune- Albright Syndrome. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2019, 11, 100-103.	0.9	6
24	Periodic reanalysis of whole-genome sequencing data enhances the diagnostic advantage over standard clinical genetic testing. European Journal of Human Genetics, 2018, 26, 740-744.	2.8	88
25	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	6.2	138
26	Severe rhizomelic shortening in a child with a complex duplication/deletion rearrangement of chromosome X. American Journal of Medical Genetics, Part A, 2018, 176, 450-454.	1.2	3
27	Improved diagnostic yield compared with targeted gene sequencing panels suggests a role for whole-genome sequencing as a first-tier genetic test. Genetics in Medicine, 2018, 20, 435-443.	2.4	404
28	Stable transmission of an unbalanced chromosome 21 derived from chromoanasynthesis in a patient with a SYNGAP1 likely pathogenic variant. Molecular Cytogenetics, 2018, 11, 50.	0.9	11
29	Biallelic mutations in DNAJC21 cause Shwachman-Diamond syndrome. Blood, 2017, 129, 1557-1562.	1.4	104
30	Phenotypic and genotypic spectrum of congenital disorders of glycosylation type I and type II. Molecular Genetics and Metabolism, 2017, 120, 235-242.	1.1	44
31	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	6.2	59
32	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289.	2.0	92
33	The genetic landscape of familial congenital hydrocephalus. Annals of Neurology, 2017, 81, 890-897.	5.3	108
34	The Ehlersâ€“Danlos syndromes, rare types. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 70-115.	1.6	168
35	The 2017 international classification of the Ehlersâ€“Danlos syndromes. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 8-26.	1.6	1,163
36	Compound heterozygous mutations in the <scp>IFT140</scp> gene cause Opitz trigonocephaly C syndrome in a patient with typical features of a ciliopathy. Clinical Genetics, 2017, 91, 640-646.	2.0	15

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37	CHARGE and Kabuki Syndromes: Gene-Specific DNA Methylation Signatures Identify Epigenetic Mechanisms Linking These Clinically Overlapping Conditions. American Journal of Human Genetics, 2017, 100, 773-788.	6.2	166
38	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
39	Buschke-Ollendorff syndrome: a novel case series and systematic review. British Journal of Dermatology, 2016, 174, 723-729.	1.5	37
40	Spell Checking Nature: Versatility of CRISPR/Cas9 for Developing Treatments for Inherited Disorders. American Journal of Human Genetics, 2016, 98, 90-101.	6.2	86
41	Borderline Lepromatous Leprosy. Journal of Cutaneous Medicine and Surgery, 2016, 20, 176-177.	1.2	0
42	Utility of wholeâ€exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clinical Genetics, 2016, 89, 275-284.	2.0	323
43	15q11.2 Duplication Encompassing Only theUBE3AGene Is Associated with Developmental Delay and Neuropsychiatric Phenotypes. Human Mutation, 2015, 36, 689-693.	2.5	67
44	MG-109â€...Revisiting a clinical diagnosis 15 years later with the aid of whole exome sequencing: Osteopetrosis versus harderophorphyria. Journal of Medical Genetics, 2015, 52, A4.1-A4.	3.2	0
45	Recessive Osteogenesis Imperfecta Caused by Missense Mutations in SPARC. American Journal of Human Genetics, 2015, 96, 979-985.	6.2	107
46	Mutations Preventing Regulated Exon Skipping in MET Cause Osteofibrous Dysplasia. American Journal of Human Genetics, 2015, 97, 837-847.	6.2	22
47	NSD1 mutations generate a genome-wide DNA methylation signature. Nature Communications, 2015, 6, 10207.	12.8	170
48	Subnuclear re-localization of SOX10 and p54NRB correlates with a unique neurological phenotype associated with SOX10 missense mutations. Human Molecular Genetics, 2015, 24, 4933-4947.	2.9	11
49	Prenatal growth restriction, retinal dystrophy, diabetes insipidus and white matter disease: expanding the spectrum of PRPS1-related disorders. European Journal of Human Genetics, 2015, 23, 310-316.	2.8	30
50	Prevalence of inherited neurotransmitter disorders in patients with movement disorders and epilepsy: a retrospective cohort study. Orphanet Journal of Rare Diseases, 2015, 10, 12.	2.7	18
51	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2015, 52, 431-437.	3.2	187
52	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	6.2	14
53	Clinical characteristics in patients with interstitial deletions of chromosome region 12q21â€“q22 and identification of a critical region associated with keratosis pilaris. American Journal of Medical Genetics, Part A, 2014, 164, 796-800.	1.2	8
54	Mutations in PLK4, encoding a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinopathy. Nature Genetics, 2014, 46, 1283-1292.	21.4	156

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55	Reciprocal deletion and duplication at 2q23.1 indicates a role for MBD5 in autism spectrum disorder. European Journal of Human Genetics, 2014, 22, 57-63.	2.8	42
56	Mutations in B3GALT6, which Encodes a Glycosaminoglycan Linker Region Enzyme, Cause a Spectrum of Skeletal and Connective Tissue Disorders. American Journal of Human Genetics, 2013, 92, 927-934.	6.2	112
57	Angelman Syndrome Due to a Termination Codon Mutation of the UBE3A Gene. Journal of Child Neurology, 2013, 28, 392-395.	1.4	5
58	Severe intellectual disability and autistic features associated with microduplication 2q23.1. European Journal of Human Genetics, 2012, 20, 398-403.	2.8	31
59	Osteopathia striata with cranial sclerosis and developmental delay in a male with a mosaic deletion in chromosome region Xq11.2. American Journal of Medical Genetics, Part A, 2012, 158A, 2946-2952.	1.2	12
60	Extracellular matrix and platelet function in patients with musculocontractural Ehlersâ€“Danlos syndrome caused by mutations in the CHST14 gene. American Journal of Medical Genetics, Part A, 2012, 158A, 1344-1354.	1.2	32
61	Definition of a critical genetic interval related to kidney abnormalities in the Potockiâ€“Lupski syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1579-1588.	1.2	9
62	Identification of three novel plasminogen (PLG) gene mutations in a series of 23 patients with low PLG activity. Thrombosis and Haemostasis, 2011, 105, 454-460.	3.4	42
63	Deletions flanked by breakpoints 3 and 4 on 15q13 may contribute to abnormal phenotypes. European Journal of Human Genetics, 2011, 19, 547-554.	2.8	18
64	Corrigendum to â€œHyperphosphatasia With Seizures, Neurologic Deficit, and Characteristic Facial Features: Five New Patients With Mabry Syndromeâ€•Am J Med Genet 152A: 1661â€“1669. American Journal of Medical Genetics, Part A, 2011, 155, 1215-1215.	1.2	2
65	Spondyloâ€epiphyseal dysplasia, Maroteaux type (pseudoâ€Morquio syndrome type 2), and parastremmatic dysplasia are caused by TRPV4 mutations. American Journal of Medical Genetics, Part A, 2010, 152A, 1443-1449.	1.2	56
66	Hyperphosphatasia with seizures, neurologic deficit, and characteristic facial features: Five new patients with Mabry syndrome. American Journal of Medical Genetics, Part A, 2010, 152A, 1661-1669.	1.2	42
67	Phenotypic features of carbohydrate sulfotransferase 3 (CHST3) deficiency in 24 patients: Congenital dislocations and vertebral changes as principal diagnostic features. American Journal of Medical Genetics, Part A, 2010, 152A, 2543-2549.	1.2	67
68	De novo ACTA2 mutation causes a novel syndrome of multisystemic smooth muscle dysfunction. American Journal of Medical Genetics, Part A, 2010, 152A, 2437-2443.	1.2	217
69	E-selectin ligandâ€“1 regulates growth plate homeostasis in mice by inhibiting the intracellular processing and secretion of mature TGF-Î². Journal of Clinical Investigation, 2010, 120, 2474-2485.	8.2	24
70	Imaging ofSHOX-Associated Anomalies. Seminars in Musculoskeletal Radiology, 2009, 13, 236-254.	0.7	10
71	Aplasia of cochlear nerves and olfactory bulbs in association with SOX10 mutation. American Journal of Medical Genetics, Part A, 2009, 149A, 431-436.	1.2	32
72	A recurrent EYA1 mutation causing alternative RNA splicing in branchioâ€otoâ€renal syndrome: Implications for molecular diagnostics and disease mechanism. American Journal of Medical Genetics, Part A, 2009, 149A, 322-327.	1.2	15

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73	De novo threeâ€way chromosome translocation 46,XY,t(4;6;21)(p16;p21.1;q21) in a male with cleidocranial dysplasia. American Journal of Medical Genetics, Part A, 2008, 146A, 453-458.	1.2	13
74	Generalized metabolic bone disease in Neurofibromatosis type I. Molecular Genetics and Metabolism, 2008, 94, 105-111.	1.1	105
75	Molecular and clinical characterization of de novo and familial cases with microduplication 3q29: guidelines for copy number variation case reporting. Cytogenetic and Genome Research, 2008, 123, 65-78.	1.1	43
76	Characterization of Potocki-Lupski Syndrome (dup(17)(p11.2p11.2)) and Delineation of a Dosage-Sensitive Critical Interval That Can Convey an Autism Phenotype. American Journal of Human Genetics, 2007, 80, 633-649.	6.2	340
77	Smithâ€“Magenis syndrome and moyamoya disease in a patient with del(17)(p11.2p13.1). American Journal of Medical Genetics, Part A, 2007, 143A, 999-1008.	1.2	19
78	Molecular diagnosis of 22q11.2 deletion and duplication by multiplex ligation dependent probe amplification. American Journal of Medical Genetics, Part A, 2007, 143A, 2924-2930.	1.2	43
79	X-LINKED RETINOSCHISIS IN THREE FEMALES FROM THE SAME FAMILY: A PHENOTYPEâ€“GENOTYPE CORRELATION. Retina, 2005, 25, 69-74.	1.7	38
80	Acute lymphoblastic leukemia in a patient with Greig cephalopolysyndactyly and interstitial deletion of chromosome 7 del(7)(p11.2 p14) involving theGLI3 andZNFN1A1 genes. Genes Chromosomes and Cancer, 2005, 42, 82-86.	2.8	13
81	Characterization of a New Syndrome That Associates Craniosynostosis, Delayed Fontanel Closure, Parietal Foramina, Imperforate Anus, and Skin Eruption: CDAGS. American Journal of Human Genetics, 2005, 77, 161-168.	6.2	18
82	Mutations and promoter SNPs in RUNX2, a transcriptional regulator of bone formation. Molecular Genetics and Metabolism, 2005, 86, 257-268.	1.1	50
83	von Voss-Cherstvoy syndrome with transient thrombocytopenia and normal psychomotor development. American Journal of Medical Genetics Part A, 2004, 126A, 299-302.	2.4	6
84	Attenuated phenotype in a child with trisomy for 1q due to unbalanced X;1 translocation [46,X,der(X),t(X;1)(q28;q32.1)]. American Journal of Medical Genetics Part A, 2004, 128A, 72-77.	2.4	10
85	Foreign-Born Physician-Scientists. Science, 2004, 305, 609-609.	12.6	0
86	Omphalocele in trisomy 3q: further delineation of phenotype. Clinical Genetics, 2003, 64, 404-413.	2.0	29
87	A Colombian family with X-linked juvenile retinoschisis with three affected females: Finding of a frameshift mutation. Ophthalmic Genetics, 1999, 20, 37-43.	1.2	38