William R Wilcox

List of Publications by Year
in descending order

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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	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
2	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
3	Fabry disease revisited: Management and treatment recommendations for adult patients. Molecular Genetics and Metabolism, 2018, 123, 416-427.	1.1	391
4	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	27.0	390
5	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
6	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
7	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
8	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
9	Lysosomal storage disorders: the need for better pediatric recognition and comprehensive care. Journal of Pediatrics, 2004, 144, S3-S14.	1.8	94
10	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
11	Small deletions in the type II collagen triple helix produce Kniest dysplasia. , 1999, 85, 105-112.		59
12	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
13	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
14	FGFR3 mutation frequency in 324 cases from the International Skeletal Dysplasia Registry. Molecular Genetics & Genomic Medicine, 2014, 2, 497-503.	1.2	49
15	De novoGRINvariants in NMDA receptor M2 channel poreâ€forming loop are associated with neurological diseases. Human Mutation, 2019, 40, 2393-2413.	2.5	48
16	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
17	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
18	Congenital Limb Deficiency Disorders. Clinics in Perinatology, 2015, 42, 281-300.	2.1	40

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19	Natural History of Perinatal and Infantile Hypophosphatasia: A Retrospective Study. Journal of Pediatrics, 2019, 209, 116-124.e4.	1.8	39
20	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
21	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
22	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
23	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
24	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
25	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
26	Changing paradigm of cancer therapy: precision medicine by next-generation sequencing. Cancer Biology and Medicine, 2016, 13, 12-8.	3.0	19
27	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
28	Pharmacokinetics and Exposureâ€“Response of Vosoritide in Children with Achondroplasia. Clinical Pharmacokinetics, 2022, 61, 263-280.	3.5	15
29	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
30	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
31	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
32	Cumming Syndrome: report of two additional cases. Pediatric Radiology, 1998, 28, 798-801.	2.0	6
33	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
34	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
35	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
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	The emerging neurological spectrum of AARS2-associated disorders. Parkinsonism and Related Disorders, 2021, 93, 50-54.	2.2	3
38	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
39	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
40	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
41	Response to Saul. Genetics in Medicine, 2015, 17, 761.	2.4	0
42	A novel skeletal disorder defines an intracellular role for FGFR2 during development. FASEB Journal, 2012, 26, 457.7.	0.5	0