Anna Lehman

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

82 papers	2,288 citations	25 h-index	46 g-index
87 ext. papers	2,973 ext. citations	6.2 avg, IF	4.05 L-index

#	Paper	IF	Citations
82	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
81	Exome Sequencing and the Management of Neurometabolic Disorders. *New England Journal of Medicine*, **2016**, 374, 2246-55	59.2	197
80	Transcriptional regulation of BACE1, the beta-amyloid precursor protein beta-secretase, by Sp1. *Molecular and Cellular Biology*, **2004**, 24, 865-74	4.8	183
79	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. *American Journal of Human Genetics*, **2013**, 93, 915-25	11	155
78	Mutations in NOTCH1 cause Adams-Oliver syndrome. *American Journal of Human Genetics*, **2014**, 95, 275-84	11	119
77	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. *American Journal of Human Genetics*, **2018**, 102, 175-187	11	108
76	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. *Brain*, **2013**, 136, 3618-24	11.2	100
75	Mitochondrial carbonic anhydrase VA deficiency resulting from CA5A alterations presents with hyperammonemia in early childhood. *American Journal of Human Genetics*, **2014**, 94, 453-61	11	66
74	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. *Brain*, **2017**, 140, 2610-2622	11.2	63
73	OCRL1 mutations in Dent 2 patients suggest a mechanism for phenotypic variability. *Nephron Physiology*, **2009**, 112, p27-36		62
72	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. *American Journal of Human Genetics*, **2015**, 97, 475-82	11	61
71	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. *American Journal of Human Genetics*, **2016**, 98, 579-587	11	60
70	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	11	58
69	The cost and diagnostic yield of exome sequencing for children with suspected genetic disorders: a benchmarking study. *Genetics in Medicine*, **2018**, 20, 1013-1021	8.1	55
68	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. *American Journal of Human Genetics*, **2017**, 100, 91-104	11	43
67	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. *American Journal of Human Genetics*, **2017**, 100, 592-604	11	42
66	Illness experience, depression, and anxiety in chronic fatigue syndrome. *Journal of Psychosomatic Research*, **2002**, 52, 461-5	4.1	36

65	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. *American Journal of Human Genetics*, **2017**, 100, 138-150	11	32
64	Etiologies of uterine malformations. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2141-72	2.5	32
63	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unit-successes and challenges. *European Journal of Pediatrics*, **2019**, 178, 1207-1218	4.1	29
62	Schinzel-Giedion syndrome: report of splenopancreatic fusion and proposed diagnostic criteria. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1299-306	2.5	29
61	Diffuse angiopathy in Adams-Oliver syndrome associated with truncating DOCK6 mutations. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 2656-62	2.5	28
60	DECIDE: a Decision Support Tool to Facilitate ParentsQChoices Regarding Genome-Wide Sequencing. *Journal of Genetic Counseling*, **2016**, 25, 1298-1308	2.5	27
59	A novel RYR2 loss-of-function mutation (I4855M) is associated with left ventricular non-compaction and atypical catecholaminergic polymorphic ventricular tachycardia. *Journal of Electrocardiology*, **2017**, 50, 227-233	1.4	26
58	A Clinical Classification Scheme for Tracheobronchomegaly (Mounier-Kuhn Syndrome). *Lung*, **2015**, 193, 815-22	2.9	25
57	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. *American Journal of Human Genetics*, **2019**, 105, 283-301	11	20
56	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. *European Journal of Medical Genetics*, **2009**, 52, 436-9	2.6	20
55	Co-occurrence of Joubert syndrome and Jeune asphyxiating thoracic dystrophy. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1411-9	2.5	19
54	Complex translocation disrupting TCF4 and altering TCF4 isoform expression segregates as mild autosomal dominant intellectual disability. *Orphanet Journal of Rare Diseases*, **2016**, 11, 62	4.2	19
53	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. *Annals of Neurology*, **2018**, 84, 788-795	9.4	18
52	Identifying candidate genes for 2p15p16.1 microdeletion syndrome using clinical, genomic, and functional analysis. *JCI Insight*, **2016**, 1, e85461	9.9	17
51	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. *American Journal of Human Genetics*, **2019**, 104, 530-541	11	17
50	GREB1L variants in familial and sporadic hereditary urogenital adysplasia and Mayer-Rokitansky-Kuster-Hauser syndrome. *Clinical Genetics*, **2020**, 98, 126-137	4	16
49	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. *American Journal of Human Genetics*, **2020**, 106, 143-152	11	16
48	Causal Attributions, Perceived Control, and Psychological Adjustment: A Study of Chronic Fatigue Syndrome1. *Journal of Applied Social Psychology*, **2006**, 36, 75-99	2.1	16

47	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	2.3	16
46	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. *European Journal of Human Genetics*, **2020**, 28, 770-782	5.3	13
45	Emphysema in an adult with galactosialidosis linked to a defect in primary elastic fiber assembly. *Molecular Genetics and Metabolism*, **2012**, 106, 99-103	3.7	13
44	19p13.2 microduplication causes a Sotos syndrome-like phenotype and alters gene expression. *Clinical Genetics*, **2012**, 81, 56-63	4	13
43	Evidence of ancillary trigeminal innervation of levator palpebrae in the general population. *Journal of Clinical Neuroscience*, **2014**, 21, 301-4	2.2	12
42	FOXP1 haploinsufficiency: Phenotypes beyond behavior and intellectual disability?. *American Journal of Medical Genetics, Part A*, **2017**, 173, 3172-3181	2.5	12
41	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. *Science Advances*, **2020**, 6,	14.3	12
40	New developmental syndromes: Understanding the family experience. *Journal of Genetic Counseling*, **2019**, 28, 202-212	2.5	11
39	Treatable inborn errors of metabolism causing neurological symptoms in adults. *Molecular Genetics and Metabolism*, **2013**, 110, 431-8	3.7	11
38	Patient Recall, Interpretation, and Perspective of an Inconclusive Long QT Syndrome Genetic Test Result. *Journal of Genetic Counseling*, **2017**, 26, 150-158	2.5	10
37	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo mutation. *Journal of Physical Education and Sports Management*, **2017**, 3, a001156	2.8	10
36	BMPER variants associated with a novel, attenuated subtype of diaphanospondylodysostosis. *Journal of Human Genetics*, **2015**, 60, 743-7	4.3	10
35	Additional post-natal diagnoses following antenatal diagnosis of isolated cleft lip +/□palate. *Archives of Disease in Childhood: Fetal and Neonatal Edition*, **2014**, 99, F286-90	4.7	10
34	p.Met233Val in a Complex Neurodegenerative Movement and Neuropsychiatric Disorder. *Journal of Movement Disorders*, **2018**, 11, 45-48	2.9	10
33	mutations identified in autism spectrum disorder using forward genetics. *ELife*, **2020**, 9,	8.9	10
32	Prenatal ultrasound and MRI findings of temporal and occipital lobe dysplasia in a twin with achondroplasia. *Ultrasound in Obstetrics and Gynecology*, **2014**, 44, 365-8	5.8	9
31	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63	14.4	9
30	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. *Genetics in Medicine*, **2020**, 22, 1215-1226	8.1	7

29	Intracranial calcification after cord blood neonatal transplantation for krabbe disease. *Neuropediatrics*, **2009**, 40, 189-91	1.6	7
28	How do Physicians Decide to Refer Their Patients for Psychiatric Genetic Counseling? A Qualitative Study of PhysiciansQPractice. *Journal of Genetic Counseling*, **2016**, 25, 1235-1242	2.5	6
27	High rate of hypertension in patients with m.3243A>G MELAS mutations and POLG variants. *Mitochondrion*, **2020**, 53, 194-202	4.9	5
26	Childhood-onset hemiatrophy caused by unilateral morphea. *Clinical Dysmorphology*, **2009**, 18, 213-4	0.9	5
25	Novel findings and expansion of phenotype in a mosaic RASopathy caused by somatic KRAS variants. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2829-2845	2.5	5
24	Hypogonadotropic Hypogonadism in Males with Glycogen Storage Disease Type 1. *JIMD Reports*, **2017**, 36, 79-84	1.9	4
23	Beyond the Electrocardiogram: Mutations in Cardiac Ion Channel Genes Underlie Nonarrhythmic Phenotypes. *Clinical Medicine Insights: Cardiology*, **2017**, 11, 1179546817698134	3.2	4
22	Child Neurology: Krabbe disease: a potentially treatable white matter disorder. *Neurology*, **2012**, 79, e170-2	6.5	4
21	Cost Analysis of Patients Referred for Inherited Heart Rhythm Disorder Evaluation. *Canadian Journal of Cardiology*, **2017**, 33, 814-821	3.8	3
20	Novel Exonic Deletions in TTC7A in a Newborn with Multiple Intestinal Atresia and Combined Immunodeficiency. *Journal of Clinical Immunology*, **2019**, 39, 616-619	5.7	3
19	Corneal findings in Parry-Romberg syndrome. *Canadian Journal of Ophthalmology*, **2014**, 49, e2-5	1.4	3
18	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	11	2
17	Variant Reinterpretation in Survivors of Cardiac Arrest With Preserved Ejection Fraction (the Cardiac Arrest Survivors With Preserved Ejection Fraction Registry) by Clinicians and Clinical Commercial Laboratories. *Circulation Genomic and Precision Medicine*, **2021**, 14, e003235	5.2	2
16	Utilization of telehealth in paediatric genome-wide sequencing: Health services implementation issues in the CAUSES Study. *Journal of Telemedicine and Telecare*, **2021**, 1357633X20982737	6.8	2
15	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. *Pediatric Neurology*, **2019**, 100, 87-91	2.9	1
14	Genomic and Cytogenetic Characterization of a Balanced Translocation Disrupting NUP98. *Cytogenetic and Genome Research*, **2017**, 152, 117-121	1.9	1
13	Anterolateral diaphragmatic hernia with body wall defect understood in relation to the abaxial domain. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1860-2	2.5	1
12	Can leaky splicing and evasion of premature termination codon surveillance contribute to the phenotypic variability in Alkuraya-Kucinskas syndrome?. *European Journal of Medical Genetics*, **2022**, 65, 104427	2.6	1

11	Renpenning syndrome in a female. *American Journal of Medical Genetics, Part A*, **2020**, 182, 498-503	2.5	1
10	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. *Genetics in Medicine*, **2021**, 23, 1873-1881	8.1	1
9	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. *American Journal of Human Genetics*, **2021**, 108, 1342-1349	11	1
8	A Novel Germline Heterozygous Variant Causing Severe Atopic Disease and Immune Dysregulation. *Frontiers in Immunology*, **2021**, 12, 788278	8.4	0
7	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. *Genetics in Medicine*, **2021**, 23, 374-383	8.1	0
6	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	11	0
5	Rare disorders have many faces: in silico characterization of rare disorder spectrum.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 76	4.2	0
4	MG-141 A further report of paediatric cancer and cleidocranial dysplasia raises the possibility of a causative association of weak effect. *Journal of Medical Genetics*, **2015**, 52, A12.1-A12	5.8	
3	Secondary biogenic amine deficiencies: genetic etiology, therapeutic interventions, and clinical effects. *Neurogenetics*, **2021**, 22, 251-262	3	
2	Integration of genetic counsellors in genomic testing triage: Outcomes of a genomic consultation service in British Columbia, Canada. *European Journal of Medical Genetics*, **2021**, 64, 104024	2.6	
1	Familial impairment of vocal cord mobility in childhood with clubfoot. *Clinical Dysmorphology*, **2018**, 27, 116-121	0.9	