Kim L Mcbride

List of Publications by Year
in descending order

103 papers	4,021 citations	168829 31 h-index	145109 60 g-index
109 all docs	109 docs citations	109 times ranked	6966 citing authors

#	Article	IF	Citations
1	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
2	Genetic Evaluation of Cardiomyopathyâ€"A Heart Failure Society of America Practice Guideline. Journal of Cardiac Failure, 2018, 24, 281-302.	0.7	280
3	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
4	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
5	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
6	NOTCH1 mutations in individuals with left ventricular outflow tract malformations reduce ligand-induced signaling. Human Molecular Genetics, 2008, 17, 2886-2893.	1.4	182
7	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
8	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
9	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
10	Genetic testing in autism: how much is enough?. Genetics in Medicine, 2007, 9, 268-274.	1.1	97
11	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
12	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
13	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
14	Rare GATA5 sequence variants identified in individuals with bicuspid aortic valve. Pediatric Research, 2014, 76, 211-216.	1.1	74
15	Utilization of Whole Exome Sequencing to Identify Causative Mutations in Familial Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2016, 9, 320-329.	5.1	71
16	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
17	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
18	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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19	Contactin 4 as an autism susceptibility locus. Autism Research, 2011, 4, 189-199.	2.1	57
20	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
21	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
22	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
23	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
24	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
25	Understanding of informed consent by parents of children enrolled in a genetic biobank. Genetics in Medicine, 2014, 16, 141-148.	1.1	43
26	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
27	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
28	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
29	MCTP2 is a dosage-sensitive gene required for cardiac outflow tract development. Human Molecular Genetics, 2013, 22, 4339-4348.	1.4	40
30	Genetic Abnormalities inFOXP1Are Associated with Congenital Heart Defects. Human Mutation, 2013, 34, 1226-1230.	1.1	39
31	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
32	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
33	Familial co-occurrence of congenital heart defects follows distinct patterns. European Heart Journal, 2018, 39, 1015-1022.	1.0	32
34	Lessons learned from 40 novel PIGA patients and a review of the literature. Epilepsia, 2020, 61, 1142-1155.	2.6	32
35	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
36	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

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37	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
38	Update in the Mucopolysaccharidoses. Seminars in Pediatric Neurology, 2021, 37, 100874.	1.0	27
39	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
40	Renal anomalies in family members of infants with bilateral renal agenesis/adysplasia. Pediatric Nephrology, 2007, 22, 52-56.	0.9	24
41	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
42	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
43	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
44	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
45	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
46	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	5.8	21
47	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
48	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
49	Heredity of bicuspid aortic valve: is family screening indicated?. Heart, 2011, 97, 1193-1195.	1.2	19
50	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
51	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
52	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
53	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
54	Genome-wide linkage scan for spontaneous DZ twinning. European Journal of Human Genetics, 2006, 14, 117-122.	1.4	16

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55	Parental Knowledge and Attitudes Toward Hypertrophic Cardiomyopathy Genetic Testing. Pediatric Cardiology, 2010, 31, 195-202.	0.6	16
56	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
57	Novel frameshift variant in MYL2 reveals molecular differences between dominant and recessive forms of hypertrophic cardiomyopathy. PLoS Genetics, 2020, 16, e1008639.	1.5	16
58	Delineating the molecular and phenotypic spectrum of the SETD1B-related syndrome. Genetics in Medicine, 2021, 23, 2122-2137.	1.1	16
59	Refinement of the Region for Split Hand/Foot Malformation 5 on 2q31.1. Molecular Syndromology, 2010, 1, 262-271.	0.3	15
60	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
61	Impact of Mendelian inheritance in cardiovascular disease. Annals of the New York Academy of Sciences, 2010, 1214, 122-137.	1.8	13
62	Evaluation of biomarkers for Sanfilippo syndrome. Molecular Genetics and Metabolism, 2019, 128, 68-74.	0.5	13
63	Natural history of echocardiographic abnormalities in mucopolysaccharidosis III. Molecular Genetics and Metabolism, 2018, 124, 131-134.	0.5	11
64	Cerebral organoids containing an AUTS2 missense variant model microcephaly. Brain, 2023, 146, 387-404.	3.7	11
65	Acute Dilated Cardiomyopathy in a Patient with Deficiency of Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase. Pediatric Cardiology, 2009, 30, 523-526.	0.6	10
66	Expansion of B4GALT7 linkeropathy phenotype to include perinatal lethal skeletal dysplasia. European Journal of Human Genetics, 2019, 27, 1569-1577.	1.4	10
67	General anesthesia with a native airway for patients with mucopolysaccharidosis type <scp>III</scp>. Paediatric Anaesthesia, 2017, 27, 370-376.	0.6	9
68	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
69	Cardiac teratogenicity in mouse maternal phenylketonuria: Defining phenotype parameters and genetic background influences. Molecular Genetics and Metabolism, 2012, 107, 650-658.	0.5	8
70	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
71	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
72	Heritability of plasma amino acid levels in different nutritional states. Molecular Genetics and Metabolism, 2007, 90, 217-220.	0.5	7

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73	Modifying Mendel. Circulation: Cardiovascular Genetics, 2012, 5, 274-276.	5.1	7
74	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
75	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
76	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
77	Longâ€read whole genome sequencing reveals HOXD13 alterations in synpolydactyly. Human Mutation, 2022, 43, 189-199.	1.1	7
78	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
79	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
80	Abnormal Longitudinal Growth of the Aorta in Children with Familial Bicuspid Aortic Valve. Pediatric Cardiology, 2017, 38, 1709-1715.	0.6	4
81	Hypomorphic alleles pose challenges in rare disease genomic variant interpretation. Clinical Genetics, 2021, 100, 775-776.	1.0	4
82	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
83	A pediatric perspective on genomics and prevention in the twenty-first century. Pediatric Research, 2020, 87, 338-344.	1.1	3
84	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
85	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
86	14-Year-Old Boy With Blurred Vision and Diplopia. Mayo Clinic Proceedings, 1999, 74, 1157-1160.	1.4	2
87	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
88	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
89	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
90	Case 27-2002: Late-Onset Infantile Neuronal Ceroid Lipofuscinosis. New England Journal of Medicine, 2003, 348, 2159-2159.	13.9	1

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91	Cardiovascular genetics clinics. American Journal of Medical Genetics, Part A, 2005, 135A, 229-229.	0.7	1
92	Idursulfase: enzyme replacement therapy for mucopolysaccharidosis Type II (Hunter syndrome). Expert Review of Endocrinology and Metabolism, 2007, 2, 19-26.	1.2	1
93	Rebuttal to the invited comment of Opitz and Carey. American Journal of Medical Genetics, Part A, 2011, 155, 2037-2038.	0.7	1
94	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
95	Lyme Disease and Pseudotumor: In reply. Mayo Clinic Proceedings, 2000, 75, 315.	1.4	0
96	Magnetic resonance imaging in neonatal citrullinemia. Journal of Pediatric Neuroradiology, 2015, 02, 169-173.	0.1	0
97	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
98	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
99	Modifying Mendel Redux. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	0
100	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
101	Direct Reprogramming of Human Fibroblasts into Myoblasts to Investigate Therapies for Neuromuscular Disorders. Journal of Visualized Experiments, 2021, , .	0.2	0
102	A qualitative assessment of parental experiences with falseâ€positive newborn screening for Krabbe disease. Journal of Genetic Counseling, 2021, , .	0.9	0
103	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