

Lynne M Bird

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.

65
papers

2,261
citations

25
h-index

47
g-index

72
ext. papers

3,031
ext. citations

6.3
avg, IF

5.11
L-index

#	Paper	IF	Citations
65	X-linked situs abnormalities result from mutations in ZIC3. <i>Nature Genetics</i> , 1997 , 17, 305-8	36.3	367
64	Identifying facial phenotypes of genetic disorders using deep learning. <i>Nature Medicine</i> , 2019 , 25, 60-64	50.5	229
63	Angelman syndrome: review of clinical and molecular aspects. <i>The Application of Clinical Genetics</i> , 2014 , 7, 93-104	3.1	128
62	Transcriptional repressor ZEB2 promotes terminal differentiation of CD8+ effector and memory T cell populations during infection. <i>Journal of Experimental Medicine</i> , 2015 , 212, 2027-39	16.6	108
61	A neurodevelopmental survey of Angelman syndrome with genotype-phenotype correlations. <i>Journal of Developmental and Behavioral Pediatrics</i> , 2010 , 31, 592-601	2.4	94
60	Angelman syndrome: Mutations influence features in early childhood. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 81-90	2.5	83
59	SAT-057 A Novel IGSF1 Variant in a Boy with Central Hypothyroidism and Epiphyseal Dysplasia. <i>Journal of the Endocrine Society</i> , 2020 , 4,	0.4	78
58	Angelman Syndrome. <i>Neurotherapeutics</i> , 2015 , 12, 641-50	6.4	75
57	Identification of novel deletions of 15q11q13 in Angelman syndrome by array-CGH: molecular characterization and genotype-phenotype correlations. <i>European Journal of Human Genetics</i> , 2007 , 15, 943-9	5.3	71
56	If not Angelman, what is it? A review of Angelman-like syndromes. <i>American Journal of Medical Genetics, Part A</i> , 2014 , 164A, 975-92	2.5	67
55	Effects of MetAP2 inhibition on hyperphagia and body weight in Prader-Willi syndrome: A randomized, double-blind, placebo-controlled trial. <i>Diabetes, Obesity and Metabolism</i> , 2017 , 19, 1751-1761	6.7	60
54	Female infant with oncocyctic cardiomyopathy and microphthalmia with linear skin defects (MLS): a clue to the pathogenesis of oncocyctic cardiomyopathy?. <i>American Journal of Medical Genetics Part A</i> , 1994 , 53, 141-8		58
53	The Koolen-de Vries syndrome: a phenotypic comparison of patients with a 17q21.31 microdeletion versus a KANSL1 sequence variant. <i>European Journal of Human Genetics</i> , 2016 , 24, 652-9	5.3	57
52	Defining the phenotypic spectrum of SLC6A1 mutations. <i>Epilepsia</i> , 2018 , 59, 389-402	6.4	54
51	Delta rhythmicity is a reliable EEG biomarker in Angelman syndrome: a parallel mouse and human analysis. <i>Journal of Neurodevelopmental Disorders</i> , 2017 , 9, 17	4.6	47
50	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotype-phenotype correlation. <i>Genetics in Medicine</i> , 2019 , 21, 867-876	8.1	43
49	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. <i>American Journal of Human Genetics</i> , 2017 , 100, 117-127	11	39

48	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. <i>American Journal of Human Genetics</i> , 2017 , 101, 206-217	11	38
47	Electrophysiological Phenotype in Angelman Syndrome Differs Between Genotypes. <i>Biological Psychiatry</i> , 2019 , 85, 752-759	7.9	33
46	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. <i>American Journal of Human Genetics</i> , 2019 , 104, 1210-1222	11	31
45	An RCT of Rapid Genomic Sequencing among Seriously Ill Infants Results in High Clinical Utility, Changes in Management, and Low Perceived Harm. <i>American Journal of Human Genetics</i> , 2020 , 107, 942-952	11	31
44	WDR26 Haploinsufficiency Causes a Recognizable Syndrome of Intellectual Disability, Seizures, Abnormal Gait, and Distinctive Facial Features. <i>American Journal of Human Genetics</i> , 2017 , 101, 139-148	11	31
43	A new syndrome of tufting enteropathy and choanal atresia, with ophthalmologic, hematologic and hair abnormalities. <i>Clinical Dysmorphology</i> , 2007 , 16, 211-21	0.9	30
42	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. <i>American Journal of Human Genetics</i> , 2017 , 101, 516-524	11	29
41	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. <i>American Journal of Human Genetics</i> , 2020 , 106, 596-610	11	26
40	Recurrence of diaphragmatic agenesis associated with multiple midline defects: evidence for an autosomal gene regulating the midline. <i>American Journal of Medical Genetics Part A</i> , 1994 , 53, 33-8		25
39	Cornelia de Lange syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 150-158	2.5	25
38	A novel SAMD9 mutation causing MIRAGE syndrome: An expansion and review of phenotype, dysmorphology, and natural history. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 415-420	2.5	24
37	Abnormal coherence and sleep composition in children with Angelman syndrome: a retrospective EEG study. <i>Molecular Autism</i> , 2018 , 9, 32	6.5	24
36	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2018 , 102, 1195-1203	11	24
35	Chromosome 2q duplications: Case report of a de novo interstitial duplication and review of the literature. <i>American Journal of Medical Genetics Part A</i> , 2001 , 100, 13-24		23
34	Cortical dysgenesis in 2 patients with chromosome 22q11 deletion. <i>Clinical Genetics</i> , 2000 , 58, 64-8	4	21
33	A dyadic approach to the delineation of diagnostic entities in clinical genomics. <i>American Journal of Human Genetics</i> , 2021 , 108, 8-15	11	19
32	Angelman syndrome genotypes manifest varying degrees of clinical severity and developmental impairment. <i>Molecular Psychiatry</i> , 2021 , 26, 3625-3633	15.1	16
31	Maladaptive behaviors in individuals with Angelman syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 983-992	2.5	14

30	A randomized controlled trial of levodopa in patients with Angelman syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 1099-1107	2.5	13
29	Healthcare burden among individuals with Angelman syndrome: Findings from the Angelman Syndrome Natural History Study. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e00734	2.3	11
28	Measuring What Matters to Individuals with Angelman Syndrome and Their Families: Development of a Patient-Centered Disease Concept Model. <i>Child Psychiatry and Human Development</i> , 2021 , 52, 654-668	2.3	10
27	Preserved expressive language as a phenotypic determinant of Mosaic Angelman Syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2019 , 7, e837	2.3	9
26	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021 , 13, 63	14.4	9
25	Widening of the genetic and clinical spectrum of Lamb-Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. <i>Genetics in Medicine</i> , 2020 , 22, 524-537	8.1	9
24	Delta power robustly predicts cognitive function in Angelman syndrome. <i>Annals of Clinical and Translational Neurology</i> , 2021 , 8, 1433-1445	5.3	8
23	Rare SUZ12 variants commonly cause an overgrowth phenotype. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2019 , 181, 532-547	3.1	8
22	Pharmacological therapies for Angelman syndrome. <i>Wiener Medizinische Wochenschrift</i> , 2017 , 167, 205-218	2.1	7
21	High-voltage, diffuse delta rhythms coincide with wakeful consciousness and complexity in Angelman syndrome. <i>Neuroscience of Consciousness</i> , 2020 , 2020, niaa005	3.3	6
20	Developmental Skills of Individuals with Angelman Syndrome Assessed Using the Bayley-III. <i>Journal of Autism and Developmental Disorders</i> , 2021 , 1	4.6	6
19	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 542-551	2.5	5
18	Tuberous sclerosis, polycystic kidney disease and mucopolipidosis III gamma caused by a microdeletion unmasking a recessive mutation. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167A, 2844-6	2.5	4
17	The STARS Phase 2 Study: A Randomized Controlled Trial of Gaboxadol in Angelman Syndrome. <i>Neurology</i> , 2021 , 96, e1024-e1035	6.5	4
16	Patterns of malformation associated with esophageal atresia/tracheoesophageal fistula: A retrospective single center study. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1351-1363	2.5	4
15	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. <i>American Journal of Medical Genetics, Part A</i> , 2019 , 179, 1543-1546	2.5	3
14	Postmortem Diagnostic Exome Sequencing Identifies a De Novo TUBB3 Alteration in a Newborn With Prenatally Diagnosed Hydrocephalus and Suspected Walker-Warburg Syndrome. <i>Pediatric and Developmental Pathology</i> , 2018 , 21, 319-323	2.2	3
13	Syndromic neurodevelopmental disorder associated with de novo variants in DDX23. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 2863-2872	2.5	3

12	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. <i>Pediatric Neurology</i> , 2021 , 123, 30-37	2.9	3
11	Differentiating molecular etiologies of Angelman syndrome through facial phenotyping using deep learning. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 2021-2026	2.5	2
10	A placebo-controlled trial of folic acid and betaine in identical twins with Angelman syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 232	4.2	1
9	A multidisciplinary approach and consensus statement to establish standards of care for Angelman syndrome.. <i>Molecular Genetics & Genomic Medicine</i> , 2022 , e1843	2.3	1
8	An observational study of pediatric healthcare burden in Angelman syndrome: results from a real-world study. <i>Orphanet Journal of Rare Diseases</i> , 2019 , 14, 239	4.2	1
7	Electrophysiological Abnormalities in Angelman Syndrome Correlate With Symptom Severity. <i>Biological Psychiatry Global Open Science</i> , 2021 , 1, 201-209		1
6	Clinical Characterization of Epilepsy in Children With Angelman Syndrome. <i>Pediatric Neurology</i> , 2021 , 124, 42-50	2.9	1
5	Genetic diagnoses and associated anomalies in fetuses prenatally diagnosed with esophageal atresia. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 1890-1895	2.5	0
4	Ending a diagnostic odyssey: Moving from exome to genome to identify cockayne syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2021 , 9, e1623	2.3	0
3	Anxiety in Angelman Syndrome.. <i>American Journal on Intellectual and Developmental Disabilities</i> , 2022 , 127, 1-10	2.2	
2	Haploinsufficiency of POU4F1 causes an ataxia syndrome with hypotonia and intention tremor. <i>Human Mutation</i> , 2021 , 42, 685-693	4.7	
1	Response to Hamosh et al. <i>American Journal of Human Genetics</i> , 2021 , 108, 1809-1810	11	