Lisa A Schimmenti

List of Publications by Year
in descending order

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Version: 2024-02-01

83	4,182	201385	118652
papers	citations	27 h-index	62 g-index

85	85	85	5988
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Mutation of the PAX2 gene in a family with optic nerve colobomas, renal anomalies and vesicoureteral reflux. Nature Genetics, 1995, 9, 358-364.	9.4	623
2	Mutations in Cypher/ZASPin patients with dilated cardiomyopathy and left ventricular non-compaction. Journal of the American College of Cardiology, 2003, 42, 2014-2027.	1.2	479
3	A Primer for Morpholino Use in Zebrafish. Zebrafish, 2009, 6, 69-77.	0.5	388
4	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	1.1	312
5	Renal-coloboma syndrome: a multi-system developmental disorder caused byPAX2mutations. Clinical Genetics, 1999, 56, 1-9.	1.0	146
6	Novel mutation in sonic hedgehog in non-syndromic colobomatous microphthalmia. American Journal of Medical Genetics Part A, 2003, 116A, 215-221.	2.4	135
7	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	9.4	131
8	Platelet endothelial cell adhesion molecule, PECAM-1, modulates cell migration. Journal of Cellular Physiology, 1992, 153, 417-428.	2.0	113
9	Update of PAX2 mutations in renal coloboma syndrome and establishment of a locus-specific database. Human Mutation, 2012, 33, 457-466.	1.1	109
10	Recurrent 10q22-q23 Deletions: A Genomic Disorder on 10q Associated with Cognitive and Behavioral Abnormalities. American Journal of Human Genetics, 2007, 80, 938-947.	2.6	101
11	Axenfeld-Rieger syndrome: new perspectives: Figure 1. British Journal of Ophthalmology, 2012, 96, 318-322.	2.1	89
12	Parentsâ€™ Perceptions of Autism Spectrum Disorder Etiology and Recurrence Risk and Effects of their Perceptions on Family Planning: Recommendations for Genetic Counselors. Journal of Genetic Counseling, 2009, 18, 507-519.	0.9	88
13	Expanded newborn screening identifies maternal primary carnitine deficiency. Molecular Genetics and Metabolism, 2007, 90, 441-445.	0.5	86
14	Renal coloboma syndrome. European Journal of Human Genetics, 2011, 19, 1207-1212.	1.4	81
15	Autosomal dominant optic nerve colobomas, vesicoureteral reflux, and renal anomalies. American Journal of Medical Genetics Part A, 1995, 59, 204-208.	2.4	68
16	Genome-Wide Reverse Genetics Framework to Identify Novel Functions of the Vertebrate Secretome. PLoS ONE, 2006, 1, e104.	1.1	67
17	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	1.1	67
18	TLR9 Polymorphisms Are Associated with Altered IFN-Î³ Levels in Children with Cerebral Malaria. American Journal of Tropical Medicine and Hygiene, 2010, 82, 548-555.	0.6	51

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19	Ellisâ€“van Creveld Syndrome and Congenital Heart Defects: Presentation of an Additional 32 Cases. Pediatric Cardiology, 2011, 32, 977-982.	0.6	50
20	Detection of Cytomegalovirus DNA in Dried Blood Spots of Minnesota Infants Who Do Not Pass Newborn Hearing Screening. Pediatric Infectious Disease Journal, 2009, 28, 1095-1098.	1.1	49
21	Homonucleotide expansion and contraction mutations ofPAX2 and inclusion of Chiari 1 malformation as part of Renal-Coloboma syndrome. , 1999, 14, 369-376.		45
22	Two-Tier Approach to the Newborn Screening of Methylenetetrahydrofolate Reductase Deficiency and Other Remethylation Disorders with Tandem Mass Spectrometry. Journal of Pediatrics, 2010, 157, 271-275.	0.9	43
23	Development and Notch Signaling Requirements of the Zebrafish Choroid Plexus. PLoS ONE, 2008, 3, e3114.	1.1	42
24	Cri du Chat Syndrome and Congenital Heart Disease: A Review of Previously Reported Cases and Presentation of an Additional 21 Cases From the Pediatric Cardiac Care Consortium. Pediatrics, 2006, 117, e924-e927.	1.0	41
25	Attitudes of the broader hearing, deaf, and hard-of-hearing community toward genetic testing for deafness. Genetics in Medicine, 2003, 5, 106-112.	1.1	40
26	A novel microdeletion/microduplication syndrome of 19p13.13. Genetics in Medicine, 2010, 12, 503-511.	1.1	37
27	Next generation sequencing in research and diagnostics of ocular birth defects. Molecular Genetics and Metabolism, 2010, 100, 184-192.	0.5	29
28	Analysis of archived newborn dried blood spots (DBS) identifies congenital cytomegalovirus as a major cause of unexplained pediatric sensorineural hearing loss. American Journal of Otolaryngology - Head and Neck Medicine and Surgery, 2017, 38, 565-570.	0.6	29
29	Next Generation Sequencing of Sporadic Vestibular Schwannoma: Necessity of Biallelic NF2 Inactivation and Implications of Accessory Non-NF2 Variants. Otology and Neurotology, 2018, 39, e860-e871.	0.7	29
30	Optic nerve dysplasia and renal insufficiency in a family with a novel PAX2 mutation, Arg115X: further ophthalmologic delineation of the renal-coloboma syndrome. Ophthalmic Genetics, 2003, 24, 191-202.	0.5	28
31	A prospective, longitudinal study of the impact of GJB2/GJB6 genetic testing on the beliefs and attitudes of parents of deaf and hardâ€ofâ€hearing infants. American Journal of Medical Genetics, Part A, 2009, 149A, 1169-1182.	0.7	28
32	Evaluation of newborn screening bloodspot-based genetic testing as second tier screen for bedside newborn hearing screening. Genetics in Medicine, 2011, 13, 1006-1010.	1.1	28
33	Additive reductions in zebrafish PRPS1 activity result in a spectrum of deficiencies modeling several human PRPS1-associated diseases. Scientific Reports, 2016, 6, 29946.	1.6	28
34	Infant hearing loss and connexin testing in a diverse population. Genetics in Medicine, 2008, 10, 517-524.	1.1	27
35	Infant with multiple congenital anomalies and deletion (9)(q34.3). American Journal of Medical Genetics Part A, 1994, 51, 140-142.	2.4	26
36	Genetic testing as part of the Early Hearing Detection and Intervention (EHDI) process. Genetics in Medicine, 2004, 6, 521-525.	1.1	25

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37	Array comparative genomic hybridization analysis in patients with anophthalmia, microphthalmia, and coloboma. Genetics in Medicine, 2011, 13, 437-442.	1.1	25
38	Identification of novel mutations and sequence variants in the SOX2 and CHX10 genes in patients with anophthalmia/microphthalmia. Molecular Vision, 2008, 14, 583-92.	1.1	25
39	The lineage-specific geneponzr1is essential for zebrafish pronephric and pharyngeal arch development. Development (Cambridge), 2012, 139, 793-804.	1.2	24
40	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	1.1	24
41	Ethnic Differences in Parental Perceptions of Genetic Testing for Deaf Infants. Journal of Genetic Counseling, 2008, 17, 129-138.	0.9	22
42	Outcomes of four patients with homocysteine remethylation disorders detected by newborn screening. Genetics in Medicine, 2016, 18, 162-167.	1.1	21
43	Profound intellectual disability caused by homozygous TRAPPC9 pathogenic variant in a man from Malta. Molecular Genetics & Genomic Medicine, 2020, 8, e1211.	0.6	20
44	New Tmc1 Deafness Mutations Impact Mechanotransduction in Auditory Hair Cells. Journal of Neuroscience, 2021, 41, 4378-4391.	1.7	18
45	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	1.1	18
46	Bosma arhinia microphthalmia syndrome: Clinical report and review of the literature. American Journal of Medical Genetics, Part A, 2016, 170, 1302-1307.	0.7	17
47	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	2.6	16
48	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India. Molecular Genetics & Genomic Medicine, 2020, 8, e1081.	0.6	13
49	Monosomy 9p24â†'pter and trisomy 5q31â†'qter: Case report and review of two cases. American Journal of Medical Genetics Part A, 1995, 57, 52-56.	2.4	12
50	Severe Methylenetetrahydrofolate Reductase (MTHFR) Deficiency: A Case Report of Nonclassical Homocystinuria. Journal of Child Neurology, 2008, 23, 823-828.	0.7	12
51	A Catalog of Genetic Syndromes in Childhood Cancer. Pediatric Blood and Cancer, 2015, 62, 2071-2075.	0.8	12
52	Building the vertebrate codex using the gene breaking protein trap library. ELife, 2020, 9, .	2.8	11
53	Sharing GJB2/GJB6 Genetic Test Information with Family Members. Journal of Genetic Counseling, 2007, 16, 313-324.	0.9	9
54	Nonhatching Decapsulated Artemia Cysts As a Replacement to Artemia Nauplii in Juvenile and Adult Zebrafish Culture. Zebrafish, 2015, 12, 457-461.	0.5	9

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55	ADULT-ONSET VITELLIFORM MACULAR DYSTROPHY SECONDARY TO A NOVEL IMPG2 GENE VARIANT. Retinal Cases and Brief Reports, 2021, 15, 356-358.	0.3	8
56	Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases. European Journal of Human Genetics, 2019, 27, 1379-1388.	1.4	8
57	Identification of aggressive Gardner syndrome phenotype associated with a de novo APC variant, c.4666dup. Journal of Physical Education and Sports Management, 2019, 5, a003640.	0.5	8
58	Identification of Two Single Nucleotide Polymorphisms in Exon 8 of PAX2. Molecular Genetics and Metabolism, 1999, 68, 507-510.	0.5	7
59	Clinical utility gene card for: renal coloboma (Papillorenal) syndrome. European Journal of Human Genetics, 2011, 19, 1017-1017.	1.4	7
60	Typical renalâ€coloboma syndrome phenotype in a patient with a submicroscopic deletion of the PAX2 gene. American Journal of Medical Genetics, Part A, 2012, 158A, 1437-1441.	0.7	7
61	Posterior staphyloma in oculocutaneous albinism: anotherÂpossible cause of reducedÂvisual acuity. Journal of AAPOS, 2015, 19, 562-564.	0.2	7
62	Congenital ichthyosis in Praderâ€“Willi syndrome associated with maternal chromosome 15 uniparental disomy: Case report and review of autosomal recessive conditions unmasked by UPD. American Journal of Medical Genetics, Part A, 2020, 182, 2442-2449.	0.7	6
63	L-type voltage-gated calcium channel agonists mitigate hearing loss and modify ribbon synapse morphology in the zebrafish model of Usher syndrome type 1. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	6
64	De novo <scp>PBX1</scp> variant in a patient with glaucoma, kidney anomalies, and developmental delay: An expansion of the <scp>CAKUTHED</scp> phenotype. American Journal of Medical Genetics, Part A, 2022, 188, 919-925.	0.7	6
65	PUS7 deficiency in human patients causes profound neurodevelopmental phenotype by dysregulating protein translation. Molecular Genetics and Metabolism, 2022, 135, 221-229.	0.5	6
66	Genetic and developmental basis of renal coloboma (papillorenal) syndrome. Expert Review of Ophthalmology, 2009, 4, 135-144.	0.3	5
67	Necrotizing Enterocolitis in Two Siblings and an Unrelated Infant with Overlapping Chromosome 6q25 Deletions. Molecular Syndromology, 2018, 9, 141-148.	0.3	5
68	Development and Validation of a Next-Generation Sequencing Panel for Syndromic and Nonsyndromic Hearing Loss. journal of applied laboratory medicine, The, 2020, 5, 467-479.	0.6	4
69	Identification of two novel polymorphisms (g.903C>T and g.1544C>T) in thePAX2 gene. Human Mutation, 2001, 17, 155-155.	1.1	3
70	Mosaic partial deletion of PTPN12 in a child with interrupted aortic arch type A. American Journal of Medical Genetics, Part A, 2015, 167, 2674-2683.	0.7	3
71	The GoAudio Quantitative Mobile Audiology Test Enhances Access to Clinical Hearing Assessments. American Journal of Audiology, 2020, 29, 887-897.	0.5	3
72	Duplication of the Down syndrome critical region does not predict facial phenotype in a baby with a ring chromosome 21. Clinical Dysmorphology, 2005, 14, 183-187.	0.1	3

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73	Keep Swimming Toward Precision Medicine Discoveries. Zebrafish, 2016, 13, 545-547.	0.5	2
74	Zebrafish and the Cancer Moonshot. Zebrafish, 2017, 14, 390-392.	0.5	2
75	Detection of PAX2 Deletions and Duplications Using Multiplex Ligation-Dependent Probe Amplification. Genetic Testing and Molecular Biomarkers, 2013, 17, 786-788.	0.3	1
76	Genetic Testing for Deaf and Hard of Hearing Individuals: Genetic Counseling. Current Genetic Medicine Reports, 2016, 4, 27-34.	1.9	1
77	The Best Retinitis Pigmentosa Masquerade. Ophthalmology, 2019, 126, 1694.	2.5	1
78	Growth hormone deficiency in a child with <scp>branchioâ€otoâ€renal</scp> spectrum disorder: Clinical evidence of <scp>EYA1</scp> in pituitary development and a recommendation for pituitary function surveillance. American Journal of Medical Genetics, Part A, 2021, 185, 261-266.	0.7	1
79	Current Approaches to the Management of Usher Syndrome for the Clinician. Perspectives of the ASHA Special Interest Groups, 2020, 5, 907-916.	0.4	1
80	Emerging Therapies and Approaches to Treat and Prevent Hearing Loss. Perspectives of the ASHA Special Interest Groups, 2020, 5, 1147-1165.	0.4	1
81	Zebrafish: A Functional Refuge at the End of an Odyssey. Zebrafish, 2016, 13, 236-238.	0.5	0
82	Mary Ella Mascia Pierpont: Geneticist, scientist, mentor, friend (1945â€“2020). American Journal of Medical Genetics, Part A, 2021, 185, 319-323.	0.7	0
83	Modeling SLC26A4 Associated Hearing Loss Using Zebrafish as a Model System. FASEB Journal, 2018, 32, 533.79.	0.2	0