

Anna Lehman

List of Publications by Year in Descending Order

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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	Can leaky splicing and evasion of premature termination codon surveillance contribute to the phenotypic variability in Alkuraya-Kucinkas syndrome?. <i>European Journal of Medical Genetics</i> , 2022 , 65, 104427	2.6	1
81	Rare disorders have many faces: in silico characterization of rare disorder spectrum.. <i>Orphanet Journal of Rare Diseases</i> , 2022 , 17, 76	4.2	0
80	A Novel Germline Heterozygous Variant Causing Severe Atopic Disease and Immune Dysregulation. <i>Frontiers in Immunology</i> , 2021 , 12, 788278	8.4	0
79	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021 , 13, 63	14.4	9
78	Novel findings and expansion of phenotype in a mosaic RASopathy caused by somatic KRAS variants. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 2829-2845	2.5	5
77	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. <i>American Journal of Human Genetics</i> , 2021 , 108, 1138-1150	11	2
76	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. <i>Circulation Genomic and Precision Medicine</i> , 2021 , 14, e003235	5.2	2
75	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. <i>Genetics in Medicine</i> , 2021 , 23, 1873-1881	8.1	1
74	Secondary biogenic amine deficiencies: genetic etiology, therapeutic interventions, and clinical effects. <i>Neurogenetics</i> , 2021 , 22, 251-262	3	
73	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2021 , 23, 374-383	8.1	0
72	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	2.6	
71	Bi-allelic variants in the ER quality-control mannosidase gene EDEM3 cause a congenital disorder of glycosylation. <i>American Journal of Human Genetics</i> , 2021 , 108, 1342-1349	11	1
70	Unique variants in CLCN3, encoding an endosomal anion/proton exchanger, underlie a spectrum of neurodevelopmental disorders. <i>American Journal of Human Genetics</i> , 2021 , 108, 1450-1465	11	0
69	Utilization of telehealth in paediatric genome-wide sequencing: Health services implementation issues in the CAUSES Study. <i>Journal of Telemedicine and Telecare</i> , 2021 , 1357633X20982737	6.8	2
68	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. <i>Genetics in Medicine</i> , 2020 , 22, 1215-1226	8.1	7
67	GREB1L variants in familial and sporadic hereditary urogenital adysplasia and Mayer-Rokitansky-Kuster-Hauser syndrome. <i>Clinical Genetics</i> , 2020 , 98, 126-137	4	16
66	High rate of hypertension in patients with m.3243A>G MELAS mutations and POLG variants. <i>Mitochondrion</i> , 2020 , 53, 194-202	4.9	5

65	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	11	16
64	De novo TBR1 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	5.3	13
63	mutations identified in autism spectrum disorder using forward genetics. <i>ELife</i> , 2020 , 9,	8.9	10
62	Renpenning syndrome in a female. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 498-503	2.5	1
61	Histone H3.3 beyond cancer: Germline mutations in cause a previously unidentified neurodegenerative disorder in 46 patients. <i>Science Advances</i> , 2020 , 6,	14.3	12
60	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unit-successes and challenges. <i>European Journal of Pediatrics</i> , 2019 , 178, 1207-1218	4.1	29
59	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. <i>Pediatric Neurology</i> , 2019 , 100, 87-91	2.9	1
58	New developmental syndromes: Understanding the family experience. <i>Journal of Genetic Counseling</i> , 2019 , 28, 202-212	2.5	11
57	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	11	20
56	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	5.7	3
55	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019 , 104, 530-541	11	17
54	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. <i>American Journal of Human Genetics</i> , 2018 , 102, 175-187	11	108
53	The cost and diagnostic yield of exome sequencing for children with suspected genetic disorders: a benchmarking study. <i>Genetics in Medicine</i> , 2018 , 20, 1013-1021	8.1	55
52	p.Met233Val in a Complex Neurodegenerative Movement and Neuropsychiatric Disorder. <i>Journal of Movement Disorders</i> , 2018 , 11, 45-48	2.9	10
51	Familial impairment of vocal cord mobility in childhood with clubfoot. <i>Clinical Dysmorphology</i> , 2018 , 27, 116-121	0.9	
50	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. <i>Annals of Neurology</i> , 2018 , 84, 788-795	9.4	18
49	The Genomic Consultation Service: A clinical service designed to improve patient selection for genome-wide sequencing in British Columbia. <i>Molecular Genetics & Genomic Medicine</i> , 2018 , 6, 592	2.3	16
48	Patient Recall, Interpretation, and Perspective of an Inconclusive Long QT Syndrome Genetic Test Result. <i>Journal of Genetic Counseling</i> , 2017 , 26, 150-158	2.5	10

47	Cost Analysis of Patients Referred for Inherited Heart Rhythm Disorder Evaluation. <i>Canadian Journal of Cardiology</i> , 2017 , 33, 814-821	3.8	3
46	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo mutation. <i>Journal of Physical Education and Sports Management</i> , 2017 , 3, a001156	2.8	10
45	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. <i>American Journal of Human Genetics</i> , 2017 , 100, 592-604	11	42
44	Hypogonadotropic Hypogonadism in Males with Glycogen Storage Disease Type 1. <i>JIMD Reports</i> , 2017 , 36, 79-84	1.9	4
43	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. <i>American Journal of Human Genetics</i> , 2017 , 100, 138-150	11	32
42	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. <i>American Journal of Human Genetics</i> , 2017 , 100, 91-104	11	43
41	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017 , 101, 664-685	11	214
40	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. <i>Brain</i> , 2017 , 140, 2610-2622	11.2	63
39	Genomic and Cytogenetic Characterization of a Balanced Translocation Disrupting NUP98. <i>Cytogenetic and Genome Research</i> , 2017 , 152, 117-121	1.9	1
38	FOXP1 haploinsufficiency: Phenotypes beyond behavior and intellectual disability?. <i>American Journal of Medical Genetics, Part A</i> , 2017 , 173, 3172-3181	2.5	12
37	Beyond the Electrocardiogram: Mutations in Cardiac Ion Channel Genes Underlie Nonarrhythmic Phenotypes. <i>Clinical Medicine Insights: Cardiology</i> , 2017 , 11, 1179546817698134	3.2	4
36	Loss-of-Function and Gain-of-Function Mutations in KCNQ5 Cause Intellectual Disability or Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2017 , 101, 65-74	11	58
35	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	1.4	26
34	DECIDE: a Decision Support Tool to Facilitate Parents' Choices Regarding Genome-Wide Sequencing. <i>Journal of Genetic Counseling</i> , 2016 , 25, 1298-1308	2.5	27
33	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. <i>American Journal of Human Genetics</i> , 2016 , 98, 579-587	11	60
32	Identifying candidate genes for 2p15p16.1 microdeletion syndrome using clinical, genomic, and functional analysis. <i>JCI Insight</i> , 2016 , 1, e85461	9.9	17
31	Etiologies of uterine malformations. <i>American Journal of Medical Genetics, Part A</i> , 2016 , 170, 2141-72	2.5	32
30	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	4.2	19

29	Exome Sequencing and the Management of Neurometabolic Disorders. <i>New England Journal of Medicine</i> , 2016 , 374, 2246-55	59.2	197
28	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	2.5	6
27	BMPER variants associated with a novel, attenuated subtype of diaphanospondylodysostosis. <i>Journal of Human Genetics</i> , 2015 , 60, 743-7	4.3	10
26	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. <i>American Journal of Human Genetics</i> , 2015 , 97, 475-82	11	61
25	A Clinical Classification Scheme for Tracheobronchomegaly (Mounier-Kuhn Syndrome). <i>Lung</i> , 2015 , 193, 815-22	2.9	25
24	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	5.8	
23	Mitochondrial carbonic anhydrase VA deficiency resulting from CA5A alterations presents with hyperammonemia in early childhood. <i>American Journal of Human Genetics</i> , 2014 , 94, 453-61	11	66
22	Additional post-natal diagnoses following antenatal diagnosis of isolated cleft lip +/-palate. <i>Archives of Disease in Childhood: Fetal and Neonatal Edition</i> , 2014 , 99, F286-90	4.7	10
21	Prenatal ultrasound and MRI findings of temporal and occipital lobe dysplasia in a twin with achondroplasia. <i>Ultrasound in Obstetrics and Gynecology</i> , 2014 , 44, 365-8	5.8	9
20	Mutations in NOTCH1 cause Adams-Oliver syndrome. <i>American Journal of Human Genetics</i> , 2014 , 95, 275-84	11	119
19	Corneal findings in Parry-Romberg syndrome. <i>Canadian Journal of Ophthalmology</i> , 2014 , 49, e2-5	1.4	3
18	Evidence of ancillary trigeminal innervation of levator palpebrae in the general population. <i>Journal of Clinical Neuroscience</i> , 2014 , 21, 301-4	2.2	12
17	Anterolateral diaphragmatic hernia with body wall defect understood in relation to the abaxial domain. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 1860-2	2.5	1
16	Diffuse angiopathy in Adams-Oliver syndrome associated with truncating DOCK6 mutations. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 2656-62	2.5	28
15	Defects in the IFT-B component IFT172 cause Jeune and Mainzer-Saldino syndromes in humans. <i>American Journal of Human Genetics</i> , 2013 , 93, 915-25	11	155
14	Treatable inborn errors of metabolism causing neurological symptoms in adults. <i>Molecular Genetics and Metabolism</i> , 2013 , 110, 431-8	3.7	11
13	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. <i>Brain</i> , 2013 , 136, 3618-24	11.2	100
12	Child Neurology: Krabbe disease: a potentially treatable white matter disorder. <i>Neurology</i> , 2012 , 79, e170-2	6.5	4

11	Emphysema in an adult with galactosialidosis linked to a defect in primary elastic fiber assembly. <i>Molecular Genetics and Metabolism</i> , 2012 , 106, 99-103	3.7	13
10	19p13.2 microduplication causes a Sotos syndrome-like phenotype and alters gene expression. <i>Clinical Genetics</i> , 2012 , 81, 56-63	4	13
9	Co-occurrence of Joubert syndrome and Jeune asphyxiating thoracic dystrophy. <i>American Journal of Medical Genetics, Part A</i> , 2010 , 152A, 1411-9	2.5	19
8	OCRL1 mutations in Dent 2 patients suggest a mechanism for phenotypic variability. <i>Nephron Physiology</i> , 2009 , 112, p27-36		62
7	Intracranial calcification after cord blood neonatal transplantation for krabbe disease. <i>Neuropediatrics</i> , 2009 , 40, 189-91	1.6	7
6	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. <i>European Journal of Medical Genetics</i> , 2009 , 52, 436-9	2.6	20
5	Childhood-onset hemiatrophy caused by unilateral morphea. <i>Clinical Dysmorphology</i> , 2009 , 18, 213-4	0.9	5
4	Schinzel-Giedion syndrome: report of splenopancreatic fusion and proposed diagnostic criteria. <i>American Journal of Medical Genetics, Part A</i> , 2008 , 146A, 1299-306	2.5	29
3	Causal Attributions, Perceived Control, and Psychological Adjustment: A Study of Chronic Fatigue Syndrome1. <i>Journal of Applied Social Psychology</i> , 2006 , 36, 75-99	2.1	16
2	Transcriptional regulation of BACE1, the beta-amyloid precursor protein beta-secretase, by Sp1. <i>Molecular and Cellular Biology</i> , 2004 , 24, 865-74	4.8	183
1	Illness experience, depression, and anxiety in chronic fatigue syndrome. <i>Journal of Psychosomatic Research</i> , 2002 , 52, 461-5	4.1	36