Lynne M Bird

List of Publications by Year in Descending Order

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	Anxiety in Angelman Syndrome.. *American Journal on Intellectual and Developmental Disabilities*, **2022**, 127, 1-10	2.2	
64	A multidisciplinary approach and consensus statement to establish standards of care for Angelman syndrome.. *Molecular Genetics & Genomic Medicine*, **2022**, e1843	2.3	1
63	The STARS Phase 2 Study: A Randomized Controlled Trial of Gaboxadol in Angelman Syndrome. *Neurology*, **2021**, 96, e1024-e1035	6.5	4
62	Angelman syndrome genotypes manifest varying degrees of clinical severity and developmental impairment. *Molecular Psychiatry*, **2021**, 26, 3625-3633	15.1	16
61	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	3.3	10
60	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. *Genome Medicine*, **2021**, 13, 63	14.4	9
59	Haploinsufficiency of POU4F1 causes an ataxia syndrome with hypotonia and intention tremor. *Human Mutation*, **2021**, 42, 685-693	4.7	
58	Delta power robustly predicts cognitive function in Angelman syndrome. *Annals of Clinical and Translational Neurology*, **2021**, 8, 1433-1445	5.3	8
57	Syndromic neurodevelopmental disorder associated with de novo variants in DDX23. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2863-2872	2.5	3
56	Ending a diagnostic odyssey: Moving from exome to genome to identify cockayne syndrome. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1623	2.3	0
55	A dyadic approach to the delineation of diagnostic entities in clinical genomics. *American Journal of Human Genetics*, **2021**, 108, 8-15	11	19
54	Developmental Skills of Individuals with Angelman Syndrome Assessed Using the Bayley-III. *Journal of Autism and Developmental Disorders*, **2021**, 1	4.6	6
53	Electrophysiological Abnormalities in Angelman Syndrome Correlate With Symptom Severity. *Biological Psychiatry Global Open Science*, **2021**, 1, 201-209		1
52	Response to Hamosh et al. *American Journal of Human Genetics*, **2021**, 108, 1809-1810	11	
51	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. *Pediatric Neurology*, **2021**, 123, 30-37	2.9	3
50	Clinical Characterization of Epilepsy in Children With Angelman Syndrome. *Pediatric Neurology*, **2021**, 124, 42-50	2.9	1
49	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	11	31

48	High-voltage, diffuse delta rhythms coincide with wakeful consciousness and complexity in Angelman syndrome. *Neuroscience of Consciousness*, **2020**, 2020, niaa005	3.3	6
47	Genetic diagnoses and associated anomalies in fetuses prenatally diagnosed with esophageal atresia. *American Journal of Medical Genetics, Part A*, **2020**, 182, 1890-1895	2.5	0
46	Differentiating molecular etiologies of Angelman syndrome through facial phenotyping using deep learning. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2021-2026	2.5	2
45	SAT-057 A Novel IGSF1 Variant in a Boy with Central Hypothyroidism and Epiphyseal Dysplasia. *Journal of the Endocrine Society*, **2020**, 4,	0.4	78
44	Widening of the genetic and clinical spectrum of Lamb-Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. *Genetics in Medicine*, **2020**, 22, 524-537	8.1	9
43	DNA Methylation Signature for EZH2 Functionally Classifies Sequence Variants in Three PRC2 Complex Genes. *American Journal of Human Genetics*, **2020**, 106, 596-610	11	26
42	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	2.5	4
41	A placebo-controlled trial of folic acid and betaine in identical twins with Angelman syndrome. *Orphanet Journal of Rare Diseases*, **2019**, 14, 232	4.2	1
40	Electrophysiological Phenotype in Angelman Syndrome Differs Between Genotypes. *Biological Psychiatry*, **2019**, 85, 752-759	7.9	33
39	Perinatal distress in 1p36 deletion syndrome can mimic hypoxic ischemic encephalopathy. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1543-1546	2.5	3
38	Healthcare burden among individuals with Angelman syndrome: Findings from the Angelman Syndrome Natural History Study. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00734	2.3	11
37	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
36	Maladaptive behaviors in individuals with Angelman syndrome. *American Journal of Medical Genetics, Part A*, **2019**, 179, 983-992	2.5	14
35	Hyperinsulinemic hypoglycemia in seven patients with de novo NSD1 mutations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 542-551	2.5	5
34	Preserved expressive language as a phenotypic determinant of Mosaic Angelman Syndrome. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e837	2.3	9
33	Rare SUZ12 variants commonly cause an overgrowth phenotype. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2019**, 181, 532-547	3.1	8
32	An observational study of pediatric healthcare burden in Angelman syndrome: results from a real-world study. *Orphanet Journal of Rare Diseases*, **2019**, 14, 239	4.2	1
31	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. *Genetics in Medicine*, **2019**, 21, 867-876	8.1	43

30	Identifying facial phenotypes of genetic disorders using deep learning. *Nature Medicine*, **2019**, 25, 60-64	50.5	229
29	Cornelia de Lange syndrome in diverse populations. *American Journal of Medical Genetics, Part A*, **2019**, 179, 150-158	2.5	25
28	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	2.5	24
27	Defining the phenotypic spectrum of SLC6A1 mutations. *Epilepsia*, **2018**, 59, 389-402	6.4	54
26	Postmortem Diagnostic Exome Sequencing Identifies a De Novo TUBB3 Alteration in a Newborn With Prenatally Diagnosed Hydrocephalus and Suspected Walker-Warburg Syndrome. *Pediatric and Developmental Pathology*, **2018**, 21, 319-323	2.2	3
25	A randomized controlled trial of levodopa in patients with Angelman syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1099-1107	2.5	13
24	Abnormal coherence and sleep composition in children with Angelman syndrome: a retrospective EEG study. *Molecular Autism*, **2018**, 9, 32	6.5	24
23	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	11	24
22	Pharmacological therapies for Angelman syndrome. *Wiener Medizinische Wochenschrift*, **2017**, 167, 205-218	2.9	7
21	Effects of MetAP2 inhibition on hyperphagia and body weight in Prader-Willi syndrome: A randomized, double-blind, placebo-controlled trial. *Diabetes, Obesity and Metabolism*, **2017**, 19, 1751-1761	6.7	60
20	Delta rhythmicity is a reliable EEG biomarker in Angelman syndrome: a parallel mouse and human analysis. *Journal of Neurodevelopmental Disorders*, **2017**, 9, 17	4.6	47
19	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. *American Journal of Human Genetics*, **2017**, 100, 117-127	11	39
18	De Novo Mutations in PPP3CA Cause Severe Neurodevelopmental Disease with Seizures. *American Journal of Human Genetics*, **2017**, 101, 516-524	11	29
17	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. *American Journal of Human Genetics*, **2017**, 101, 206-217	11	38
16	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	11	31
15	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, 652-9	5.3	57
14	Angelman Syndrome. *Neurotherapeutics*, **2015**, 12, 641-50	6.4	75
13	Transcriptional repressor ZEB2 promotes terminal differentiation of CD8+ effector and memory T cell populations during infection. *Journal of Experimental Medicine*, **2015**, 212, 2027-39	16.6	108

12	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**, 167A, 2844-6	2.5	4
11	If not Angelman, what is it? A review of Angelman-like syndromes. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 975-92	2.5	67
10	Angelman syndrome: review of clinical and molecular aspects. *The Application of Clinical Genetics*, **2014**, 7, 93-104	3.1	128
9	Angelman syndrome: Mutations influence features in early childhood. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 81-90	2.5	83
8	A neurodevelopmental survey of Angelman syndrome with genotype-phenotype correlations. *Journal of Developmental and Behavioral Pediatrics*, **2010**, 31, 592-601	2.4	94
7	Identification of novel deletions of 15q11q13 in Angelman syndrome by array-CGH: molecular characterization and genotype-phenotype correlations. *European Journal of Human Genetics*, **2007**, 15, 943-9	5.3	71
6	A new syndrome of tufting enteropathy and choanal atresia, with ophthalmologic, hematologic and hair abnormalities. *Clinical Dysmorphology*, **2007**, 16, 211-21	0.9	30
5	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		23
4	Cortical dysgenesis in 2 patients with chromosome 22q11 deletion. *Clinical Genetics*, **2000**, 58, 64-8	4	21
3	X-linked situs abnormalities result from mutations in ZIC3. *Nature Genetics*, **1997**, 17, 305-8	36.3	367
2	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, 33-8		25
1	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, 141-8		58