Beth A Kozel

List of Publications by Year
in descending order

67 papers	2,423 citations	236925 25 h-index	214800 47 g-index
70 all docs	70 docs citations	70 times ranked	3901 citing authors

#	Article	IF	Citations
1	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	3.2	447
2	Elastic fiber formation: A dynamic view of extracellular matrix assembly using timer reporters. Journal of Cellular Physiology, 2006, 207, 87-96.	4.1	170
3	Tropoelastin Interacts with Cell-surface Glycosaminoglycans via Its COOH-terminal Domain. Journal of Biological Chemistry, 2005, 280, 40939-40947.	3.4	138
4	Domains in Tropoelastin That Mediate Elastin Depositionin Vitro and in Vivo. Journal of Biological Chemistry, 2003, 278, 18491-18498.	3.4	122
5	Williams syndrome. Nature Reviews Disease Primers, 2021, 7, 42.	30.5	103
6	Elastic fiber macro-assembly is a hierarchical, cell motion-mediated process. Journal of Cellular Physiology, 2006, 207, 97-106.	4.1	93
7	BMP4 loss-of-function mutations in developmental eye disorders including SHORT syndrome. Human Genetics, 2011, 130, 495-504.	3.8	92
8	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. Genetics in Medicine, 2017, 19, 1040-1048.	2.4	85
9	Infantile cardiomyopathy caused by a mutation in the overlapping region of mitochondrial ATPase 6 and 8 genes. Journal of Medical Genetics, 2009, 46, 308-314.	3.2	84
10	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
11	Williams Syndrome Predisposes to Vascular Stiffness Modified by Antihypertensive Use and Copy Number Changes in NCF1. Hypertension, 2014, 63, 74-79.	2.7	69
12	Elastin-driven genetic diseases. Matrix Biology, 2018, 71-72, 144-160.	3.6	69
13	Deposition of tropoelastin into the extracellular matrix requires a competent elastic fiber scaffold but not live cells. Matrix Biology, 2004, 23, 23-34.	3.6	68
14	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
15	Effects of Obesity and Hypertension on Pulse Wave Velocity in Children. Journal of Clinical Hypertension, 2017, 19, 221-226.	2.0	52
16	Elastic fiber ultrastructure and assembly. Matrix Biology, 2019, 84, 31-40.	3.6	52
17	Hypercalcemia in Patients with Williams-Beuren Syndrome. Journal of Pediatrics, 2016, 178, 254-260.e4.	1.8	45
18	Minoxidil improves vascular compliance, restores cerebral blood flow, and alters extracellular matrix gene expression in a model of chronic vascular stiffness. American Journal of Physiology - Heart and Circulatory Physiology, 2018, 315, H18-H32.	3.2	44

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19	Wholeâ€genome copy number variation analysis in anophthalmia and microphthalmia. Clinical Genetics, 2013, 84, 473-481.	2.0	43
20	Delineation of a deletion region critical for corpus callosal abnormalities in chromosome 1q43â€“q44. European Journal of Human Genetics, 2012, 20, 176-179.	2.8	42
21	Genetic Modifiers of Cardiovascular Phenotype Caused by Elastin Haploinsufficiency Act by Extrinsic Noncomplementation. Journal of Biological Chemistry, 2011, 286, 44926-44936.	3.4	34
22	Altered reactivity of resistance vasculature contributes to hypertension in elastin insufficiency. American Journal of Physiology - Heart and Circulatory Physiology, 2014, 306, H654-H666.	3.2	32
23	Distinct Clinical and Histopathological Presentations of Danon Cardiomyopathy in Young Women. Journal of the American College of Cardiology, 2010, 55, 408-410.	2.8	29
24	KATP channel gain-of-function leads to increased myocardial L-type Ca2+ current and contractility in Cantu syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 6773-6778.	7.1	29
25	Alternative Splicing and Tissue-specific Elastin Misassembly Act as Biological Modifiers of Human Elastin Gene Frameshift Mutations Associated with Dominant Cutis Laxa. Journal of Biological Chemistry, 2012, 287, 22055-22067.	3.4	28
26	A Yeast Artificial Chromosome (YAC) Contig Encompassing the Critical Region of the X-Linked Lymphoproliferative Disease (XLP) Locus. Genomics, 1997, 39, 55-65.	2.9	23
27	Skin findings in Williams syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2217-2225.	1.2	22
28	Chronic antihypertensive treatment improves pulse pressure but not large artery mechanics in a mouse model of congenital vascular stiffness. American Journal of Physiology - Heart and Circulatory Physiology, 2015, 309, H1008-H1016.	3.2	21
29	Prenatal exposure to pesticides and risk for holoprosencephaly: a case-control study. Environmental Health, 2020, 19, 65.	4.0	20
30	Whole exome sequencing in patients with Williamsâ€“Beuren syndrome followed by disease modeling in mice points to four novel pathways that may modify stenosis risk. Human Molecular Genetics, 2020, 29, 2035-2050.	2.9	15
31	Identifying environmental risk factors and <scp>geneâ€“environment</scp> interactions in holoprosencephaly. Birth Defects Research, 2021, 113, 63-76.	1.5	14
32	Developmental vascular malformations in EPAS1 gain-of-function syndrome. JCI Insight, 2021, 6, .	5.0	14
33	Vascular elastic fiber heterogeneity in health and disease. Current Opinion in Hematology, 2020, 27, 190-196.	2.5	14
34	Impaired angiogenesis and extracellular matrix metabolism in autosomal-dominant hyper-IgE syndrome. Journal of Clinical Investigation, 2020, 130, 4167-4181.	8.2	13
35	Copy number analysis of NIPBL in a cohort of 510 patients reveals rare copy number variants and a mosaic deletion. Molecular Genetics & Genomic Medicine, 2014, 2, 115-123.	1.2	12
36	Social, neurodevelopmental, endocrine, and head size differences associated with atypical deletions in Williamsâ€“Beuren syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 1008-1020.	1.2	12

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37	Biomechanical Properties of the Skin in Cutis Laxa. Journal of Investigative Dermatology, 2014, 134, 2836-2838.	0.7	11
38	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. Bone, 2019, 120, 354-363.	2.9	11
39	Inhibition of NOX1 Mitigates Blood Pressure Increases in Elastin Insufficiency. Function, 2021, 2, zqab015.	2.3	10
40	Elastin. , 2011, , 267-301.		10
41	Clinical utility gene card for: Williamsâ€“Beuren Syndrome [7q11.23]. European Journal of Human Genetics, 2014, 22, 1153-1153.	2.8	9
42	Exome sequencing of 85 Williamsâ€“Beuren syndrome cases rules out coding variation as a major contributor to remaining variance in social behavior. Molecular Genetics & Genomic Medicine, 2018, 6, 749-765.	1.2	9
43	Elastin Insufficiency Predisposes Mice to Impaired Glucose Metabolism. Journal of Molecular and Genetic Medicine: an International Journal of Biomedical Research, 2014, 08, .	0.1	8
44	X-linked creatine transporter deficiency results in prolonged QTc and increased sudden death risk in humans and disease model. Genetics in Medicine, 2021, 23, 1864-1872.	2.4	8
45	Cardiac pathologies in mouse loss of imprinting models are due to misexpression of H19 long noncoding RNA. ELife, 2021, 10, .	6.0	8
46	Mild macrocytosis in Williams-Beuren syndrome. European Journal of Medical Genetics, 2020, 63, 103740.	1.3	6
47	Vascular Casting of Adult and Early Postnatal Mouse Lungs for Micro-CT Imaging. Journal of Visualized Experiments, 2020, , .	0.3	6
48	Loss of Angiotensin II Type 2 Receptor Improves Blood Pressure in Elastin Insufficiency. Frontiers in Cardiovascular Medicine, 2021, 8, 782138.	2.4	6
49	Non-invasive in situ visualization of the murine cranial vasculature. Cell Reports Methods, 2022, 2, 100151.	2.9	6
50	Neuraxial dysraphism in EPAS1-associated syndrome due to improper mesenchymal transition. Neurology: Genetics, 2020, 6, e414.	1.9	5
51	Xâ€ray microtomosynthesis of unstained pathology tissue samples. Journal of Microscopy, 2021, 283, 9-20.	1.8	5
52	Variegation of autism related traits across seven neurogenetic disorders. Translational Psychiatry, 2022, 12, 149.	4.8	5
53	Perspectives on Cognitive Phenotypes and Models of Vascular Disease. Arteriosclerosis, Thrombosis, and Vascular Biology, 2022, , 101161ATVBAHA122317395.	2.4	4
54	Copper-Binding Domain Variation in a Novel Murine Lysyl Oxidase Model Produces Structurally Inferior Aortic Elastic Fibers Whose Failure Is Modified by Age, Sex, and Blood Pressure. International Journal of Molecular Sciences, 2022, 23, 6749.	4.1	4

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55	Emerging mechanisms of elastin transcriptional regulation. American Journal of Physiology - Cell Physiology, 2022, 323, C666-C677.	4.6	4
56	Novel ophthalmic findings and deep phenotyping in Williams-Beuren syndrome. British Journal of Ophthalmology, 2023, 107, 1554-1559.	3.9	3
57	Frequency of QTc Interval Prolongation in Children and Adults with Williams Syndrome. Pediatric Cardiology, 2022, 43, 1559-1567.	1.3	2
58	Elastin Insufficiency Confers Proximal and Distal Pulmonary Vasculopathy in Mice, Partially Remedied by the KATP Channel Opener Minoxidil: Considerations and Cautions for the Treatment of People With Williams-Beuren Syndrome. Frontiers in Cardiovascular Medicine, 2022, 9, .	2.4	2
59	448: Maternal carriage of the K121Q single nucleotide polymorphism (SNP) in the ENPP1 (PC-1) gene is associated with preeclampsia in a Hispanic population. American Journal of Obstetrics and Gynecology, 2007, 197, S132.	1.3	1
60	GENERAL ANESTHESIA RISK IN WILLIAMS BEUREN SYNDROME PATIENTS IN THE CURRENT ERA. Journal of the American College of Cardiology, 2017, 69, 598.	2.8	1
61	Special Therapy and Psychosocial Needs Identified in a Multidisciplinary Cancer Predisposition Syndrome Clinic. Journal of Pediatric Hematology/Oncology, 2019, 41, 133-136.	0.6	1
62	Pathology of the Elastic Matrix. , 2016, , 31-80.		1
63	Airflow Obstruction in Adults with Williams Syndrome and Mice with Elastin Insufficiency. Diagnostics, 2022, 12, 1438.	2.6	1
64	705: Maternal carriage of the K121Q single nucleotide polymorphism (SNP) in the ENPP (PC-1) gene is associated with preterm birth and diminished birthweight in a hispanic population. American Journal of Obstetrics and Gynecology, 2007, 197, S201.	1.3	0
65	Cover Image, Volume 176A, Number 5, May 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0
66	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
67	Timeâ€lapse imaging of extracellular matrix assembly. FASEB Journal, 2008, 22, 101.1.	0.5	0