Anna Lehman

List of Publications by Year
in descending order

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85 papers	3,455 citations	182225 30 h-index	175968 55 g-index
87 all docs	87 docs citations	87 times ranked	8164 citing authors

#	Article	IF	Citations
1	Utilization of telehealth in paediatric genome-wide sequencing: Health services implementation issues in the CAUSES Study. Journal of Telemedicine and Telecare, 2023, 29, 318-327.	1.4	5
2	Return of Results Policies for Genomic Research: Current Practices and the Hearts in Rhythm Organization (HiRO) Approach. Canadian Journal of Cardiology, 2022, 38, 526-535.	0.8	3
3	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.	0.7	3
4	Rare disorders have many faces: in silico characterization of rare disorder spectrum. Orphanet Journal of Rare Diseases, 2022, 17, 76.	1.2	6
5	The effect of rapid exome sequencing on downstream health care utilization for infants with suspected genetic disorders in an intensive care unit. Genetics in Medicine, 2022, 24, 1675-1683.	1.1	3
6	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	1.1	13
7	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.	0.7	0
8	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	3.6	50
9	Novel findings and expansion of phenotype in a mosaic <scp>RASopathy</scp> caused by somatic <scp>KRAS</scp> variants. American Journal of Medical Genetics, Part A, 2021, 185, 2829-2845.	0.7	23
10	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.	2.6	17
11	Variant Reinterpretation in Survivors of Cardiac Arrest With Preserved Ejection Fraction (the Cardiac) Tj ETQq1 1 0.784314 rgBT /Ove Laboratories. Circulation Genomic and Precision Medicine, 2021, 14, e003235.	1.6	10
12	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	1.1	5
13	Secondary biogenic amine deficiencies: genetic etiology, therapeutic interventions, and clinical effects. Neurogenetics, 2021, 22, 251-262.	0.7	1
14	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.	2.6	9
15	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.	2.6	16
16	A Novel Germline Heterozygous BCL11B Variant Causing Severe Atopic Disease and Immune Dysregulation. Frontiers in Immunology, 2021, 12, 788278.	2.2	9
17	Renpenning syndrome in a female. American Journal of Medical Genetics, Part A, 2020, 182, 498-503.	0.7	4
18	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43

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19	Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7. Genetics in Medicine, 2020, 22, 1215-1226.	1.1	22
20	<scp>GREB1L</scp> variants in familial and sporadic hereditary urogenital adysplasia and <scp>Mayerâ€Rokitanskyâ€Küsterâ€Hauser</scp> syndrome. Clinical Genetics, 2020, 98, 126-137.	1.0	32
21	High rate of hypertension in patients with m.3243A>G MELAS mutations and POLG variants. Mitochondrion, 2020, 53, 194-202.	1.6	7
22	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. American Journal of Human Genetics, 2020, 106, 143-152.	2.6	30
23	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.	1.4	27
24	KDM5A mutations identified in autism spectrum disorder using forward genetics. ELife, 2020, 9, .	2.8	27
25	De Novo Heterozygous POLR2A Variants Cause a Neurodevelopmental Syndrome with Profound Infantile-Onset Hypotonia. American Journal of Human Genetics, 2019, 105, 283-301.	2.6	46
26	Novel Exonic Deletions in TTC7A in a Newborn with Multiple Intestinal Atresia and Combined Immunodeficiency. Journal of Clinical Immunology, 2019, 39, 616-619.	2.0	3
27	RAPIDOMICS: rapid genome-wide sequencing in a neonatal intensive care unitâ€"successes and challenges. European Journal of Pediatrics, 2019, 178, 1207-1218.	1.3	59
28	Strabismus in Children With Intellectual Disability: Part of a Broader Motor Control Phenotype?. Pediatric Neurology, 2019, 100, 87-91.	1.0	4
29	New developmental syndromes: Understanding the family experience. Journal of Genetic Counseling, 2019, 28, 202-212.	0.9	21
30	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.	2.6	30
31	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	2.6	204
32	The cost and diagnostic yield of exome sequencing for children with suspected genetic disorders: a benchmarking study. Genetics in Medicine, 2018, 20, 1-9.	1.1	79
33	Familial impairment of vocal cord mobility in childhood with clubfoot. Clinical Dysmorphology, 2018, 27, 116-121.	0.1	0
34	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	2.8	44
35	The Genomic Consultation Service: A clinical service designed to improve patient selection for genomeâ€wide sequencing in British Columbia. Molecular Genetics & Genomic Medicine, 2018, 6, 592-600.	0.6	18
36	PSEN1 p.Met233Val in a Complex Neurodegenerative Movement and Neuropsychiatric Disorder. Journal of Movement Disorders, 2018, 11, 45-48.	0.7	12

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37	Patient Recall, Interpretation, and Perspective of an Inconclusive Long QT Syndrome Genetic Test Result. Journal of Genetic Counseling, 2017, 26, 150-158.	0.9	13
38	Cost Analysis of Patients Referred for Inherited Heart Rhythm Disorder Evaluation. Canadian Journal of Cardiology, 2017, 33, 814-821.	0.8	3
39	Optic atrophy, cataracts, lipodystrophy/lipoatrophy, and peripheral neuropathy caused by a de novo OPA3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a001156.	0.5	11
40	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	2.6	61
41	Hypogonadotropic Hypogonadism in Males with Glycogen Storage Disease Type 1. JIMD Reports, 2017, 36, 79-84.	0.7	8
42	De Novo Mutations in EBF3 Cause a Neurodevelopmental Syndrome. American Journal of Human Genetics, 2017, 100, 138-150.	2.6	52
43	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	2.6	72
44	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
45	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	3.7	102
46	Genomic and Cytogenetic Characterization of a Balanced Translocation Disrupting <i>NUP98</i>. Cytogenetic and Genome Research, 2017, 152, 117-121.	0.6	1
47	FOXP1 haploinsufficiency: Phenotypes beyond behavior and intellectual disability?. American Journal of Medical Genetics, Part A, 2017, 173, 3172-3181.	0.7	18
48	Beyond the Electrocardiogram: Mutations in Cardiac Ion Channel Genes Underlie Nonarrhythmic Phenotypes. Clinical Medicine Insights: Cardiology, 2017, 11, 117954681769813.	0.6	7
49	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.	2.6	99
50	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.	0.4	47
51	Etiologies of uterine malformations. American Journal of Medical Genetics, Part A, 2016, 170, 2141-2172.	0.7	45
52	Complex translocation disrupting TCF4 and altering TCF4 isoform expression segregates as mild autosomal dominant intellectual disability. Orphanet Journal of Rare Diseases, 2016, 11, 62.	1.2	35
53	Exome Sequencing and the Management of Neurometabolic Disorders. New England Journal of Medicine, 2016, 374, 2246-2255.	13.9	254
54	How do Physicians Decide to Refer Their Patients for Psychiatric Genetic Counseling? A Qualitative Study of Physiciansâ€™ Practice. Journal of Genetic Counseling, 2016, 25, 1235-1242.	0.9	8

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55	DECIDE: a Decision Support Tool to Facilitate Parentsâ€™ Choices Regarding Genomeâ€Wide Sequencing. Journal of Genetic Counseling, 2016, 25, 1298-1308.	0.9	36
56	Mosaic Activating Mutations in FGFR1 Cause Encephalocraniocutaneous Lipomatosis. American Journal of Human Genetics, 2016, 98, 579-587.	2.6	88
57	Identifying candidate genes for 2p15p16.1 microdeletion syndrome using clinical, genomic, and functional analysis. JCI Insight, 2016, 1, e85461.	2.3	22
58	MG-141â€…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.	1.5	0
59	BMPER variants associated with a novel, attenuated subtype of diaphanospondylodysostosis. Journal of Human Genetics, 2015, 60, 743-747.	1.1	15
60	Heterozygous Loss-of-Function Mutations in DLL4 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2015, 97, 475-482.	2.6	73
61	A Clinical Classification Scheme for Tracheobronchomegaly (Mounier-Kuhn Syndrome). Lung, 2015, 193, 815-822.	1.4	27
62	Anterolateral diaphragmatic hernia with body wall defect understood in relation to the abaxial domain. American Journal of Medical Genetics, Part A, 2014, 164, 1860-1862.	0.7	1
63	Diffuse angiopathy in Adamsâ€Oliver syndrome associated with truncating DOCK6 mutations. American Journal of Medical Genetics, Part A, 2014, 164, 2656-2662.	0.7	32
64	Mitochondrial Carbonic Anhydrase VA Deficiency Resulting from CA5A Alterations Presents with Hyperammonemia in Early Childhood. American Journal of Human Genetics, 2014, 94, 453-461.	2.6	82
65	Additional post-natal diagnoses following antenatal diagnosis of isolated cleft lip +/â˜ palate. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2014, 99, F286-F290.	1.4	14
66	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.	0.9	15
67	Mutations in NOTCH1 Cause Adams-Oliver Syndrome. American Journal of Human Genetics, 2014, 95, 275-284.	2.6	150
68	Corneal findings in Parryâ€“Romberg syndrome. Canadian Journal of Ophthalmology, 2014, 49, e2-e5.	0.4	5
69	Evidence of ancillary trigeminal innervation of levator palpebrae in the general population. Journal of Clinical Neuroscience, 2014, 21, 301-304.	0.8	16
70	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	2.6	196
71	Treatable inborn errors of metabolism causing neurological symptoms in adults. Molecular Genetics and Metabolism, 2013, 110, 431-438.	0.5	16
72	Mutations in B4GALNT1 (GM2 synthase) underlie a new disorder of ganglioside biosynthesis. Brain, 2013, 136, 3618-3624.	3.7	115

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73	Fetal Progeria: Prenatal Sonographic Findings in Petty Syndrome. Journal of Ultrasound in Medicine, 2013, 32, 881-883.	0.8	0
74	Child Neurology: Krabbe disease. Neurology, 2012, 79, e170-2.	1.5	6
75	Emphysema in an adult with galactosialidosis linked to a defect in primary elastic fiber assembly. Molecular Genetics and Metabolism, 2012, 106, 99-103.	0.5	15
76	19p13.2 microduplication causes a Sotos syndromeâ€like phenotype and alters gene expression. Clinical Genetics, 2012, 81, 56-63.	1.0	14
77	Coâ€occurrence of Joubert syndrome and Jeune asphyxiating thoracic dystrophy. American Journal of Medical Genetics, Part A, 2010, 152A, 1411-1419.	0.7	30
78	<i>OCRL1</i> Mutations in Dent 2 Patients Suggest a Mechanism for Phenotypic Variability. Nephron Physiology, 2009, 112, p27-p36.	1.5	79
79	Intracranial Calcification after Cord Blood Neonatal Transplantation for Krabbe Disease. Neuropediatrics, 2009, 40, 189-191.	0.3	7
80	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. European Journal of Medical Genetics, 2009, 52, 436-439.	0.7	21
81	Childhood-onset hemiatrophy caused by unilateral morphea. Clinical Dysmorphology, 2009, 18, 213-214.	0.1	6
82	Schinzelâ€“Giedion syndrome: Report of splenopancreatic fusion and proposed diagnostic criteria. American Journal of Medical Genetics, Part A, 2008, 146A, 1299-1306.	0.7	45
83	Causal Attributions, Perceived Control, and Psychological Adjustment: A Study of Chronic Fatigue Syndrome¹. Journal of Applied Social Psychology, 2006, 36, 75-99.	1.3	20
84	Transcriptional Regulation of BACE1, the Î²-Amyloid Precursor Protein Î²-Secretase, by Sp1. Molecular and Cellular Biology, 2004, 24, 865-874.	1.1	207
85	Illness experience, depression, and anxiety in chronic fatigue syndrome. Journal of Psychosomatic Research, 2002, 52, 461-465.	1.2	41