Richard A Lewis

List of Publications by Year
in descending order

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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	A photoreceptor cell-specific ATP-binding transporter gene (ABCR) is mutated in recessive Starqardt macular dystrophy. Nature Genetics, 1997, 15, 236-246.	21.4	1,277
2	The Lowe's oculocerebrorenal syndrome gene encodes a protein highly homologous to inositol polyphosphate-5-phosphatase. Nature, 1992, 358, 239-242.	27.8	467
3	Mutations in MKKS cause obesity, retinal dystrophy and renal malformations associated with Bardet-Biedl syndrome. Nature Genetics, 2000, 26, 67-70.	21.4	311
4	Prevalence of mutations causing retinitis pigmentosa and other inherited retinopathies. Human Mutation, 2001, 17, 42-51.	2.5	292
5	Clinical manifestations in a cohort of 41 Rothmund-Thomson syndrome patients. American Journal of Medical Genetics Part A, 2001, 102, 11-17.	2.4	290
6	Multi-disciplinary clinical study of Smith-Magenis syndrome (deletion 17p11.2). American Journal of Medical Genetics Part A, 1996, 62, 247-254.	2.4	285
7	Cloning of the gene for ocular albinism type 1 from the distal short arm of the X chromosome. Nature Genetics, 1995, 10, 13-19.	21.4	190
8	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
9	Bardetâ€“Biedl syndrome is linked to DNA markers on chromosome 11 q and is genetically heterogeneous. Nature Genetics, 1994, 7, 108-112.	21.4	179
10	A recurrent deletion in the ubiquitously expressed NEMO (IKK-gamma) gene accounts for the vast majority of incontinentia pigmenti mutations. Human Molecular Genetics, 2001, 10, 2171-2179.	2.9	165
11	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
12	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
13	NR2F1 Mutations Cause Optic Atrophy with Intellectual Disability. American Journal of Human Genetics, 2014, 94, 303-309.	6.2	125
14	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	6.2	112
15	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
16	Analysis of the OA1 gene reveals mutations in only one-third of patients with X-linked ocular albinism. Human Molecular Genetics, 1995, 4, 2319-2325.	2.9	80
17	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	6.2	80
18	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80

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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. Genetics in Medicine, 2017, 19, 412-420.	2.4	73
20	A novel locus for Leber congenital amaurosis on chromosome 14q24. Human Genetics, 1998, 103, 328-333.	3.8	69
21	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
22	Dependable and Efficient Clinical Utility of Target Capture-Based Deep Sequencing in Molecular Diagnosis of Retinitis Pigmentosa. , 2014, 55, 6213.		67
23	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
24	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150.	2.4	64
25	OA1 Mutations and Deletions in X-Linked Ocular Albinism. American Journal of Human Genetics, 1998, 62, 800-809.	6.2	60
26	Multiple endocrine syndrome type IIb in early childhood. Cancer, 1991, 68, 1832-1834.	4.1	48
27	A Dominant Mutation in Hexokinase 1 (HK1) Causes Retinitis Pigmentosa. , 2014, 55, 7147.		43
28	ADIPOR1Is Mutated in Syndromic Retinitis Pigmentosa. Human Mutation, 2016, 37, 246-249.	2.5	41
29	Novel mutations in XLRS1 causing retinoschisis, including first evidence of putative leader sequence change. Human Mutation, 1999, 14, 423-427.	2.5	40
30	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
31	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
32	Genetic causes of optic nerve hypoplasia. Journal of Medical Genetics, 2017, 54, 441-449.	3.2	30
33	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
34	Neoplasms in neurofibromatosis 1 are related to gender but not to family history of cancer. Genetic Epidemiology, 2001, 20, 75-86.	1.3	24
35	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
36	Improved Diagnosis of Inherited Retinal Dystrophies by High-Fidelity PCR of ORF15 followed by Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 817-824.	2.8	21

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37	The phenotypic variability of HK1-associated retinal dystrophy. Scientific Reports, 2017, 7, 7051.	3.3	21
38	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a TUBB3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984.	1.2	18
39	Risk of Cataract among Subjects with Acquired Immune Deficiency Syndrome FreeÂof Ocular Opportunistic Infections. Ophthalmology, 2014, 121, 2317-2324.	5.2	15
40	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
41	Syndromic congenital myelofibrosis associated with a loss-of-function variant in RBSN. Blood, 2018, 132, 658-662.	1.4	9
42	Retinal Diseases Caused by Mutations in Genes Not Specifically Associated with the Clinical Diagnosis. PLoS ONE, 2016, 11, e0165405.	2.5	9
43	Identification of a Novel Gene on 10q22.1 Causing Autosomal Dominant Retinitis Pigmentosa (adRP). Advances in Experimental Medicine and Biology, 2016, 854, 193-200.	1.6	8
44	Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma. Neurology: Genetics, 2018, 4, e248.	1.9	7
45	Novel mutations in two Saudi patients with congenital retinal dystrophy. Middle East African Journal of Ophthalmology, 2016, 23, 139.	0.3	6
46	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	2.9	6
47	Filamin (FLN1),plexin (SEX), major palmitoylated proteinp55 (MPP1), and von-Hippel Lindau binding protein (VBP1) are not involved in incontinentia pigmenti type 2. American Journal of Medical Genetics Part A, 2000, 94, 79-84.	2.4	5
48	A novel, de novo intronic variant in <scp>POGZ</scp> causes <scp>Whiteâ€“Sutton</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2198-2203.	1.2	4
49	Human homologue of the murinebare patches/striated gene is not mutated in incontinentia pigmenti type 2. , 2000, 91, 241-244.		3
50	Early onset and severe clinical course associated with the m.5540G>A mutation in MT - TW. Molecular Genetics and Metabolism Reports, 2014, 1, 61-65.	1.1	3
51	GNAI3: Another Candidate Gene to Screen in Persons with Ocular Albinism. PLoS ONE, 2016, 11, e0162273.	2.5	3
52	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
53	PRUNE1 c.<scp>933G</scp>>A synonymous variant induces exon 7 skipping, disrupts the <scp>DHHA2</scp> domain, and leads to an atypical <scp>NMIHBA</scp> syndrome presentation: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 1868-1874.	1.2	2
54	Dominant retinal dystrophies and Stargardt disease. Ophthalmic Genetics, 1999, 20, 69-70.	1.2	1