Lynne M Bird

List of Publications by Year
in descending order

69 papers	3,538 citations	172443 29 h-index	149686 56 g-index
72 all docs	72 docs citations	72 times ranked	5740 citing authors

#	Article	IF	Citations
1	Identifying facial phenotypes of genetic disorders using deep learning. Nature Medicine, 2019, 25, 60-64.	30.7	449
2	X-linked situs abnormalities result from mutations in ZIC3. Nature Genetics, 1997, 17, 305-308.	21.4	406
3	Transcriptional repressor ZEB2 promotes terminal differentiation of CD8+ effector and memory T cell populations during infection. Journal of Experimental Medicine, 2015, 212, 2027-2039.	8.5	206
4	Angelman syndrome: review of clinical and molecular aspects. The Application of Clinical Genetics, 2014, 7, 93.	3.0	167
5	A Neurodevelopmental Survey of Angelman Syndrome With Genotype-Phenotype Correlations. Journal of Developmental and Behavioral Pediatrics, 2010, 31, 592-601.	1.1	123
6	Angelman Syndrome. Neurotherapeutics, 2015, 12, 641-650.	4.4	112
7	An RCT of Rapid Genomic Sequencing among Seriously Ill Infants Results in High Clinical Utility, Changes in Management, and Low Perceived Harm. American Journal of Human Genetics, 2020, 107, 942-952.	6.2	110
8	Angelman syndrome: Mutations influence features in early childhood. American Journal of Medical Genetics, Part A, 2011, 155, 81-90.	1.2	109
9	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. European Journal of Human Genetics, 2016, 24, 652-659.	2.8	108
10	Defining the phenotypic spectrum of SLC6A1 mutations. Epilepsia, 2018, 59, 389-402.	5.1	99
11	Effects of <scp>MetAP2</scp> inhibition on hyperphagia and body weight in Praderâ€“Willi syndrome: A randomized, doubleâ€blind, placeboâ€controlled trial. Diabetes, Obesity and Metabolism, 2017, 19, 1751-1761.	4.4	88
12	If not Angelman, what is it? a review of Angelmanâ€like syndromes. American Journal of Medical Genetics, Part A, 2014, 164, 975-992.	1.2	80
13	Identification of novel deletions of 15q11q13 in Angelman syndrome by array-CGH: molecular characterization and genotypeâ€“phenotype correlations. European Journal of Human Genetics, 2007, 15, 943-949.	2.8	75
14	Delta rhythmicity is a reliable EEG biomarker in Angelman syndrome: a parallel mouse and human analysis. Journal of Neurodevelopmental Disorders, 2017, 9, 17.	3.1	74
15	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
16	Female infant with oncocytic cardiomyopathy and microphthalmia with linear skin defects (MLS): A clue to the pathogenesis of oncocytic cardiomyopathy?. American Journal of Medical Genetics Part A, 1994, 53, 141-148.	2.4	69
17	Electrophysiological Phenotype in Angelman Syndrome Differs Between Genotypes. Biological Psychiatry, 2019, 85, 752-759.	1.3	65
18	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	6.2	62

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19	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
20	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. American Journal of Human Genetics, 2020, 106, 596-610.	6.2	59
21	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
22	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217.	6.2	51
23	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
24	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. American Journal of Human Genetics, 2017, 101, 139-148.	6.2	45
25	Abnormal coherence and sleep composition in children with Angelman syndrome: a retrospective EEG study. Molecular Autism, 2018, 9, 32.	4.9	44
26	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. American Journal of Human Genetics, 2017, 101, 516-524.	6.2	43
27	Angelman syndrome genotypes manifest varying degrees of clinical severity and developmental impairment. Molecular Psychiatry, 2021, 26, 3625-3633.	7.9	41
28	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
29	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37
30	Measuring What Matters to Individuals with Angelman Syndrome and Their Families: Development of a Patient-Centered Disease Concept Model. Child Psychiatry and Human Development, 2021, 52, 654-668.	1.9	34
31	A new syndrome of tufting enteropathy and choanal atresia, with ophthalmologic, hematologic and hair abnormalities. Clinical Dysmorphology, 2007, 16, 211-221.	0.3	33
32	A novel SAMD9 mutation causing MIRAGE syndrome: An expansion and review of phenotype, dysmorphology, and natural history. American Journal of Medical Genetics, Part A, 2018, 176, 415-420.	1.2	30
33	Maladaptive behaviors in individuals with Angelman syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 983-992.	1.2	29
34	Cortical dysgenesis in 2 patients with chromosome 22q11 deletion. Clinical Genetics, 2000, 58, 64-68.	2.0	28
35	Recurrence of diaphragmatic agenesis associated with multiple midline defects: Evidence for an autosomal gene regulating the midline. American Journal of Medical Genetics Part A, 1994, 53, 33-38.	2.4	27
36	Chromosome 2q duplications: Case report of a de novo interstitial duplication and review of the literature. American Journal of Medical Genetics Part A, 2001, 100, 13-24.	2.4	25

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37	Rare SUZ12 variants commonly cause an overgrowth phenotype. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 532-547.	1.6	23
38	Delta power robustly predicts cognitive function in Angelman syndrome. Annals of Clinical and Translational Neurology, 2021, 8, 1433-1445.	3.7	23
39	Healthcare burden among individuals with Angelman syndrome: Findings from the Angelman Syndrome Natural History Study. Molecular Genetics & Genomic Medicine, 2019, 7, e00734.	1.2	22
40	Developmental Skills of Individuals with Angelman Syndrome Assessed Using the Bayley-III. Journal of Autism and Developmental Disorders, 2023, 53, 720-737.	2.7	22
41	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
42	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
43	A randomized controlled trial of levodopa in patients with Angelman syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1099-1107.	1.2	18
44	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. American Journal of Medical Genetics, Part A, 2019, 179, 542-551.	1.2	16
45	High-voltage, diffuse delta rhythms coincide with wakeful consciousness and complexity in Angelman syndrome. Neuroscience of Consciousness, 2020, 2020, niaa005.	2.6	15
46	Preserved expressive language as a phenotypic determinant of Mosaic Angelman Syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e837.	1.2	14
47	Patterns of malformation associated with esophageal atresia/tracheoesophageal fistula: A retrospective single center study. American Journal of Medical Genetics, Part A, 2020, 182, 1351-1363.	1.2	14
48	A multidisciplinary approach and consensus statement to establish standards of care for Angelman syndrome. Molecular Genetics & Genomic Medicine, 2022, 10, e1843.	1.2	14
49	Electrophysiological Abnormalities in Angelman Syndrome Correlate With Symptom Severity. Biological Psychiatry Global Open Science, 2021, 1, 201-209.	2.2	13
50	The STARS Phase 2 Study. Neurology, 2021, 96, e1024-e1035.	1.1	12
51	Pharmacological therapies for Angelman syndrome. Wiener Medizinische Wochenschrift, 2017, 167, 205-218.	1.1	10
52	Clinical Characterization of Epilepsy in Children With Angelman Syndrome. Pediatric Neurology, 2021, 124, 42-50.	2.1	9
53	Syndromic neurodevelopmental disorder associated with de novo variants in <scp>DDX23</scp>. American Journal of Medical Genetics, Part A, 2021, 185, 2863-2872.	1.2	8
54	Postmortem Diagnostic Exome Sequencing Identifies a De Novo TUBB3 Alteration in a Newborn With Prenatally Diagnosed Hydrocephalus and Suspected Walkerâ€“Warburg Syndrome. Pediatric and Developmental Pathology, 2018, 21, 319-323.	1.0	7

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55	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. American Journal of Medical Genetics, Part A, 2019, 179, 1543-1546.	1.2	5
56	An observational study of pediatric healthcare burden in Angelman syndrome: results from a real-world study. Orphanet Journal of Rare Diseases, 2019, 14, 239.	2.7	5
57	The Efficacy, Safety, and Pharmacology of a Ghrelin O-Acyltransferase Inhibitor for the Treatment of Prader-Willi Syndrome. Journal of Clinical Endocrinology and Metabolism, 2022, 107, e2373-e2380.	3.6	5
58	Longitudinal EEG model detects antisense oligonucleotide treatment effect and increased UBE3A in Angelman syndrome. Brain Communications, 2022, 4, .	3.3	5
59	The MAP3K7 gene: Further delineation of clinical characteristics and genotype/phenotype correlations. Human Mutation, 2022, 43, 1377-1395.	2.5	5
60	Tuberous sclerosis, polycystic kidney disease and mucolipidosis III gamma caused by a microdeletion unmasking a recessive mutation. American Journal of Medical Genetics, Part A, 2015, 167, 2844-2846.	1.2	4
61	Differentiating molecular etiologies of Angelman syndrome through facial phenotyping using deep learning. American Journal of Medical Genetics, Part A, 2020, 182, 2021-2026.	1.2	4
62	Ending a diagnostic odyssey: Moving from exome to genome to identify cockayne syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1623.	1.2	3
63	A placebo-controlled trial of folic acid and betaine in identical twins with Angelman syndrome. Orphanet Journal of Rare Diseases, 2019, 14, 232.	2.7	2
64	Genetic diagnoses and associated anomalies in fetuses prenatally diagnosed with esophageal atresia. American Journal of Medical Genetics, Part A, 2020, 182, 1890-1895.	1.2	2
65	Anxiety in Angelman Syndrome. American Journal on Intellectual and Developmental Disabilities, 2022, 127, 1-10.	1.6	2
66	Haploinsufficiency of POU4F1 causes an ataxia syndrome with hypotonia and intention tremor. Human Mutation, 2021, 42, 685-693.	2.5	0
67	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
68	SAT-057 A Novel IGSF1 Variant in a Boy with Central Hypothyroidism and Epiphyseal Dysplasia. Journal of the Endocrine Society, 2020, 4, .	0.2	0
69	A unique pancreatic phenotype in a child with a <scp>WDR19</scp>â€related ciliopathy: A case report and literature review ofÂpancreatic involvement in ciliopathies. American Journal of Medical Genetics, Part A, 2022, 188, 2242-2245.	1.2	0