

Anna Lehman

List of Publications by Year in descending order

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85
papers

3,455
citations

159585

30
h-index

155660

55
g-index

87
all docs

87
docs citations

87
times ranked

7581
citing authors

#	ARTICLE	IF	CITATIONS
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
2	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	27.0	254
3	Transcriptional Regulation of BACE1, the β -Amyloid Precursor Protein β -Secretase, by Sp1. Molecular and Cellular Biology, 2004, 24, 865-874.	2.3	207
4	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
5	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
6	Mutations in NOTCH1 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2014, 95, 275-284.	6.2	150
7	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	7.6	115
8	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
9	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. American Journal of Human Genetics, 2017, 101, 65-74.	6.2	99
10	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	6.2	88
11	Mitochondrial Carbonic Anhydrase VA Deficiency Resulting from CA5A Alterations Presents with Hyperammonemia in Early Childhood. American Journal of Human Genetics, 2014, 94, 453-461.	6.2	82
12	<i>OCRL1</i> Mutations in Dent 2 Patients Suggest a Mechanism for Phenotypic Variability. Nephron Physiology, 2009, 112, p27-p36.	1.2	79
13	The cost and diagnostic yield of exome sequencing for children with suspected genetic disorders: a benchmarking study. Genetics in Medicine, 2018, 20, 1-9.	2.4	79
14	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	6.2	73
15	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	6.2	72
16	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
17	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unit—successes and challenges. European Journal of Pediatrics, 2019, 178, 1207-1218.	2.7	59
18	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	6.2	52

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19	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	8.2	50
20	A novel RYR2 loss-of-function mutation (I4855M) is associated with left ventricular non-compaction and atypical catecholaminergic polymorphic ventricular tachycardia. <i>Journal of Electrocardiology</i> , 2017, 50, 227-233.	0.9	47
21	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. <i>American Journal of Human Genetics</i> , 2019, 105, 283-301.	6.2	46
22	Schinzel-Giedion syndrome: Report of splenopancreatic fusion and proposed diagnostic criteria. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1299-1306.	1.2	45
23	Etiologies of uterine malformations. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2141-2172.	1.2	45
24	<i>NBEA</i> : Developmental disease gene with early generalized epilepsy phenotypes. <i>Annals of Neurology</i> , 2018, 84, 788-795.	5.3	44
25	Histone H3.3 beyond cancer: Germline mutations in <i>Histone 3 Family 3A and 3B</i> cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , 2020, 6, .	10.3	43
26	Illness experience, depression, and anxiety in chronic fatigue syndrome. <i>Journal of Psychosomatic Research</i> , 2002, 52, 461-465.	2.6	41
27	DECIDE: a Decision Support Tool to Facilitate Parents' Choices Regarding Genome-Wide Sequencing. <i>Journal of Genetic Counseling</i> , 2016, 25, 1298-1308.	1.6	36
28	Complex translocation disrupting TCF4 and altering TCF4 isoform expression segregates as mild autosomal dominant intellectual disability. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 62.	2.7	35
29	Diffuse angiopathy in Adams-Oliver syndrome associated with truncating <i>DOCK6</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2656-2662.	1.2	32
30	<i>GREB1L</i> variants in familial and sporadic hereditary urogenital adysplasia and Mayer-Rokitansky-Kuster-Hauser syndrome. <i>Clinical Genetics</i> , 2020, 98, 126-137.	2.0	32
31	Co-occurrence of Joubert syndrome and Jeune asphyxiating thoracic dystrophy. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1411-1419.	1.2	30
32	Missense Variants in the Histone Acetyltransferase Complex Component Gene <i>TRRAP</i> Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	6.2	30
33	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. <i>American Journal of Human Genetics</i> , 2020, 106, 143-152.	6.2	30
34	A Clinical Classification Scheme for Tracheobronchomegaly (Mounier-Kuhn Syndrome). <i>Lung</i> , 2015, 193, 815-822.	3.3	27
35	De novo <i>TBR1</i> variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. <i>European Journal of Human Genetics</i> , 2020, 28, 770-782.	2.8	27
36	<i>KDM5A</i> mutations identified in autism spectrum disorder using forward genetics. <i>ELife</i> , 2020, 9, .	6.0	27

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37	Novel findings and expansion of phenotype in a mosaic <scp>RASopathy</scp> caused by somatic <scp><i>KRAS</i></scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 2829-2845.	1.2	23
38	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	2.4	22
39	Identifying candidate genes for 2p15p16.1 microdeletion syndrome using clinical, genomic, and functional analysis. JCI Insight, 2016, 1, e85461.	5.0	22
40	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. European Journal of Medical Genetics, 2009, 52, 436-439.	1.3	21
41	New developmental syndromes: Understanding the family experience. Journal of Genetic Counseling, 2019, 28, 202-212.	1.6	21
42	Causal Attributions, Perceived Control, and Psychological Adjustment: A Study of Chronic Fatigue Syndrome¹. Journal of Applied Social Psychology, 2006, 36, 75-99.	2.0	20
43	FOXP1 haploinsufficiency: Phenotypes beyond behavior and intellectual disability?. American Journal of Medical Genetics, Part A, 2017, 173, 3172-3181.	1.2	18
44	The Genomic Consultation Service: A clinical service designed to improve patient selection for genome-wide sequencing in British Columbia. Molecular Genetics & Genomic Medicine, 2018, 6, 592-600.	1.2	18
45	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
46	Treatable inborn errors of metabolism causing neurological symptoms in adults. Molecular Genetics and Metabolism, 2013, 110, 431-438.	1.1	16
47	Evidence of ancillary trigeminal innervation of levator palpebrae in the general population. Journal of Clinical Neuroscience, 2014, 21, 301-304.	1.5	16
48	Unique variants in CLCN3, encoding an endosomal anion/proton exchanger, underlie a spectrum of neurodevelopmental disorders. American Journal of Human Genetics, 2021, 108, 1450-1465.	6.2	16
49	Emphysema in an adult with galactosialidosis linked to a defect in primary elastic fiber assembly. Molecular Genetics and Metabolism, 2012, 106, 99-103.	1.1	15
50	Prenatal ultrasound and MRI findings of temporal and occipital lobe dysplasia in a twin with achondroplasia. Ultrasound in Obstetrics and Gynecology, 2014, 44, 365-368.	1.7	15
51	BMPER variants associated with a novel, attenuated subtype of diaphanospondylodysostosis. Journal of Human Genetics, 2015, 60, 743-747.	2.3	15
52	19p13.2 microduplication causes a Sotos syndrome-like phenotype and alters gene expression. Clinical Genetics, 2012, 81, 56-63.	2.0	14
53	Additional post-natal diagnoses following antenatal diagnosis of isolated cleft lip +/âˆƒ palate. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2014, 99, F286-F290.	2.8	14
54	Patient Recall, Interpretation, and Perspective of an Inconclusive Long QT Syndrome Genetic Test Result. Journal of Genetic Counseling, 2017, 26, 150-158.	1.6	13

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55	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2021, 23, 374-383.	2.4	13
56	PSEN1 p.Met233Val in a Complex Neurodegenerative Movement and Neuropsychiatric Disorder. <i>Journal of Movement Disorders</i> , 2018, 11, 45-48.	1.3	12
57	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo <i>OPA3</i> mutation. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a001156.	1.2	11
58	Variant Reinterpretation in Survivors of Cardiac Arrest With Preserved Ejection Fraction (the Cardiac) Tj ETQq0 0 0 rgBT /Overlock 10 Tf Laboratories. <i>Circulation Genomic and Precision Medicine</i> , 2021, 14, e003235.	3.6	10
59	Bi-allelic variants in the ER quality-control mannosidase gene <i>EDEM3</i> cause a congenital disorder of glycosylation. <i>American Journal of Human Genetics</i> , 2021, 108, 1342-1349.	6.2	9
60	A Novel Germline Heterozygous <i>BCL11B</i> Variant Causing Severe Atopic Disease and Immune Dysregulation. <i>Frontiers in Immunology</i> , 2021, 12, 788278.	4.8	9
61	How do Physicians Decide to Refer Their Patients for Psychiatric Genetic Counseling? A Qualitative Study of Physicians' Practice. <i>Journal of Genetic Counseling</i> , 2016, 25, 1235-1242.	1.6	8
62	Hypogonadotropic Hypogonadism in Males with Glycogen Storage Disease Type 1. <i>JIMD Reports</i> , 2017, 36, 79-84.	1.5	8
63	Intracranial Calcification after Cord Blood Neonatal Transplantation for Krabbe Disease. <i>Neuropediatrics</i> , 2009, 40, 189-191.	0.6	7
64	Beyond the Electrocardiogram: Mutations in Cardiac Ion Channel Genes Underlie Nonarrhythmic Phenotypes. <i>Clinical Medicine Insights: Cardiology</i> , 2017, 11, 117954681769813.	1.8	7
65	High rate of hypertension in patients with m.3243A>G MELAS mutations and <i>POLG</i> variants. <i>Mitochondrion</i> , 2020, 53, 194-202.	3.4	7
66	Childhood-onset hemiatrophy caused by unilateral morphea. <i>Clinical Dysmorphology</i> , 2009, 18, 213-214.	0.3	6
67	Child Neurology: Krabbe disease. <i>Neurology</i> , 2012, 79, e170-2.	1.1	6
68	Rare disorders have many faces: in silico characterization of rare disorder spectrum. <i>Orphanet Journal of Rare Diseases</i> , 2022, 17, 76.	2.7	6
69	Corneal findings in Parry-Romberg syndrome. <i>Canadian Journal of Ophthalmology</i> , 2014, 49, e2-e5.	0.7	5
70	<i>PIGG</i> variant pathogenicity assessment reveals characteristic features within 19 families. <i>Genetics in Medicine</i> , 2021, 23, 1873-1881.	2.4	5
71	Utilization of telehealth in paediatric genome-wide sequencing: Health services implementation issues in the CAUSES Study. <i>Journal of Telemedicine and Telecare</i> , 2023, 29, 318-327.	2.7	5
72	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. <i>Pediatric Neurology</i> , 2019, 100, 87-91.	2.1	4

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73	Renpenning syndrome in a female. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 498-503.	1.2	4
74	Cost Analysis of Patients Referred for Inherited Heart Rhythm Disorder Evaluation. <i>Canadian Journal of Cardiology</i> , 2017, 33, 814-821.	1.7	3
75	Novel Exonic Deletions in TTC7A in a Newborn with Multiple Intestinal Atresia and Combined Immunodeficiency. <i>Journal of Clinical Immunology</i> , 2019, 39, 616-619.	3.8	3
76	Return of Results Policies for Genomic Research: Current Practices and the Hearts in Rhythm Organization (HiRO) Approach. <i>Canadian Journal of Cardiology</i> , 2022, 38, 526-535.	1.7	3
77	Can leaky splicing and evasion of premature termination codon surveillance contribute to the phenotypic variability in Alkuraya-Kucinskas syndrome?. <i>European Journal of Medical Genetics</i> , 2022, 65, 104427.	1.3	3
78	The effect of rapid exome sequencing on downstream health care utilization for infants with suspected genetic disorders in an intensive care unit. <i>Genetics in Medicine</i> , 2022, 24, 1675-1683.	2.4	3
79	Anterolateral diaphragmatic hernia with body wall defect understood in relation to the abaxial domain. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1860-1862.	1.2	1
80	Genomic and Cytogenetic Characterization of a Balanced Translocation Disrupting <i>NUIP98</i> . <i>Cytogenetic and Genome Research</i> , 2017, 152, 117-121.	1.1	1
81	Secondary biogenic amine deficiencies: genetic etiology, therapeutic interventions, and clinical effects. <i>Neurogenetics</i> , 2021, 22, 251-262.	1.4	1
82	MG-141...A further report of paediatric cancer and cleidocranial dysplasia raises the possibility of a causative association of weak effect. <i>Journal of Medical Genetics</i> , 2015, 52, A12.1-A12.	3.2	0
83	Familial impairment of vocal cord mobility in childhood with clubfoot. <i>Clinical Dysmorphology</i> , 2018, 27, 116-121.	0.3	0
84	Integration of genetic counsellors in genomic testing triage: Outcomes of a genomic consultation service in British Columbia, Canada. <i>European Journal of Medical Genetics</i> , 2021, 64, 104024.	1.3	0
85	Fetal Progeria: Prenatal Sonographic Findings in Petty Syndrome. <i>Journal of Ultrasound in Medicine</i> , 2013, 32, 881-883.	1.7	0