Roberto Mendoza-Londono

List of Publications by Year
in descending order

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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 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
2	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
3	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
4	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
5	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
6	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
7	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
8	NSD1 mutations generate a genome-wide DNA methylation signature. Nature Communications, 2015, 6, 10207.	12.8	170
9	The Ehlersâ€“Danlos syndromes, rare types. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2017, 175, 70-115.	1.6	168
10	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
11	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
12	Functional Dysregulation of CDC42 Causes Diverse Developmental Phenotypes. American Journal of Human Genetics, 2018, 102, 309-320.	6.2	138
13	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
14	The genetic landscape of familial congenital hydrocephalus. Annals of Neurology, 2017, 81, 890-897.	5.3	108
15	Recessive Osteogenesis Imperfecta Caused by Missense Mutations in SPARC. American Journal of Human Genetics, 2015, 96, 979-985.	6.2	107
16	Generalized metabolic bone disease in Neurofibromatosis type I. Molecular Genetics and Metabolism, 2008, 94, 105-111.	1.1	105
17	Biallelic mutations in DNAJC21 cause Shwachman-Diamond syndrome. Blood, 2017, 129, 1557-1562.	1.4	104
18	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289.	2.0	92

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19	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
20	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
21	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
22	15q11.2 Duplication Encompassing Only theUBE3AGene Is Associated with Developmental Delay and Neuropsychiatric Phenotypes. Human Mutation, 2015, 36, 689-693.	2.5	67
23	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	6.2	59
24	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
25	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
26	Mutations and promoter SNPs in RUNX2, a transcriptional regulator of bone formation. Molecular Genetics and Metabolism, 2005, 86, 257-268.	1.1	50
27	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109.	5.9	47
28	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
29	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
30	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
31	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
32	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
33	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
34	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
35	X-LINKED RETINOSCHISIS IN THREE FEMALES FROM THE SAME FAMILY: A PHENOTYPEâ€“GENOTYPE CORRELATION. Retina, 2005, 25, 69-74.	1.7	38
36	Buschke-Ollendorff syndrome: a novel case series and systematic review. British Journal of Dermatology, 2016, 174, 723-729.	1.5	37

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37	Clinical utility of genomic sequencing: a measurement toolkit. Npj Genomic Medicine, 2020, 5, 56.	3.8	37
38	ATP6AP2 variant impairs CNS development and neuronal survival to cause fulminant neurodegeneration. Journal of Clinical Investigation, 2019, 129, 2145-2162.	8.2	37
39	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
40	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
41	Severe intellectual disability and autistic features associated with microduplication 2q23.1. European Journal of Human Genetics, 2012, 20, 398-403.	2.8	31
42	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
43	Omphalocele in trisomy 3q: further delineation of phenotype. Clinical Genetics, 2003, 64, 404-413.	2.0	29
44	New insights into DNA methylation signatures: SMARCA2 variants in Nicolaides-Baraitser syndrome. BMC Medical Genomics, 2019, 12, 105.	1.5	25
45	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
46	Mutations Preventing Regulated Exon Skipping in MET Cause Osteofibrous Dysplasia. American Journal of Human Genetics, 2015, 97, 837-847.	6.2	22
47	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
48	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
49	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
50	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
51	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
52	<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
53	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
54	New cases that expand the genotypic and phenotypic spectrum of Congenital NAD Deficiency Disorder. Human Mutation, 2021, 42, 862-876.	2.5	16

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55	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
56	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
57	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	6.2	14
58	Blood pressure in adults with short stature skeletal dysplasias. American Journal of Medical Genetics, Part A, 2020, 182, 150-161.	1.2	14
59	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
60	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
61	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
62	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
63	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
64	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
65	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
66	Imaging ofSHOX-Associated Anomalies. Seminars in Musculoskeletal Radiology, 2009, 13, 236-254.	0.7	10
67	Definition of a critical genetic interval related to kidney abnormalities in the Potockâ€“Lupski syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1579-1588.	1.2	9
68	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
69	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
70	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
71	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
72	Angelman Syndrome Due to a Termination Codon Mutation of the UBE3A Gene. Journal of Child Neurology, 2013, 28, 392-395.	1.4	5

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73	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
74	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
75	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
76	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
77	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
78	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
79	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
80	16q22.1 microdeletion and anticipatory guidance. American Journal of Medical Genetics, Part A, 2019, 179, 1287-1292.	1.2	1
81	COG5 variants lead to complex early onset retinal degeneration, upregulation of PERK and DNA damage. Scientific Reports, 2020, 10, 21269.	3.3	1
82	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
83	Diagnostic outcomes for molecular genetic testing in children with suspected Ehlersâ€“Danlos syndrome. American Journal of Medical Genetics, Part A, 2022, , .	1.2	1
84	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
85	Borderline Lepromatous Leprosy. Journal of Cutaneous Medicine and Surgery, 2016, 20, 176-177.	1.2	0
86	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
87	Foreign-Born Physician-Scientists. Science, 2004, 305, 609-609.	12.6	0