Kim L Mcbride

List of Publications by Year
in descending order

103	4,021	147726 31	128225 60
papers	citations	h-index	g-index

109	109	109	6464
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Cerebral organoids containing an AUTS2 missense variant model microcephaly. Brain, 2023, 146, 387-404.	3.7	11
2	Longâ€read whole genome sequencing reveals HOXD13 alterations in synpolydactyly. Human Mutation, 2022, 43, 189-199.	1.1	7
3	Biallelic SEPSECS variants in two siblings with pontocerebellar hypoplasia type 2D underscore the relevance of splice-disrupting synonymous variants in disease.. Journal of Physical Education and Sports Management, 2022, , mcs.a006165.	0.5	0
4	Use of machine learning to classify high-risk variants of uncertain significance in lamin A/C cardiac disease. Heart Rhythm, 2022, 19, 676-685.	0.3	3
5	Exome sequencing in multiplex families with left-sided cardiac defects has high yield for disease gene discovery. PLoS Genetics, 2022, 18, e1010236.	1.5	8
6	A Decade's Experience in Pediatric Chromosomal Microarray Reveals Distinct Characteristics Across Ordering Specialties. Journal of Molecular Diagnostics, 2022, 24, 1031-1040.	1.2	3
7	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	5.8	21
8	Update in the Mucopolysaccharidoses. Seminars in Pediatric Neurology, 2021, 37, 100874.	1.0	27
9	Direct Reprogramming of Human Fibroblasts into Myoblasts to Investigate Therapies for Neuromuscular Disorders. Journal of Visualized Experiments, 2021, , .	0.2	0
10	Longitudinal MRI brain volume changes over one year in children with mucopolysaccharidosis types IIIA and IIIB. Molecular Genetics and Metabolism, 2021, 133, 193-200.	0.5	2
11	A qualitative assessment of parental experiences with falseâ€positive newborn screening for Krabbe disease. Journal of Genetic Counseling, 2021, , .	0.9	0
12	Common deletion variants causing protocadherin-Î± deficiency contribute to the complex genetics of BAV and left-sided congenital heart disease. Human Genetics and Genomics Advances, 2021, 2, 100037.	1.0	7
13	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16
14	A Multi-Omics Approach Using a Mouse Model of Cardiac Malformations for Prioritization of Human Congenital Heart Disease Contributing Genes. Frontiers in Cardiovascular Medicine, 2021, 8, 683074.	1.1	2
15	Hypomorphic alleles pose challenges in rare disease genomic variant interpretation. Clinical Genetics, 2021, 100, 775-776.	1.0	4
16	Decoding Genetics of Congenital Heart Disease Using Patient-Derived Induced Pluripotent Stem Cells (iPSCs). Frontiers in Cell and Developmental Biology, 2021, 9, 630069.	1.8	17
17	A pediatric perspective on genomics and prevention in the twenty-first century. Pediatric Research, 2020, 87, 338-344.	1.1	3
18	Treatment of mucopolysaccharidosis type II (Hunter syndrome): a Delphi derived practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1735-1742.	1.1	8

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19	Novel frameshift variant in MYL2 reveals molecular differences between dominant and recessive forms of hypertrophic cardiomyopathy. PLoS Genetics, 2020, 16, e1008639.	1.5	16
20	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
21	Expansion of B4GALT7 linkeropathy phenotype to include perinatal lethal skeletal dysplasia. European Journal of Human Genetics, 2019, 27, 1569-1577.	1.4	10
22	Reclassification of Variants of Uncertain Significance in Children with Inherited Arrhythmia Syndromes is Predicted by Clinical Factors. Pediatric Cardiology, 2019, 40, 1679-1687.	0.6	24
23	De novo loss-of-function variants in NSD2 (WHSC1) associate with a subset of Wolfâ€“Hirschhorn syndrome. Journal of Physical Education and Sports Management, 2019, 5, a004044.	0.5	20
24	Evaluation of biomarkers for Sanfilippo syndrome. Molecular Genetics and Metabolism, 2019, 128, 68-74.	0.5	13
25	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	3.6	23
26	Novel in-frame FLNB deletion causes Larsen syndrome in a three-generation pedigree. Journal of Physical Education and Sports Management, 2019, 5, a004176.	0.5	1
27	Phenylalanine and tyrosine measurements across gestation by tandem mass spectrometer on dried blood spot cards from normal pregnant women. Genetics in Medicine, 2019, 21, 1821-1826.	1.1	7
28	Genetic Evaluation of Cardiomyopathyâ€”A Heart Failure Society of America Practice Guideline. Journal of Cardiac Failure, 2018, 24, 281-302.	0.7	280
29	Familial co-occurrence of congenital heart defects follows distinct patterns. European Heart Journal, 2018, 39, 1015-1022.	1.0	32
30	In-frame de novo mutation in BICD2 in two patients with muscular atrophy and arthrogryposis. Journal of Physical Education and Sports Management, 2018, 4, a003160.	0.5	14
31	Natural history of echocardiographic abnormalities in mucopolysaccharidosis III. Molecular Genetics and Metabolism, 2018, 124, 131-134.	0.5	11
32	Genetic evaluation of cardiomyopathy: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 899-909.	1.1	172
33	General anesthesia with a native airway for patients with mucopolysaccharidosis type <scp>III</scp>. Paediatric Anaesthesia, 2017, 27, 370-376.	0.6	9
34	Systemic gene transfer of scAAV9.U1a.hSGSH for MPS IIIA: tolerability and preliminary evidence for a biochemical effect. Molecular Genetics and Metabolism, 2017, 120, S47.	0.5	0
35	Differential Prevalence of Antibodies Against Adeno-Associated Virus in Healthy Children and Patients with Mucopolysaccharidosis III: Perspective for AAV-Mediated Gene Therapy. Human Gene Therapy Clinical Development, 2017, 28, 187-196.	3.2	31
36	A cohort study of multiple families with FBN1 p.R650C variant, ectopia lentis, and low but not absent risk for aortopathy. American Journal of Medical Genetics, Part A, 2017, 173, 2995-3002.	0.7	4

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37	Abnormal Longitudinal Growth of the Aorta in Children with Familial Bicuspid Aortic Valve. Pediatric Cardiology, 2017, 38, 1709-1715.	0.6	4
38	Modifying Mendel Redux. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	0
39	Assessment of large copy number variants in patients with apparently isolated congenital leftâ€sided cardiac lesions reveals clinically relevant genomic events. American Journal of Medical Genetics, Part A, 2017, 173, 2176-2188.	0.7	17
40	Differential prevalence of antibodies against adeno-associated virus in healthy children and patients with mucopolysaccharidosis III: perspective for AAV-mediated gene therapy. Human Gene Therapy Clinical Development, 2017, , .	3.2	0
41	A prospective one-year natural history study of mucopolysaccharidosis types IIIA and IIIB: Implications for clinical trial design. Molecular Genetics and Metabolism, 2016, 119, 239-248.	0.5	41
42	Utilization of Whole Exome Sequencing to Identify Causative Mutations in Familial Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2016, 9, 320-329.	5.1	71
43	A genome-wide association study of congenital cardiovascular left-sided lesions shows association with a locus on chromosome 20. Human Molecular Genetics, 2016, 25, 2331-2341.	1.4	31
44	Magnetic resonance imaging in neonatal citrullinemia. Journal of Pediatric Neuroradiology, 2015, 02, 169-173.	0.1	0
45	78. An IND-Enabling GLP-Toxicology and Biodistribution Study Assessing Systemic rAAV9-hNAGLU Gene Delivery for Treating MPS IIIB: Genotype- and Sex-Specific Dose-Limiting Acute Liver Toxicity in Male Wild Type C57BL/6 Mice. Molecular Therapy, 2015, 23, S34.	3.7	0
46	A GLP-Compliant Toxicology and Biodistribution Study: Systemic Delivery of an rAAV9 Vector for the Treatment of Mucopolysaccharidosis IIIB. Human Gene Therapy Clinical Development, 2015, 26, 228-242.	3.2	19
47	Rationale for the Cytogenomics of Cardiovascular Malformations Consortium: A Phenotype Intensive Registry Based Approach. Journal of Cardiovascular Development and Disease, 2015, 2, 76-92.	0.8	7
48	Lifetime Prevalence of Sexual Intercourse and Contraception Use at Last Sex Among Adolescents and Young Adults With Congenital Heart Disease. Journal of Adolescent Health, 2015, 56, 396-401.	1.2	22
49	Novel familial dilated cardiomyopathy mutation in <scp>MYL</scp>2 affects the structure and function of myosin regulatory light chain. FEBS Journal, 2015, 282, 2379-2393.	2.2	42
50	Rare GATA5 sequence variants identified in individuals with bicuspid aortic valve. Pediatric Research, 2014, 76, 211-216.	1.1	74
51	Understanding of informed consent by parents of children enrolled in a genetic biobank. Genetics in Medicine, 2014, 16, 141-148.	1.1	43
52	Neurodevelopmental disorders among individuals with duplication of 4p13 to 4p12 containing a GABAA receptor subunit gene cluster. European Journal of Human Genetics, 2014, 22, 105-109.	1.4	20
53	Genetic knowledge and attitudes of parents of children with congenital heart defects. American Journal of Medical Genetics, Part A, 2014, 164, 3069-3075.	0.7	18
54	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	13.9	101

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55	Use of a targeted, combinatorial next-generation sequencing approach for the study of bicuspid aortic valve. BMC Medical Genomics, 2014, 7, 56.	0.7	50
56	Feasibility and Safety of Systemic rAAV9-hNAGLU Delivery for Treating Mucopolysaccharidosis IIIB: Toxicology, Biodistribution, and Immunological Assessments in Primates. Human Gene Therapy Clinical Development, 2014, 25, 72-84.	3.2	79
57	Severe hypertrophic cardiomyopathy in Noonan syndromeâ€"consider sequencing genes encoding sarcomeric proteins. American Journal of Medical Genetics, Part A, 2013, 161, 230-231.	0.7	2
58	MCTP2 is a dosage-sensitive gene required for cardiac outflow tract development. Human Molecular Genetics, 2013, 22, 4339-4348.	1.4	40
59	Successful Medical Therapy for Hypophosphatemic Rickets due to Mitochondrial Complex I Deficiency Induced de Toni-DebrÃ©-Fanconi Syndrome. Case Reports in Pediatrics, 2013, 2013, 1-5.	0.2	4
60	Genetic Abnormalities inFOXP1Are Associated with Congenital Heart Defects. Human Mutation, 2013, 34, 1226-1230.	1.1	39
61	Modifying Mendel. Circulation: Cardiovascular Genetics, 2012, 5, 274-276.	5.1	7
62	Cardiac teratogenicity in mouse maternal phenylketonuria: Defining phenotype parameters and genetic background influences. Molecular Genetics and Metabolism, 2012, 107, 650-658.	0.5	8
63	NOTCH1 missense alleles associated with left ventricular outflow tract defects exhibit impaired receptor processing and defective EMT. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2011, 1812, 121-129.	1.8	24
64	Incidence and timing of infusion-related reactions in patients with mucopolysaccharidosis type II (Hunter syndrome) on idursulfase therapy in the real-world setting: A perspective from the Hunter Outcome Survey (HOS). Molecular Genetics and Metabolism, 2011, 103, 113-120.	0.5	55
65	Contactin 4 as an autism susceptibility locus. Autism Research, 2011, 4, 189-199.	2.1	57
66	Pediatric Subâ€specialist Controversies in the Treatment of Congenital Heart Disease in Trisomy 13 or 18. Journal of Genetic Counseling, 2011, 20, 495-509.	0.9	33
67	Rebuttal to the invited comment of Opitz and Carey. American Journal of Medical Genetics, Part A, 2011, 155, 2037-2038.	0.7	1
68	Coronary artery disease in a Werner syndromeâ€like form of progeria characterized by low levels of progerin, a splice variant of lamin A. American Journal of Medical Genetics, Part A, 2011, 155, 3002-3006.	0.7	55
69	Association of common variants in ERBB4 with congenital left ventricular outflow tract obstruction defects. Birth Defects Research Part A: Clinical and Molecular Teratology, 2011, 91, 162-168.	1.6	43
70	Heredity of bicuspid aortic valve: is family screening indicated?. Heart, 2011, 97, 1193-1195.	1.2	19
71	Novel X-linked glomerulopathy is associated with a COL4A5 missense mutation in a non-collagenous interruption. Kidney International, 2011, 79, 120-127.	2.6	16
72	Importance of surgical history in diagnosing mucopolysaccharidosis type II (Hunter syndrome): Data from the Hunter Outcome Survey. Genetics in Medicine, 2010, 12, 816-822.	1.1	63

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73	Identification of a Recurrent Microdeletion at 17q23.1q23.2 Flanked by Segmental Duplications Associated with Heart Defects and Limb Abnormalities. American Journal of Human Genetics, 2010, 86, 454-461.	2.6	85
74	Confirmation study of PTEN mutations among individuals with autism or developmental delays/mental retardation and macrocephaly. Autism Research, 2010, 3, 137-141.	2.1	218
75	Parental Knowledge and Attitudes Toward Hypertrophic Cardiomyopathy Genetic Testing. Pediatric Cardiology, 2010, 31, 195-202.	0.6	16
76	Impact of Mendelian inheritance in cardiovascular disease. Annals of the New York Academy of Sciences, 2010, 1214, 122-137.	1.8	13
77	Significant contributions of the extraembryonic membranes and maternal genotype to the placental pathology in heterozygous Nsdhl deficient female embryos. Human Molecular Genetics, 2010, 19, 364-373.	1.4	3
78	Refinement of the Region for Split Hand/Foot Malformation 5 on 2q31.1. Molecular Syndromology, 2010, 1, 262-271.	0.3	15
79	Early orthotopic liver transplantation in urea cycle defects: Follow up of a developmental outcome study. Molecular Genetics and Metabolism, 2010, 100, S84-S87.	0.5	53
80	Home treatment with intravenous enzyme replacement therapy with idursulfase for mucopolysaccharidosis type II â€" data from the Hunter Outcome Survey. Molecular Genetics and Metabolism, 2010, 101, 123-129.	0.5	26
81	Acute Dilated Cardiomyopathy in a Patient with Deficiency of Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase. Pediatric Cardiology, 2009, 30, 523-526.	0.6	10
82	Linkage analysis of left ventricular outflow tract malformations (aortic valve stenosis, coarctation) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 811-819.	1.4	81
83	The prevalence of PTEN mutations in a clinical pediatric cohort with autism spectrum disorders, developmental delay, and macrocephaly. Genetics in Medicine, 2009, 11, 111-117.	1.1	251
84	NOTCH1 mutations in individuals with left ventricular outflow tract malformations reduce ligand-induced signaling. Human Molecular Genetics, 2008, 17, 2886-2893.	1.4	182
85	Idursulfase: enzyme replacement therapy for mucopolysaccharidosis Type II (Hunter syndrome). Expert Review of Endocrinology and Metabolism, 2007, 2, 19-26.	1.2	1
86	Heritability of plasma amino acid levels in different nutritional states. Molecular Genetics and Metabolism, 2007, 90, 217-220.	0.5	7
87	Gene Copy-Number Variation and Associated Polymorphisms of Complement Component C4 in Human Systemic Lupus Erythematosus (SLE): Low Copy Number Is a Risk Factor for and High Copy Number Is a Protective Factor against SLE Susceptibility in European Americans. American Journal of Human Genetics, 2007, 80, 1037-1054.	2.6	411
88	Genetic testing in autism: how much is enough?. Genetics in Medicine, 2007, 9, 268-274.	1.1	97
89	Renal anomalies in family members of infants with bilateral renal agenesis/adysplasia. Pediatric Nephrology, 2007, 22, 52-56.	0.9	24
90	Genome-wide linkage scan for spontaneous DZ twinning. European Journal of Human Genetics, 2006, 14, 117-122.	1.4	16

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91	Epidemiology of noncomplex left ventricular outflow tract obstruction malformations (aortic valve) Tj ETQq1 1 0.784314 rgBT /Overlock Research Part A: Clinical and Molecular Teratology, 2005, 73, 555-561.	1.6	64
92	Inheritance analysis of congenital left ventricular outflow tract obstruction malformations: Segregation, multiplex relative risk, and heritability. American Journal of Medical Genetics, Part A, 2005, 134A, 180-186.	0.7	198
93	Cardiovascular genetics clinics. American Journal of Medical Genetics, Part A, 2005, 135A, 229-229.	0.7	1
94	Developmental Outcomes With Early Orthotopic Liver Transplantation for Infants With Neonatal-Onset Urea Cycle Defects and a Female Patient With Late-Onset Ornithine Transcarbamylase Deficiency. Pediatrics, 2004, 114, e523-e526.	1.0	65
95	Echocardiographic Evaluation of Asymptomatic Parental and Sibling Cardiovascular Anomalies Associated With Congenital Left Ventricular Outflow Tract Lesions. Pediatrics, 2004, 114, 691-696.	1.0	102
96	A family-based association study of congenital left-sided heart malformations and 5,10 methylenetetrahydrofolate reductase. Birth Defects Research Part A: Clinical and Molecular Teratology, 2004, 70, 825-830.	1.6	32
97	Toward a genetic etiology of CHARGE syndrome: I. A systematic scan for submicroscopic deletions. American Journal of Medical Genetics Part A, 2003, 118A, 260-266.	2.4	23
98	Case 27-2002: Late-Onset Infantile Neuronal Ceroid Lipofuscinosis. New England Journal of Medicine, 2003, 348, 2159-2159.	13.9	1
99	Aortoesophageal fistula in a 13-yr-old girl: Complication after nasogastric tube placement in the setting of right-sided aortic arch. Pediatric Critical Care Medicine, 2002, 3, 378-380.	0.2	8
100	Hb DARTMOUTH [Î±66(E15)Leu â†' Pro (Î±2) (CTG â†' CCG)]: A NOVEL Î±2-GLOBIN GENE MUTATION ASSOCIATED WITH SEVERE NEONATAL ANEMIA WHEN INHERITED IN TRANS WITH SOUTHEAST ASIAN Î±-THALASSEMIA-1. Hemoglobin, 2001, 25, 375-382.	0.4	28
101	Severe 6-Thioguanine-induced Marrow Aplasia in a Child With Acute Lymphoblastic Leukemia and Inherited Thiopurine Methyltransferase Deficiency. The American Journal of Pediatric Hematology/oncology, 2000, 22, 441-445.	1.3	42
102	Lyme Disease and Pseudotumor: In reply. Mayo Clinic Proceedings, 2000, 75, 315.	1.4	0
103	14-Year-Old Boy With Blurred Vision and Diplopia. Mayo Clinic Proceedings, 1999, 74, 1157-1160.	1.4	2