Elizabeth J Bhoj

List of Publications by Year
in descending order

82 papers	1,713 citations	331670 21 h-index	361022 35 g-index
88 all docs	88 docs citations	88 times ranked	3975 citing authors

#	Article	IF	Citations
1	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
2	ARAF recurrent mutation causes central conducting lymphatic anomaly treatable with a MEK inhibitor. Nature Medicine, 2019, 25, 1116-1122.	30.7	136
3	Pathogenic variant in EPHB4 results in central conducting lymphatic anomaly. Human Molecular Genetics, 2018, 27, 3233-3245.	2.9	73
4	Gene domain-specific DNA methylation episignatures highlight distinct molecular entities of ADNP syndrome. Clinical Epigenetics, 2019, 11, 64.	4.1	71
5	Maternal uniparental disomy of chromosome 20: a novel imprinting disorder of growth failure. Genetics in Medicine, 2016, 18, 309-315.	2.4	69
6	Automated Clinical Exome Reanalysis Reveals Novel Diagnoses. Journal of Molecular Diagnostics, 2019, 21, 38-48.	2.8	68
7	De Novo Mutations of RERE Cause a Genetic Syndrome with Features that Overlap Those Associated with Proximal 1p36 Deletions. American Journal of Human Genetics, 2016, 98, 963-970.	6.2	67
8	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebroâ€“costoâ€“mandibular syndrome. Nature Communications, 2014, 5, 4483.	12.8	57
9	Mapping RNA splicing variations in clinically accessible and nonaccessible tissues to facilitate Mendelian disease diagnosis using RNA-seq. Genetics in Medicine, 2020, 22, 1181-1190.	2.4	54
10	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. American Journal of Human Genetics, 2016, 98, 782-788.	6.2	50
11	Monoallelic BMP2 Variants Predicted to Result in Haploinsufficiency Cause Craniofacial, Skeletal, and Cardiac Features Overlapping Those of 20p12 Deletions. American Journal of Human Genetics, 2017, 101, 985-994.	6.2	44
12	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, .	10.3	43
13	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	1.3	42
14	De novo variants in Myelin regulatory factor (MYRF) as candidates of a new syndrome of cardiac and urogenital anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 969-972.	1.2	39
15	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
16	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
17	Truncating SRCAP variants outside the Floating-Harbor syndrome locus cause a distinct neurodevelopmental disorder with a specific DNA methylation signature. American Journal of Human Genetics, 2021, 108, 1053-1068.	6.2	31
18	Novel findings with reassessment of exome data: implications for validation testing and interpretation of genomic data. Genetics in Medicine, 2018, 20, 329-336.	2.4	28

#	Article	IF	Citations
19	Tracheal cartilaginous sleeves in children with syndromic craniosynostosis. Genetics in Medicine, 2017, 19, 62-68.	2.4	27
20	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.	2.8	27
21	Expanding the SPECC1L mutation phenotypic spectrum to include Teebi hypertelorism syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 2497-2502.	1.2	26
22	Exome sequencing expands the mechanism of SOX5â€associated intellectual disability: A case presentation with review of soxâ€related disorders. American Journal of Medical Genetics, Part A, 2015, 167, 2548-2554.	1.2	26
23	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
24	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25
25	Phenotypic spectrum associated with SPECC1L pathogenic variants: new families and critical review of the nosology of Teebi, Opitz GBBB, and Baraitser-Winter syndromes. European Journal of Medical Genetics, 2019, 62, 103588.	1.3	24
26	A genome-wide association study of anorexia nervosa suggests a risk locus implicated in dysregulated leptin signaling. Scientific Reports, 2017, 7, 3847.	3.3	23
27	Cerebroâ€“costoâ€“mandibular syndrome: Clinical, radiological, and genetic findings. American Journal of Medical Genetics, Part A, 2016, 170, 1115-1126.	1.2	21
28	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
29	Activating variants in <scp>PDGFRB</scp> result in a spectrum of disorders responsive to imatinib monotherapy. American Journal of Medical Genetics, Part A, 2020, 182, 1576-1591.	1.2	21
30	Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human Molecular Genetics, 2020, 29, 1900-1921.	2.9	21
31	Expanding the spectrum of microdeletion 4q21 syndrome: A partial phenotype with incomplete deletion of the minimal critical region and a new association with cleft palate and pierre robin sequence. American Journal of Medical Genetics, Part A, 2013, 161, 2327-2333.	1.2	20
32	SMARCE1, a rare cause of Coffinâ€“Siris Syndrome: Clinical description of three additional cases. American Journal of Medical Genetics, Part A, 2016, 170, 1967-1973.	1.2	18
33	An Additional Individual with a De Novo Variant in Myelin Regulatory Factor (MYRF) with Cardiac and Urogenital Anomalies: Further Proof of Causality: Comments on the article by Pinz et al. (). American Journal of Medical Genetics, Part A, 2018, 176, 2041-2043.	1.2	18
34	Bi-allelic Loss-of-Function Variants in NUP188 Cause a Recognizable Syndrome Characterized by Neurologic, Ocular, and Cardiac Abnormalities. American Journal of Human Genetics, 2020, 106, 623-631.	6.2	18
35	Pathogenic variants in SMARCA5 , a chromatin remodeler, cause a range of syndromic neurodevelopmental features. Science Advances, 2021, 7, .	10.3	17
36	EP300 â€related Rubinsteinâ€“Taybi syndrome: Highlighted rare phenotypic findings and a genotypeâ€–phenotype metaâ€analysis of 74 patients. American Journal of Medical Genetics, Part A, 2020, 182, 2926-2938.	1.2	16

#	Article	IF	Citations
37	De novo loss-of-function variants in X-linked MED12 are associated with Hardikar syndrome in females. Genetics in Medicine, 2021, 23, 637-644.	2.4	16
38	Phenotypic predictors and final diagnoses in patients referred for RASopathy testing by targeted next-generation sequencing. Genetics in Medicine, 2017, 19, 715-718.	2.4	14
39	De novo variants in CACNA1E found in patients with intellectual disability, developmental regression and social cognition deficit but no seizures. Molecular Autism, 2021, 12, 69.	4.9	12
40	Bi-allelic variants in OGDHL cause a neurodevelopmental spectrum disease featuring epilepsy, hearing loss, visual impairment, and ataxia. American Journal of Human Genetics, 2021, 108, 2368-2384.	6.2	12
41	Expanding the phenotypic spectrum of TP63â€related disorders including the first set of monozygotic twins. American Journal of Medical Genetics, Part A, 2018, 176, 75-81.	1.2	11
42	Missense Mutations in NKAP Cause a Disorder of Transcriptional Regulation Characterized by Marfanoid Habitus and Cognitive Impairment. American Journal of Human Genetics, 2019, 105, 987-995.	6.2	11
43	Heterozygous de novo variants in <scp>CSNK1G1</scp> are associated with syndromic developmental delay and autism spectrum disorder. Clinical Genetics, 2020, 98, 571-576.	2.0	10
44	Further delineation of the phenotypic spectrum of nevus comedonicus syndrome to include congenital pulmonary airway malformation of the lung and aneurysm. American Journal of Medical Genetics, Part A, 2020, 182, 746-754.	1.2	9
45	ALG13 Xâ€linked intellectual disability: New variants, glycosylation analysis, and expanded phenotypes. Journal of Inherited Metabolic Disease, 2021, 44, 1001-1012.	3.6	9
46	Congenital diaphragmatic hernia as a prominent feature of a SPECC1L â€related syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2919-2925.	1.2	8
47	Experiences with offering pro bono medical genetics services in the West Indies: Benefits to patients, physicians, and the community. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 1030-1041.	1.6	8
48	Expanding the clinical and phenotypic heterogeneity associated with biallelic variants in ACO2. Annals of Clinical and Translational Neurology, 2020, 7, 1013-1028.	3.7	8
49	Extension of the mutational and clinical spectrum of SOX2 related disorders: Description of six new cases and a novel association with suprasellar teratoma. American Journal of Medical Genetics, Part A, 2018, 176, 2710-2719.	1.2	7
50	Isolated vocal cord paralysis in two siblings with compound heterozygous variants inMUSK: Expanding the phenotypic spectrum. American Journal of Medical Genetics, Part A, 2019, 179, 655-658.	1.2	7
51	Clinical variability of TUBB â€associated disorders: Diagnosis through reanalysis. American Journal of Medical Genetics, Part A, 2020, 182, 3035-3039.	1.2	7
52	Molecular Diagnostic Outcomes from 700 Cases. Journal of Molecular Diagnostics, 2022, 24, 274-286.	2.8	7
53	Early Infantile Epileptic Encephalopathy in anSTXBP1Patient with Lactic Acidemia and Normal Mitochondrial Respiratory Chain Function. Case Reports in Genetics, 2016, 2016, 1-5.	0.2	6
54	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. American Journal of Human Genetics, 2021, 108, 749-756.	6.2	6

#	Article	IF	Citations
55	Clinical Phenotypic Spectrum of 4095 Individuals with Down Syndrome from Text Mining of Electronic Health Records. Genes, 2021, 12, 1159.	2.4	6
56	Variants in ADD1 cause intellectual disability, corpus callosum dysgenesis, and ventriculomegaly in humans. Genetics in Medicine, 2022, 24, 319-331.	2.4	6
57	Variants in PHF8 cause a spectrum of X-linked neurodevelopmental disorders and facial dysmorphology. Human Genetics and Genomics Advances, 2022, 3, 100102.	1.7	5
58	Expanding the phenotypic spectrum of ARCN1-related syndrome. Genetics in Medicine, 2022, 24, 1227-1237.	2.4	5
59	Imprinted genes in clinical exome sequencing: Review of 538 cases and exploration of mouse-human conservation in the identification of novel human disease loci. European Journal of Medical Genetics, 2020, 63, 103903.	1.3	4
60	Pathogenic variants in CDH11 impair cell adhesion and cause Teebi hypertelorism syndrome. Human Genetics, 2021, 140, 1061-1076.	3.8	4
61	Phenotypic modifications of patients with full chromosome aneuploidies and concurrent suspected or confirmed second diagnoses. American Journal of Medical Genetics, Part A, 2015, 167, 2168-2175.	1.2	3
62	Contemporary Evaluation of the Neonate with Congenital Anomalies. NeoReviews, 2017, 18, e522-e531.	0.8	3
63	A homozygous truncating NALCN variant in two Afroâ€Caribbean siblings with hypotonia and dolichocephaly. American Journal of Medical Genetics, Part A, 2020, 182, 1877-1880.	1.2	3
64	Congenital polyvalvular disease expands the cardiac phenotype of the RASopathies. American Journal of Medical Genetics, Part A, 2021, 185, 1486-1493.	1.2	3
65	Genetic skin disorders: The value of a multidisciplinary clinic. American Journal of Medical Genetics, Part A, 2021, 185, 1159-1167.	1.2	3
66	Analysis of histone variant constraint and tissue expression suggests five potential novel human disease genes: H2AFY2, H2AFZ, H2AFY, H2AFV, H1F0. Human Genetics, 2022, 141, 1409-1421.	3.8	3
67	Further supporting <scp>SMARCC2</scp>â€related neurodevelopmental disorder through exome analysis and reanalysis in two patients. American Journal of Medical Genetics, Part A, 2022, 188, 878-882.	1.2	3
68	Application of exome sequencing to diagnose a novel presentation of the Cornelia de Lange syndrome in an Afroâ€Caribbean family. Molecular Genetics & Genomic Medicine, 2020, 8, e1318.	1.2	2
69	Inborn error of metabolism patients after liver transplantation: Outcomes of 35 patients over 27â€‰years in one pediatric quaternary hospital. American Journal of Medical Genetics, Part A, 2022, 188, 1443-1447.	1.2	2
70	Aortic coarctation and carotid artery aneurysm in a patient with hardikar syndrome: Cardiovascular implications for affected individuals. American Journal of Medical Genetics, Part A, 2016, 170, 482-486.	1.2	1
71	An Algorithm for the Assessment of Facial Asymmetry in Children With Focus on Etiology and Treatment. Cleft Palate-Craniofacial Journal, 2019, 56, 419-424.	0.9	1
72	Generalized, severe epidermolysis bullosa simplex caused by a Keratin 5 p.E477K mutation. Pediatric Dermatology, 2019, 36, 1007-1009.	0.9	1

#	Article	IF	Citations
73	Muenke syndrome: Medical and surgical comorbidities and longâ€term management. American Journal of Medical Genetics, Part A, 2019, 179, 1442-1450.	1.2	1
74	3548 De novo germline variants in Histone 3 Family 3A (H3F3A) and Histone 3 Family 3B (H3F3B) cause a severe neurodegenerative disorder and functional effects unique from their somatic mutations. Journal of Clinical and Translational Science, 2019, 3, 103-103.	0.6	1
75	Discovery of a neuromuscular syndrome caused by biallelic variants in ASCC3. Human Genetics and Genomics Advances, 2021, 2, 100024.	1.7	1
76	Cleft palate morphology, genetic etiology, and risk of mortality in infants with Robin sequence. American Journal of Medical Genetics, Part A, 2021, 185, 3694-3700.	1.2	1
77	A novel unbalanced translocation between chromosomes 5p and 18q leading to dysmorphology and global developmental delay. Molecular Genetics & Genomic Medicine, 2022, , e1900.	1.2	1
78	Exome and <scp>RNAâ€Seq</scp> analyses of an incomplete penetrance variant in <scp> USP9X </scp> in femaleâ€specific syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2022, , .	1.2	1
79	Cover Image, Volume 170A, Number 5, May 2016. , 2016, 170, i-i.		0
80	What not to expect when you're expecting: Unusual cases of placental mosaicism detected on non-invasive prenatal screening. European Journal of Medical Genetics, 2020, 63, 103895.	1.3	0
81	Elucidating the clinical spectrum and molecular basis of HYAL2 deficiency. Genetics in Medicine, 2022, 24, 631-644.	2.4	0
82	Contribution of Mendelian disorders in a population-based pediatric neurodegeneration cohort. Journal of Pediatrics, 2022, , .	1.8	0