Lisa A Schimmenti

List of Publications by Year
in descending order

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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	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
2	PUS7 deficiency in human patients causes profound neurodevelopmental phenotype by dysregulating protein translation. Molecular Genetics and Metabolism, 2022, 135, 221-229.	0.5	6
3	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
4	ADULT-ONSET VITELLIFORM MACULAR DYSTROPHY SECONDARY TO A NOVEL IMPG2 GENE VARIANT. Retinal Cases and Brief Reports, 2021, 15, 356-358.	0.3	8
5	Impact of integrated translational research on clinical exome sequencing. Genetics in Medicine, 2021, 23, 498-507.	1.1	24
6	Mary Ella Mascia Pierpont: Geneticist, scientist, mentor, friend (1945â€“2020). American Journal of Medical Genetics, Part A, 2021, 185, 319-323.	0.7	0
7	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
8	New Tmc1 Deafness Mutations Impact Mechanotransduction in Auditory Hair Cells. Journal of Neuroscience, 2021, 41, 4378-4391.	1.7	18
9	Disease-specific ACMG/AMP guidelines improve sequence variant interpretation for hearing loss. Genetics in Medicine, 2021, 23, 2208-2212.	1.1	18
10	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
11	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
12	Profound intellectual disability caused by homozygous TRAPPC9 pathogenic variant in a man from Malta. Molecular Genetics & Genomic Medicine, 2020, 8, e1211.	0.6	20
13	The GoAudio Quantitative Mobile Audiology Test Enhances Access to Clinical Hearing Assessments. American Journal of Audiology, 2020, 29, 887-897.	0.5	3
14	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
15	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
16	Building the vertebrate codex using the gene breaking protein trap library. ELife, 2020, 9, .	2.8	11
17	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
18	Emerging Therapies and Approaches to Treat and Prevent Hearing Loss. Perspectives of the ASHA Special Interest Groups, 2020, 5, 1147-1165.	0.4	1

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19	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
20	ClinGen expert clinical validity curation of 164 hearing loss geneâ€“disease pairs. Genetics in Medicine, 2019, 21, 2239-2247.	1.1	67
21	The Best Retinitis Pigmentosa Masquerade. Ophthalmology, 2019, 126, 1694.	2.5	1
22	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
23	Expert specification of the ACMG/AMP variant interpretation guidelines for genetic hearing loss. Human Mutation, 2018, 39, 1593-1613.	1.1	312
24	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
25	Necrotizing Enterocolitis in Two Siblings and an Unrelated Infant with Overlapping Chromosome 6q25 Deletions. Molecular Syndromology, 2018, 9, 141-148.	0.3	5
26	Modeling SLC26A4 Associated Hearing Loss Using Zebrafish as a Model System. FASEB Journal, 2018, 32, 533.79.	0.2	0
27	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
28	Zebrafish and the Cancer Moonshot. Zebrafish, 2017, 14, 390-392.	0.5	2
29	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
30	Genetic Testing for Deaf and Hard of Hearing Individuals: Genetic Counseling. Current Genetic Medicine Reports, 2016, 4, 27-34.	1.9	1
31	Zebrafish: A Functional Refuge at the End of an Odyssey. Zebrafish, 2016, 13, 236-238.	0.5	0
32	Keep Swimming Toward Precision Medicine Discoveries. Zebrafish, 2016, 13, 545-547.	0.5	2
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	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
35	Outcomes of four patients with homocysteine remethylation disorders detected by newborn screening. Genetics in Medicine, 2016, 18, 162-167.	1.1	21
36	Posterior staphyloma in oculocutaneous albinism: anotherÂpossible cause of reducedÂvisual acuity. Journal of AAPOS, 2015, 19, 562-564.	0.2	7

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37	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
38	A Catalog of Genetic Syndromes in Childhood Cancer. Pediatric Blood and Cancer, 2015, 62, 2071-2075.	0.8	12
39	Nonhatching Decapsulated Artemia Cysts As a Replacement to Artemia Nauplii in Juvenile and Adult Zebrafish Culture. Zebrafish, 2015, 12, 457-461.	0.5	9
40	Detection of PAX2 Deletions and Duplications Using Multiplex Ligation-Dependent Probe Amplification. Genetic Testing and Molecular Biomarkers, 2013, 17, 786-788.	0.3	1
41	The lineage-specific geneponzr1is essential for zebrafish pronephric and pharyngeal arch development. Development (Cambridge), 2012, 139, 793-804.	1.2	24
42	Axenfeld-Rieger syndrome: new perspectives: Figure 1. British Journal of Ophthalmology, 2012, 96, 318-322.	2.1	89
43	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
44	Update of PAX2 mutations in renal coloboma syndrome and establishment of a locus-specific database. Human Mutation, 2012, 33, 457-466.	1.1	109
45	Clinical utility gene card for: renal coloboma (Papillorenal) syndrome. European Journal of Human Genetics, 2011, 19, 1017-1017.	1.4	7
46	Ellisâ€“van Creveld Syndrome and Congenital Heart Defects: Presentation of an Additional 32 Cases. Pediatric Cardiology, 2011, 32, 977-982.	0.6	50
47	Array comparative genomic hybridization analysis in patients with anophthalmia, microphthalmia, and coloboma. Genetics in Medicine, 2011, 13, 437-442.	1.1	25
48	Renal coloboma syndrome. European Journal of Human Genetics, 2011, 19, 1207-1212.	1.4	81
49	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
50	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
51	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
52	A novel microdeletion/microduplication syndrome of 19p13.13. Genetics in Medicine, 2010, 12, 503-511.	1.1	37
53	Next generation sequencing in research and diagnostics of ocular birth defects. Molecular Genetics and Metabolism, 2010, 100, 184-192.	0.5	29
54	A Primer for Morpholino Use in Zebrafish. Zebrafish, 2009, 6, 69-77.	0.5	388

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55	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
56	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
57	Genetic and developmental basis of renal coloboma (papillorenal) syndrome. Expert Review of Ophthalmology, 2009, 4, 135-144.	0.3	5
58	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
59	Ethnic Differences in Parental Perceptions of Genetic Testing for Deaf Infants. Journal of Genetic Counseling, 2008, 17, 129-138.	0.9	22
60	Infant hearing loss and connexin testing in a diverse population. Genetics in Medicine, 2008, 10, 517-524.	1.1	27
61	Severe Methylenetetrahydrofolate Reductase (MTHFR) Deficiency: A Case Report of Nonclassical Homocystinuria. Journal of Child Neurology, 2008, 23, 823-828.	0.7	12
62	Development and Notch Signaling Requirements of the Zebrafish Choroid Plexus. PLoS ONE, 2008, 3, e3114.	1.1	42
63	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
64	Expanded newborn screening identifies maternal primary carnitine deficiency. Molecular Genetics and Metabolism, 2007, 90, 441-445.	0.5	86
65	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
66	Sharing GJB2/GJB6 Genetic Test Information with Family Members. Journal of Genetic Counseling, 2007, 16, 313-324.	0.9	9
67	Genome-Wide Reverse Genetics Framework to Identify Novel Functions of the Vertebrate Secretome. PLoS ONE, 2006, 1, e104.	1.1	67
68	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
69	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
70	Genetic testing as part of the Early Hearing Detection and Intervention (EHDI) process. Genetics in Medicine, 2004, 6, 521-525.	1.1	25
71	Novel mutation in sonic hedgehog in non-syndromic colobomatous microphthalmia. American Journal of Medical Genetics Part A, 2003, 116A, 215-221.	2.4	135
72	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

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73	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
74	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
75	Identification of two novel polymorphisms (g.903C>T and g.1544C>T) in thePAX2 gene. Human Mutation, 2001, 17, 155-155.	1.1	3
76	Renal-coloboma syndrome: a multi-system developmental disorder caused byPAX2mutations. Clinical Genetics, 1999, 56, 1-9.	1.0	146
77	Homonucleotide expansion and contraction mutations ofPAX2 and inclusion of Chiari 1 malformation as part of Renal-Coloboma syndrome. , 1999, 14, 369-376.		45
78	Identification of Two Single Nucleotide Polymorphisms in Exon 8 of PAX2. Molecular Genetics and Metabolism, 1999, 68, 507-510.	0.5	7
79	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
80	Autosomal dominant optic nerve colobomas, vesicoureteral reflux, and renal anomalies. American Journal of Medical Genetics Part A, 1995, 59, 204-208.	2.4	68
81	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
82	Infant with multiple congenital anomalies and deletion (9)(q34.3). American Journal of Medical Genetics Part A, 1994, 51, 140-142.	2.4	26
83	Platelet endothelial cell adhesion molecule, PECAM-1, modulates cell migration. Journal of Cellular Physiology, 1992, 153, 417-428.	2.0	113