

# Beth A Kozel

## List of Publications by Year in descending order

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Version: 2024-02-01

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	Novel ophthalmic findings and deep phenotyping in Williams-Beuren syndrome. <i>British Journal of Ophthalmology</i> , 2023, 107, 1554-1559.	3.9	3
2	Non-invasive in situ visualization of the murine cranial vasculature. <i>Cell Reports Methods</i> , 2022, 2, 100151.	2.9	6
3	Frequency of QTc Interval Prolongation in Children and Adults with Williams Syndrome. <i>Pediatric Cardiology</i> , 2022, 43, 1559-1567.	1.3	2
4	Variegation of autism related traits across seven neurogenetic disorders. <i>Translational Psychiatry</i> , 2022, 12, 149.	4.8	5
5	Perspectives on Cognitive Phenotypes and Models of Vascular Disease. <i>Arteriosclerosis, Thrombosis, and Vascular Biology</i> , 2022, , 101161ATVBAHA122317395.	2.4	4
6	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. <i>Frontiers in Cardiovascular Medicine</i> , 2022, 9, .	2.4	2
7	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. <i>International Journal of Molecular Sciences</i> , 2022, 23, 6749.	4.1	4
8	Airflow Obstruction in Adults with Williams Syndrome and Mice with Elastin Insufficiency. <i>Diagnostics</i> , 2022, 12, 1438.	2.6	1
9	Emerging mechanisms of elastin transcriptional regulation. <i>American Journal of Physiology - Cell Physiology</i> , 2022, 323, C666-C677.	4.6	4
10	Identifying environmental risk factors and <sc>gene&quot;environment</sc> interactions in holoprosencephaly. <i>Birth Defects Research</i> , 2021, 113, 63-76.	1.5	14
11	A dyadic approach to the delineation of diagnostic entities in clinical genomics. <i>American Journal of Human Genetics</i> , 2021, 108, 8-15.	6.2	71
12	X-ray microtomosynthesis of unstained pathology tissue samples. <i>Journal of Microscopy</i> , 2021, 283, 9-20.	1.8	5
13	Developmental vascular malformations in EPAS1 gain-of-function syndrome. <i>JCI Insight</i> , 2021, 6, .	5.0	14
14	Inhibition of NOX1 Mitigates Blood Pressure Increases in Elastin Insufficiency. <i>Function</i> , 2021, 2, zqab015.	2.3	10
15	X-linked creatine transporter deficiency results in prolonged QTc and increased sudden death risk in humans and disease model. <i>Genetics in Medicine</i> , 2021, 23, 1864-1872.	2.4	8
16	Williams syndrome. <i>Nature Reviews Disease Primers</i> , 2021, 7, 42.	30.5	103
17	Cardiac pathologies in mouse loss of imprinting models are due to misexpression of H19 long noncoding RNA. <i>ELife</i> , 2021, 10, .	6.0	8
18	Response to Hamosh et al.. <i>American Journal of Human Genetics</i> , 2021, 108, 1809-1810.	6.2	0

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19	Loss of Angiotensin II Type 2 Receptor Improves Blood Pressure in Elastin Insufficiency. <i>Frontiers in Cardiovascular Medicine</i> , 2021, 8, 782138.	2.4	6
20	Mild macrocytosis in Williams-Beuren syndrome. <i>European Journal of Medical Genetics</i> , 2020, 63, 103740.	1.3	6
21	Neuraxial dysraphism in EPAS1-associated syndrome due to improper mesenchymal transition. <i>Neurology: Genetics</i> , 2020, 6, e414.	1.9	5
22	Whole exome sequencing in patients with Williams-Beuren syndrome followed by disease modeling in mice points to four novel pathways that may modify stenosis risk. <i>Human Molecular Genetics</i> , 2020, 29, 2035-2050.	2.9	15
23	Prenatal exposure to pesticides and risk for holoprosencephaly: a case-control study. <i>Environmental Health</i> , 2020, 19, 65.	4.0	20
24	Social, neurodevelopmental, endocrine, and head size differences associated with atypical deletions in Williams-Beuren syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1008-1020.	1.2	12
25	Vascular elastic fiber heterogeneity in health and disease. <i>Current Opinion in Hematology</i> , 2020, 27, 190-196.	2.5	14
26	Impaired angiogenesis and extracellular matrix metabolism in autosomal-dominant hyper-IgE syndrome. <i>Journal of Clinical Investigation</i> , 2020, 130, 4167-4181.	8.2	13
27	Vascular Casting of Adult and Early Postnatal Mouse Lungs for Micro-CT Imaging. <i>Journal of Visualized Experiments</i> , 2020, , .	0.3	6
28	Elastic fiber ultrastructure and assembly. <i>Matrix Biology</i> , 2019, 84, 31-40.	3.6	52
29	Special Therapy and Psychosocial Needs Identified in a Multidisciplinary Cancer Predisposition Syndrome Clinic. <i>Journal of Pediatric Hematology/Oncology</i> , 2019, 41, 133-136.	0.6	1
30	Lamin B receptor-related disorder is associated with a spectrum of skeletal dysplasia phenotypes. <i>Bone</i> , 2019, 120, 354-363.	2.9	11
31	Elastin-driven genetic diseases. <i>Matrix Biology</i> , 2018, 71-72, 144-160.	3.6	69
32	Williams-Beuren syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1128-1136.	1.2	55
33	Minoxidil improves vascular compliance, restores cerebral blood flow, and alters extracellular matrix gene expression in a model of chronic vascular stiffness. <i>American Journal of Physiology - Heart and Circulatory Physiology</i> , 2018, 315, H18-H32.	3.2	44
34	Exome sequencing of 85 Williams-Beuren syndrome cases rules out coding variation as a major contributor to remaining variance in social behavior. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2018, 6, 749-765.	1.2	9
35	Cover Image, Volume 176A, Number 5, May 2018. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, .	1.2	0
36	The Exome Clinic and the role of medical genetics expertise in the interpretation of exome sequencing results. <i>Genetics in Medicine</i> , 2017, 19, 1040-1048.	2.4	85

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37	GENERAL ANESTHESIA RISK IN WILLIAMS BEUREN SYNDROME PATIENTS IN THE CURRENT ERA. <i>Journal of the American College of Cardiology</i> , 2017, 69, 598.	2.8	1
38	Effects of Obesity and Hypertension on Pulse Wave Velocity in Children. <i>Journal of Clinical Hypertension</i> , 2017, 19, 221-226.	2.0	52
39	Hypercalcemia in Patients with Williams-Beuren Syndrome. <i>Journal of Pediatrics</i> , 2016, 178, 254-260.e4.	1.8	45
40	KATP channel gain-of-function leads to increased myocardial L-type Ca <sup>2+</sup> current and contractility in Cantu syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, 6773-6778.	7.1	29
41	Pathology of the Elastic Matrix. , 2016, , 31-80.		1
42	Chronic antihypertensive treatment improves pulse pressure but not large artery mechanics in a mouse model of congenital vascular stiffness. <i>American Journal of Physiology - Heart and Circulatory Physiology</i> , 2015, 309, H1008-H1016.	3.2	21
43	Elastin Insufficiency Predisposes Mice to Impaired Glucose Metabolism. <i>Journal of Molecular and Genetic Medicine: an International Journal of Biomedical Research</i> , 2014, 08, .	0.1	8
44	Biomechanical Properties of the Skin in Cutis Laxa. <i>Journal of Investigative Dermatology</i> , 2014, 134, 2836-2838.	0.7	11
45	Clinical utility gene card for: Williams's Beuren Syndrome [7q11.23]. <i>European Journal of Human Genetics</i> , 2014, 22, 1153-1153.	2.8	9
46	Skin findings in Williams syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 2217-2225.	1.2	22
47	Williams Syndrome Predisposes to Vascular Stiffness Modified by Antihypertensive Use and Copy Number Changes in <i>NCF1</i> . <i>Hypertension</i> , 2014, 63, 74-79.	2.7	69
48	Copy number analysis of <i>NIPBL</i> in a cohort of 510 patients reveals rare copy number variants and a mosaic deletion. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2014, 2, 115-123.	1.2	12
49	Altered reactivity of resistance vasculature contributes to hypertension in elastin insufficiency. <i>American Journal of Physiology - Heart and Circulatory Physiology</i> , 2014, 306, H654-H666.	3.2	32
50	Whole-genome copy number variation analysis in anophthalmia and microphthalmia. <i>Clinical Genetics</i> , 2013, 84, 473-481.	2.0	43
51	Alternative Splicing and Tissue-specific Elastin Misassembly Act as Biological Modifiers of Human Elastin Gene Frameshift Mutations Associated with Dominant Cutis Laxa. <i>Journal of Biological Chemistry</i> , 2012, 287, 22055-22067.	3.4	28
52	Delineation of a deletion region critical for corpus callosal abnormalities in chromosome 1q43-q44. <i>European Journal of Human Genetics</i> , 2012, 20, 176-179.	2.8	42
53	BMP4 loss-of-function mutations in developmental eye disorders including SHORT syndrome. <i>Human Genetics</i> , 2011, 130, 495-504.	3.8	92
54	Genetic Modifiers of Cardiovascular Phenotype Caused by Elastin Haploinsufficiency Act by Extrinsic Noncomplementation. <i>Journal of Biological Chemistry</i> , 2011, 286, 44926-44936.	3.4	34

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55	Elastin. , 2011, , 267-301.		10
56	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
57	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
58	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
59	Time-lapse imaging of extracellular matrix assembly. FASEB Journal, 2008, 22, 101.1.	0.5	0
60	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
61	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
62	Elastic fiber formation: A dynamic view of extracellular matrix assembly using timer reporters. Journal of Cellular Physiology, 2006, 207, 87-96.	4.1	170
63	Elastic fiber macro-assembly is a hierarchical, cell motion-mediated process. Journal of Cellular Physiology, 2006, 207, 97-106.	4.1	93
64	Tropoelastin Interacts with Cell-surface Glycosaminoglycans via Its COOH-terminal Domain. Journal of Biological Chemistry, 2005, 280, 40939-40947.	3.4	138
65	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
66	Domains in Tropoelastin That Mediate Elastin Depositionin Vitro and in Vivo. Journal of Biological Chemistry, 2003, 278, 18491-18498.	3.4	122
67	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