

Richard A Lewis

List of Publications by Year in descending order

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Version: 2024-02-01

54
papers

5,454
citations

186265

28
h-index

182427

51
g-index

55
all docs

55
docs citations

55
times ranked

7058
citing authors

#	ARTICLE	IF	CITATIONS
1	<i><i>PRUNE1</i> c.<sc>933G</sc>&gt;A synonymous variant induces exon 7 skipping, disrupts the <sc>DHHA2</sc> domain, and leads to an atypical <sc>NMIHBA</sc> syndrome presentation: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 1868-1874.</i>	1.2	2
2	A novel, de novo intronic variant in <sc><i>POGZ</i></sc> causes <sc>Whiteâ€“Sutton</sc> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2198-2203.	1.2	4
3	The microRNA processor<i>DROSHA</i> is a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	2.9	6
4	Consensus recommendations for the use of retinoids in ichthyosis and other disorders of cornification in children and adolescents. Pediatric Dermatology, 2021, 38, 164-180.	0.9	34
5	A novel de novo intronic variant in ITPR1 causes Gillespie syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2315-2324.	1.2	2
6	Nr2f1 heterozygous knockout mice recapitulate neurological phenotypes of Bosch-Boonstra-Schaaf optic atrophy syndrome and show impaired hippocampal synaptic plasticity. Human Molecular Genetics, 2020, 29, 705-715.	2.9	12
7	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nature Genetics, 2020, 52, 1145-1150.	21.4	22
8	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	6.2	128
9	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
10	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
11	Epidemiology of anophthalmia and microphthalmia: Prevalence and patterns in Texas, 1999â€“2009. American Journal of Medical Genetics, Part A, 2018, 176, 1810-1818.	1.2	26
12	Syndromic congenital myelofibrosis associated with a loss-of-function variant in RBSN. Blood, 2018, 132, 658-662.	1.4	9
13	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
14	Genetic causes of optic nerve hypoplasia. Journal of Medical Genetics, 2017, 54, 441-449.	3.2	30
15	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a <i>TUBB3</i> mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984.	1.2	18
16	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
17	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
18	The phenotypic variability of HK1-associated retinal dystrophy. Scientific Reports, 2017, 7, 7051.	3.3	21

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19	Whole-exome sequencing in the molecular diagnosis of individuals with congenital anomalies of the kidney and urinary tract and identification of a new causative gene. <i>Genetics in Medicine</i> , 2017, 19, 412-420.	2.4	73
20	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. <i>PLoS Genetics</i> , 2017, 13, e1006905.	3.5	80
21	<i>ADIPOR1</i> Is Mutated in Syndromic Retinitis Pigmentosa. <i>Human Mutation</i> , 2016, 37, 246-249.	2.5	41
22	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. <i>American Journal of Human Genetics</i> , 2016, 99, 318-336.	6.2	112
23	Recurrent De Novo and Biallelic Variation of <i>ATAD3A</i> , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. <i>American Journal of Human Genetics</i> , 2016, 99, 831-845.	6.2	146
24	Improved Diagnosis of Inherited Retinal Dystrophies by High-Fidelity PCR of <i>ORF15</i> followed by Next-Generation Sequencing. <i>Journal of Molecular Diagnostics</i> , 2016, 18, 817-824.	2.8	21
25	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotype-phenotype correlations. <i>Genetics in Medicine</i> , 2016, 18, 1143-1150.	2.4	64
26	Identification of a Novel Gene on 10q22.1 Causing Autosomal Dominant Retinitis Pigmentosa (adRP). <i>Advances in Experimental Medicine and Biology</i> , 2016, 854, 193-200.	1.6	8
27	<i>GNAI3</i> : Another Candidate Gene to Screen in Persons with Ocular Albinism. <i>PLoS ONE</i> , 2016, 11, e0162273.	2.5	3
28	Retinal Diseases Caused by Mutations in Genes Not Specifically Associated with the Clinical Diagnosis. <i>PLoS ONE</i> , 2016, 11, e0165405.	2.5	9
29	Novel mutations in two Saudi patients with congenital retinal dystrophy. <i>Middle East African Journal of Ophthalmology</i> , 2016, 23, 139.	0.3	6
30	Dependable and Efficient Clinical Utility of Target Capture-Based Deep Sequencing in Molecular Diagnosis of Retinitis Pigmentosa. , 2014, 55, 6213.		67
31	A Dominant Mutation in Hexokinase 1 (<i>HK1</i>) Causes Retinitis Pigmentosa. , 2014, 55, 7147.		43
32	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. <i>Kidney International</i> , 2014, 85, 880-887.	5.2	67
33	Early onset and severe clinical course associated with the m.5540G>A mutation in <i>MT-TW</i> . <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 61-65.	1.1	3
34	Risk of Cataract among Subjects with Acquired Immune Deficiency Syndrome Free of Ocular Opportunistic Infections. <i>Ophthalmology</i> , 2014, 121, 2317-2324.	5.2	15
35	<i>NR2F1</i> Mutations Cause Optic Atrophy with Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 303-309.	6.2	125
36	Recurrent CNVs and SNVs at the <i>NPHP1</i> Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. <i>American Journal of Human Genetics</i> , 2014, 94, 745-754.	6.2	80

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37	Clinical manifestations in a cohort of 41 Rothmund-Thomson syndrome patients. <i>American Journal of Medical Genetics Part A</i> , 2001, 102, 11-17.	2.4	290
38	Prevalence of mutations causing retinitis pigmentosa and other inherited retinopathies. <i>Human Mutation</i> , 2001, 17, 42-51.	2.5	292
39	Neoplasms in neurofibromatosis 1 are related to gender but not to family history of cancer. <i>Genetic Epidemiology</i> , 2001, 20, 75-86.	1.3	24
40	A recurrent deletion in the ubiquitously expressed NEMO (IKK-gamma) gene accounts for the vast majority of incontinentia pigmenti mutations. <i>Human Molecular Genetics</i> , 2001, 10, 2171-2179.	2.9	165
41	Human homologue of the murine bare patches/striated gene is not mutated in incontinentia pigmenti type 2. , 2000, 91, 241-244.		3
42	Filamin (FLN1), plexin (SEX), major palmitoylated protein p55 (MPP1), and von-Hippel Lindau binding protein (VBP1) are not involved in incontinentia pigmenti type 2. <i>American Journal of Medical Genetics Part A</i> , 2000, 94, 79-84.	2.4	5
43	Mutations in MKKS cause obesity, retinal dystrophy and renal malformations associated with Bardet-Biedl syndrome. <i>Nature Genetics</i> , 2000, 26, 67-70.	21.4	311
44	Dominant retinal dystrophies and Stargardt disease. <i>Ophthalmic Genetics</i> , 1999, 20, 69-70.	1.2	1
45	Novel mutations in XLR51 causing retinoschisis, including first evidence of putative leader sequence change. <i>Human Mutation</i> , 1999, 14, 423-427.	2.5	40
46	A novel locus for Leber congenital amaurosis on chromosome 14q24. <i>Human Genetics</i> , 1998, 103, 328-333.	3.8	69
47	OA1 Mutations and Deletions in X-Linked Ocular Albinism. <i>American Journal of Human Genetics</i> , 1998, 62, 800-809.	6.2	60
48	A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Stargardt macular dystrophy. <i>Nature Genetics</i> , 1997, 15, 236-246.	21.4	1,277
49	Multi-disciplinary clinical study of Smith-Magenis syndrome (deletion 17p11.2). <i>American Journal of Medical Genetics Part A</i> , 1996, 62, 247-254.	2.4	285
50	Cloning of the gene for ocular albinism type 1 from the distal short arm of the X chromosome. <i>Nature Genetics</i> , 1995, 10, 13-19.	21.4	190
51	Analysis of the OA1 gene reveals mutations in only one-third of patients with X-linked ocular albinism. <i>Human Molecular Genetics</i> , 1995, 4, 2319-2325.	2.9	80
52	Bardet-Biedl syndrome is linked to DNA markers on chromosome 11 q and is genetically heterogeneous. <i>Nature Genetics</i> , 1994, 7, 108-112.	21.4	179
53	The Lowe's oculocerebrorenal syndrome gene encodes a protein highly homologous to inositol polyphosphate-5-phosphatase. <i>Nature</i> , 1992, 358, 239-242.	27.8	467
54	Multiple endocrine syndrome type IIb in early childhood. <i>Cancer</i> , 1991, 68, 1832-1834.	4.1	48