William R Wilcox

List of Publications by Year
in descending order

42 papers	3,716 citations	304743 22 h-index	302126 39 g-index
42 all docs	42 docs citations	42 times ranked	3819 citing authors

#	Article	IF	Citations
1	Pharmacokinetics and Exposureâ€“Response of Vosoritide in Children with Achondroplasia. Clinical Pharmacokinetics, 2022, 61, 263-280.	3.5	15
2	Safe and persistent growth-promoting effects of vosoritide in children with achondroplasia: 2-year results from an open-label, phase 3 extension study. Genetics in Medicine, 2021, 23, 2443-2447.	2.4	36
3	Health care practitioners' experience-based opinions on providing care after a positive newborn screen for Pompe disease. Molecular Genetics and Metabolism, 2021, 134, 20-28.	1.1	5
4	The emerging neurological spectrum of AARS2-associated disorders. Parkinsonism and Related Disorders, 2021, 93, 50-54.	2.2	3
5	MO035HISTORICAL CONTROL ANALYSIS DEMONSTRATES SUPERIOR REDUCTION OF PLASMA GLOBOTRIAOSYLCERAMIDE BY VENGLUSTAT COMPARED WITH PLACEBO OR AGALSIDASE BETA IN CLASSIC FABRY DISEASE PATIENTS. Nephrology Dialysis Transplantation, 2020, 35, .	0.7	2
6	P0062GLUCOSYLCERAMIDE SYNTHASE INHIBITION WITH VENGLUSTAT IN CLASSIC FABRY DISEASE PATIENTS LEADS TO PROGRESSIVE REDUCTION OF ENDOTHELIAL CELL GLOBOTRIAOSYLCERAMIDE INCLUSION VOLUME. Nephrology Dialysis Transplantation, 2020, 35, .	0.7	1
7	Improvement of gastrointestinal symptoms in a significant proportion of male patients with classic Fabry disease treated with agalsidase beta: A Fabry Registry analysis stratified by phenotype. Molecular Genetics and Metabolism Reports, 2020, 25, 100670.	1.1	6
8	Newborn Screening for X-Linked Adrenoleukodystrophy in Georgia: Experiences from a Pilot Study Screening of 51,081 Newborns. International Journal of Neonatal Screening, 2020, 6, 81.	3.2	19
9	Two-Tiered Newborn Screening with Post-Analytical Tools for Pompe Disease and Mucopolysaccharidosis Type I Results in Performance Improvement and Future Direction. International Journal of Neonatal Screening, 2020, 6, 2.	3.2	23
10	Use of a rare disease registry for establishing phenotypic classification of previously unassigned GLA variants: a consensus classification system by a multispecialty Fabry disease genotypeâ€“phenotype workgroup. Journal of Medical Genetics, 2020, 57, 542-551.	3.2	43
11	Fabry disease and COVID-19: international expert recommendations for management based on real-world experience. CKJ: Clinical Kidney Journal, 2020, 13, 913-925.	2.9	11
12	De novoGRINvariants in NMDA receptor M2 channel poreâ€forming loop are associated with neurological diseases. Human Mutation, 2019, 40, 2393-2413.	2.5	48
13	Natural History of Perinatal and Infantile Hypophosphatasia: A Retrospective Study. Journal of Pediatrics, 2019, 209, 116-124.e4.	1.8	39
14	Fabry disease revisited: Management and treatment recommendations for adult patients. Molecular Genetics and Metabolism, 2018, 123, 416-427.	1.1	391
15	SP004EFFECTS OF LONG-TERM MIGALASTAT TREATMENT ON RENAL FUNCTION BY BASELINE PROTEINURIA IN PATIENTS (PTS) WITH FABRY DISEASE. Nephrology Dialysis Transplantation, 2018, 33, i347-i348.	0.7	4
16	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. Journal of Medical Genetics, 2017, 54, 288-296.	3.2	262
17	Improvement of Fabry Disease-Related Gastrointestinal Symptoms in a Significant Proportion of Female Patients Treated with Agalsidase Beta: Data from the Fabry Registry. JIMD Reports, 2017, 38, 45-51.	1.5	18
18	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. Genetics in Medicine, 2017, 19, 430-438.	2.4	157

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19	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	27.0	390
20	Risk factors for severe clinical events in male and female patients with Fabry disease treated with agalsidase beta enzyme replacement therapy: Data from the Fabry Registry. Molecular Genetics and Metabolism, 2016, 119, 151-159.	1.1	35
21	Exome Sequencing Identified a Splice Site Mutation in FHL1 that Causes Uruguay Syndrome, an X-Linked Disorder With Skeletal Muscle Hypertrophy and Premature Cardiac Death. Circulation: Cardiovascular Genetics, 2016, 9, 130-135.	5.1	8
22	The management and treatment of children with Fabry disease: A United States-based perspective. Molecular Genetics and Metabolism, 2016, 117, 104-113.	1.1	85
23	Genetic evaluation and testing for hereditary forms of cancer in the era of next-generation sequencing. Cancer Biology and Medicine, 2016, 13, 55-67.	3.0	35
24	Changing paradigm of cancer therapy: precision medicine by next-generation sequencing. Cancer Biology and Medicine, 2016, 13, 12-8.	3.0	19
25	Response to Saul. Genetics in Medicine, 2015, 17, 761.	2.4	0
26	Antiproteinuric therapy and Fabry nephropathy: factors associated with preserved kidney function during agalsidase-beta therapy. Journal of Medical Genetics, 2015, 52, 860-866.	3.2	53
27	A second locus for schneckenbecken dysplasia identified by a mutation in the gene encoding inositol polyphosphate phosphataseâ€like 1 (INPPL1). American Journal of Medical Genetics, Part A, 2015, 167, 2470-2473.	1.2	9
28	Congenital Limb Deficiency Disorders. Clinics in Perinatology, 2015, 42, 281-300.	2.1	40
29	Ten-year outcome of enzyme replacement therapy with agalsidase beta in patients with Fabry disease. Journal of Medical Genetics, 2015, 52, 353-358.	3.2	266
30	Solving the molecular diagnostic testing conundrum for Mendelian disorders in the era of next-generation sequencing: single-gene, gene panel, or exome/genome sequencing. Genetics in Medicine, 2015, 17, 444-451.	2.4	288
31	FGFR3 mutation frequency in 324 cases from the International Skeletal Dysplasia Registry. Molecular Genetics & Genomic Medicine, 2014, 2, 497-503.	1.2	49
32	Fibroblast Growth Factor Receptor 3 Interacts with and Activates TGFÎ²-Activated Kinase 1 Tyrosine Phosphorylation and NFÎºB Signaling in Multiple Myeloma and Bladder Cancer. PLoS ONE, 2014, 9, e86470.	2.5	27
33	Anti-Î±-galactosidase A antibody response to agalsidase beta treatment: Data from the Fabry Registry. Molecular Genetics and Metabolism, 2012, 105, 443-449.	1.1	58
34	A novel skeletal disorder defines an intracellular role for FGFR2 during development. FASEB Journal, 2012, 26, 457.7.	0.5	0
35	Females with Fabry disease frequently have major organ involvement: Lessons from the Fabry Registry. Molecular Genetics and Metabolism, 2008, 93, 112-128.	1.1	442
36	Distinguishing Pacman dysplasia from mucolipidosis II: Comment on Saul et al. [2005]. American Journal of Medical Genetics, Part A, 2005, 135A, 333-333.	1.2	3

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37	Lysosomal storage disorders: the need for better pediatric recognition and comprehensive care. Journal of Pediatrics, 2004, 144, S3-S14.	1.8	94
38	Fabry Disease, an Under-Recognized Multisystemic Disorder: Expert Recommendations for Diagnosis, Management, and Enzyme Replacement Therapy. Annals of Internal Medicine, 2003, 138, 338.	3.9	619
39	Subtle radiographic findings of achondroplasia in patients with Crouzon syndrome with acanthosis nigricans due to an Ala391Glu substitution in FGFR3. American Journal of Medical Genetics Part A, 2001, 98, 75-91.	2.4	47
40	Small deletions in the type II collagen triple helix produce Kniest dysplasia. , 1999, 85, 105-112.		59
41	Small deletions in the type II collagen triple helix produce Kniest dysplasia. American Journal of Medical Genetics Part A, 1999, 85, 105-112.	2.4	1
42	Cumming Syndrome: report of two additional cases. Pediatric Radiology, 1998, 28, 798-801.	2.0	6