R Rodney Howell

List of Publications by Year
in descending order

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99	5,323	87888	82547
papers	citations	38 h-index	72 g-index

107	107	107	3410
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Pompe disease diagnosis and management guideline. Genetics in Medicine, 2006, 8, 267-288.	2.4	473
2	Newborn Screening: Toward a Uniform Screening Panel and Systemâ€"Executive Summary. Pediatrics, 2006, 117, S296-S307.	2.1	386
3	Strategies for Implementing Screening for Critical Congenital Heart Disease. Pediatrics, 2011, 128, e1259-e1267.	2.1	344
4	Executive Summary. Genetics in Medicine, 2006, 8, S1-S11.	2.4	310
5	A PARADOXICAL EFFECT OF ACTINOMYCIN D: THE MECHANISM OF REGULATION OF ENZYME SYNTHESIS BY HYDROCORTISONE. Proceedings of the National Academy of Sciences of the United States of America, 1964, 52, 1121-1129.	7.1	233
6	Pompe disease in infants and children. Journal of Pediatrics, 2004, 144, S35-S43.	1.8	226
7	Menkes disease: a biochemical abnormality in cultured human fibroblasts.. Proceedings of the National Academy of Sciences of the United States of America, 1976, 73, 604-606.	7.1	170
8	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. Journal of Neuromuscular Diseases, 2018, 5, 145-158.	2.6	148
9	Biochemistry of Uric Acid and Its Relation to Gout. New England Journal of Medicine, 1963, 268, 712-716.	27.0	144
10	Turnover of Ribosomal RNA in Rat Liver. Science, 1965, 149, 1093-1095.	12.6	132
11	Hepatic Adenomata With Type 1 Glycogen Storage Disease. JAMA - Journal of the American Medical Association, 1976, 236, 1481.	7.4	119
12	THE RENAL EXCRETION OF URIC ACID IN GOUT. Journal of Clinical Investigation, 1962, 41, 1094-1098.	8.2	101
13	Relationships between glycogen storage disease and tophaceous gout. American Journal of Medicine, 1967, 42, 58-66.	1.5	98
14	Infantile Metachromatic Leukodystrophy. New England Journal of Medicine, 1970, 282, 1336-1340.	27.0	90
15	Revised Recommendations for the Treatment of Infants Diagnosed with Spinal Muscular Atrophy Via Newborn Screening Who Have 4 Copies of SMN2. Journal of Neuromuscular Diseases, 2020, 7, 97-100.	2.6	89
16	Mammalian enzyme induction by hydrocortisone. Journal of Molecular Biology, 1964, 9, 100-108.	4.2	79
17	Committee report: Method for evaluating conditions nominated for population-based screening of newborns and children. Genetics in Medicine, 2010, 12, 153-159.	2.4	78
18	Xeroderma Pigmentosum: A Rapid Sensitive Method for Prenatal Diagnosis. Science, 1971, 174, 147-150.	12.6	75

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19	Excessive production of uric acid in type I glycogen storage disease. Journal of Pediatrics, 1968, 72, 488-496.	1.8	72
20	Committee report: Considerations and recommendations for national guidance regarding the retention and use of residual dried blood spot specimens after newborn screening. Genetics in Medicine, 2011, 13, 621-624.	2.4	72
21	The old and new concepts of acute gouty arthritis. Arthritis and Rheumatism, 1962, 5, 616-623.	6.7	71
22	Hepatic adenomata with type 1 glycogen storage disease. JAMA - Journal of the American Medical Association, 1976, 236, 1481-1484.	7.4	68
23	The interrelationship of glycogen storage disease and gout. Arthritis and Rheumatism, 1965, 8, 780-785.	6.7	66
24	Echocardiographic abnormalities in the mucopolysaccharide storage diseases. American Journal of Cardiology, 1988, 61, 170-176.	1.6	62
25	Neurotransmitter defects and treatment of disorders of hyperphenylalaninemia. Journal of Pediatrics, 1981, 98, 729-733.	1.8	60
26	Cardiac involvement in glycogen storage disease III: Morphologic and biochemical characterization with endomyocardial biopsy. American Journal of Cardiology, 1984, 53, 980-981.	1.6	59
27	PROPERTIES OF ACATALASIC CELLS GROWING IN VITRO. Journal of Experimental Medicine, 1962, 115, 313-328.	8.5	58
28	Long-term follow-up after diagnosis resulting from newborn screening: Statement of the US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. Genetics in Medicine, 2008, 10, 259-261.	2.4	56
29	The regulation of enzyme synthesis by steroid hormones: The role of translation. Journal of Cellular and Comparative Physiology, 1965, 66, 137-151.	1.8	53
30	Diagnostic challenges for Pompe disease: An under-recognized cause of floppy baby syndrome. Genetics in Medicine, 2006, 8, 289-296.	2.4	52
31	Newborn screening for spinal muscular atrophy: Anticipating an imminent need. Seminars in Perinatology, 2015, 39, 217-229.	2.5	51
32	Type IV glycogen storage disease: Branching enzyme deficiency in skin fibroblasts and possible heterozygote detection. Journal of Pediatrics, 1971, 78, 638-642.	1.8	47
33	CHARACTERIZATION OF RIBOSOMAL AGGREGATES ISOLATED FROM LIVER. Proceedings of the National Academy of Sciences of the United States of America, 1964, 52, 1241-1248.	7.1	45
34	Type IV Glycogen-storage Disease: Light-microscopic, Electron-microscopic, and Enzymatic Study. American Journal of Clinical Pathology, 1976, 66, 702-709.	0.7	43
35	X-ray diffraction studies of the tophaceous deposits in gout. Arthritis and Rheumatism, 1963, 6, 97-103.	6.7	39
36	Familial recurrent rhabdomyolysis due to carnitine palmityl transferase deficiency. American Journal of Medicine, 1979, 67, 167-171.	1.5	39

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37	Insulin Secretion in Type I Glycogen Storage Disease. Diabetes, 1969, 18, 755-758.	0.6	38
38	The Iduronidase-Deficient Mucopolysaccharidoses: Clinical and Roentgenographic Features. Pediatrics, 1976, 57, 111-122.	2.1	38
39	Ectodermal manifestations in Menkes disease. Clinical Genetics, 1985, 28, 532-540.	2.0	35
40	A blueprint for maternal and child health primary care physician education in medical genetics and genomic medicine: Recommendations of the United States Secretary for Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children. Genetics in Medicine, 2010, 12, 77-80.	2.4	35
41	Newborn screening for Duchenne muscular dystrophy in China: follow-up diagnosis and subsequent treatment. World Journal of Pediatrics, 2017, 13, 197-201.	1.8	31
42	Committee Report: Advancing the current recommended panel of conditions for newborn screening. Genetics in Medicine, 2007, 9, 792-796.	2.4	30
43	Developing a national collaborative study system for rare genetic diseases. Genetics in Medicine, 2008, 10, 325-329.	2.4	30
44	Carrier testing for spinal muscular atrophy. Genetics in Medicine, 2010, 12, 621-622.	2.4	29
45	PILI TORTI AS MARKER FOR CARRIERS OF MENKES DISEASE. Lancet, The, 1978, 311, 607-608.	13.7	28
46	We Need Expanded Newborn Screening. Pediatrics, 2006, 117, 1800-1805.	2.1	28
47	Synthesis of a metallothionein-like protein in cultured human skin fibroblasts: Relation to abnormal copper distribution in Menkes' disease. Journal of Cellular Physiology, 1981, 106, 339-348.	4.1	25
48	Evidence-Based Consensus and Systematic Review on Reducing the Time to Diagnosis of Duchenne Muscular Dystrophy. Journal of Pediatrics, 2019, 204, 305-313.e14.	1.8	24
49	Developmental changes in amino acid concentrations in human amniotic fluid: Abnormal findings in maternal phenylketonuria. American Journal of Obstetrics and Gynecology, 1971, 111, 38-42.	1.3	23
50	The high price of false positives. Molecular Genetics and Metabolism, 2006, 87, 180-183.	1.1	22
51	Including ELSI research questions in newborn screening pilot studies. Genetics in Medicine, 2019, 21, 525-533.	2.4	21
52	Appropriateness of Newborn Screening for Î±1â€Antitrypsin Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, 199-203.	1.8	20
53	Expanding research to provide an evidence base for nutritional interventions for the management of inborn errors of metabolism. Molecular Genetics and Metabolism, 2013, 109, 319-328.	1.1	19
54	A QUANTITATIVE STUDY OF RECYCLING OF ISOTOPE FROM GLYCINE-1-C14,Î±-N15 INTO VARIOUS SUBUNITS OF THE URIC ACID MOLECULE IN A NORMAL SUBJECT*. Journal of Clinical Investigation, 1961, 40, 2076-2082.	8.2	18

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55	Genetic evidence for the common identity of glucose-6-phosphatase, pyrophosphate-glucose phosphotransferase, carbamyl phosphate-glucose phosphotransferase and inorganic pyrophosphatase. Biochimica Et Biophysica Acta - General Subjects, 1977, 496, 431-435.	2.4	17
56	Continuing Lessons from Glycogen Storage Diseases. New England Journal of Medicine, 1991, 324, 55-56.	27.0	16
57	Issues in implementing prenatal screening for cystic fibrosis: Results of a working conference. Genetics in Medicine, 1999, 1, 129-135.	2.4	15
58	The diagnostic value ofserum enzyme measurements. Journal of Pediatrics, 1966, 68, 121-134.	1.8	14
59	Maximizing the Benefit of Life-Saving Treatments for Pompe Disease, Spinal Muscular Atrophy, and Duchenne Muscular Dystrophy Through Newborn Screening. JAMA Neurology, 2019, 76, 978.	9.0	14
60	The Progress and Future of US Newborn Screening. International Journal of Neonatal Screening, 2022, 8, 41.	3.2	14
61	The simultaneous occurrence of histidinemia and congenital hypoplastic anemia. Journal of Pediatrics, 1969, 75, 878-880.	1.8	12
62	Structures for clinical followâ€up: Newborn screening. Journal of Inherited Metabolic Disease, 2007, 30, 600-605.	3.6	12
63	From Developing Guidelines to Implementing Legislation: Actions of the US Advisory Committee on Heritable Disorders in Newborns and Children Toward Advancing and Improving Newborn Screening. Seminars in Perinatology, 2010, 34, 121-124.	2.5	11
64	Arylsulfatase activity in human urine: quantitative studies on patients with lysosomal disorders including metachromatic leukodystrophy. Clinica Chimica Acta, 1972, 36, 99-103.	1.1	10
65	Secretary's Advisory Committee on Heritable Disorders in Newborns and Children response to the President's Council on Bioethics report: The changing moral focus of newborn screening. Genetics in Medicine, 2011, 13, 301-304.	2.4	10
66	Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. Mental Retardation and Developmental Disabilities Research Reviews, 2006, 12, 313-315.	3.6	9
67	Ethical Issues Surrounding Newborn Screening. International Journal of Neonatal Screening, 2021, 7, 3.	3.2	9
68	Ultraviolet-Absorbing Compounds in Urine of Normal Newborns and Young Children. Clinical Chemistry, 1970, 16, 702-706.	3.2	8
69	The Fate of Newborn Screening Blood Spots. Pediatric Research, 2010, 67, 237-237.	2.3	8
70	Enzymatic and biochemical characterization of the avian glycogen body. Comparative Biochemistry and Physiology Part B: Comparative Biochemistry, 1975, 50, 525-530.	0.2	7
71	Hypertension in a Child With Type IA Glycogen Storage Disease. American Journal of Kidney Diseases, 1988, 11, 264-266.	1.9	6
72	Duchenne Muscular Dystrophy Newborn Screening, a Case Study for Examining Ethical and Legal Issues for Pilots for Emerging Disorders: Considerations and Recommendations. International Journal of Neonatal Screening, 2018, 4, 6.	3.2	6

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73	MOVRâ€"NeuroMuscular ObserVational Research, a unified data hub for neuromuscular diseases. Genetics in Medicine, 2019, 21, 536-538.	2.4	6
74	Prenatal Diagnosis in the Prevention of Handicapping Disorders. Pediatric Clinics of North America, 1973, 20, 141-149.	1.8	5
75	Introduction: Newborn screening. Mental Retardation and Developmental Disabilities Research Reviews, 2006, 12, 229-229.	3.6	5
76	A Disservice to Advances in Newborn Genetic Screening. Journal of Health and Social Behavior, 2011, 52, 277-278.	4.8	5
77	MANNOSIDOSIS AND MATERNAL PENICILLAMINE THERAPY. Lancet, The, 1976, 307, 312-313.	13.7	4
78	From a Single Child to Uniform Newborn Screening: My Lucky Life in Pediatric Medical Genetics. Annual Review of Genomics and Human Genetics, 2018, 19, 1-14.	6.2	4
79	Phenylketonuria in the General Population. New England Journal of Medicine, 1970, 282, 1486-1488.	27.0	3
80	Genetic Disease: The Present Status of Treatment. Hospital Practice (1995), 1972, 7, 75-84.	1.0	3
81	Quality improvement of newborn screening in real time. Genetics in Medicine, 2011, 13, 205.	2.4	3
82	Fifty years of newborn screening. Molecular Genetics and Metabolism, 2014, 113, 4-5.	1.1	3
83	We must now put in place an updated, comprehensive newborn screening program for deaf and hard-of-hearing infants. Genetics in Medicine, 2019, 21, 2439-2441.	2.4	3
84	A Visit with Dr. Louis Woolf, Recognizing His 100th Birthday and His Contributions to the Diagnosis and Treatment of Phenylketonuria. International Journal of Neonatal Screening, 2020, 6, 45.	3.2	3
85	The effects of colchicine on the metabolic accompaniments of phagocytosis. Arthritis and Rheumatism, 1965, 8, 749-751.	6.7	2
86	Some Medical and Social Aspects of the Treatment for Genetic-Metabolic Diseases. Annals of the American Academy of Political and Social Science, 1972, 399, 30-37.	1.6	2
87	Some major milestones and future directions. Genetics in Medicine, 2000, 2, 255-258.	2.4	2
88	Every Child Is Priceless: Debating Effective Newborn Screening Policy. Hastings Center Report, 2009, 39, 4-8.	1.0	2
89	Newborn Screening for Krabbe Disease: A Model of Cooperation. Pediatric Neurology, 2009, 40, 256-257.	2.1	2
90	THE METABOLISM OF TRYPTOPHAN IN MONGOLISM. Annals of the New York Academy of Sciences, 1970, 171, 578-586.	3.8	1

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91	Juvenile Gouty Arthritis. JAMA Pediatrics, 1985, 139, 547.	3.0	1
92	Propagation Delays in Fixed-Priority Scheduling of Periodic Tasks. , 2010, , .		1
93	Committee Report: Considerations and Recommendations for National Guidance Regarding the Retention and Use of Residual Dried Blood Spot Specimens After Newborn Screening. Obstetrical and Gynecological Survey, 2011, 66, 687-689.	0.4	1
94	Histidinemia in a Negro Child. JAMA Pediatrics, 1971, 122, 212.	3.0	0
95	617 HUMAN MILK TRACE METALS â€“ APPLICATION OF X-RAY FLUORESCENCE SPECTROMETRY TO QUANTITATION AND SCREENING. Pediatric Research, 1981, 15, 543-543.	2.3	0
96	Detection of Human Acid Î±-Glucosidase in Fibroblasts Using Monoclonal Antibodies in a Biotin-Avidin Amplified ELISA. Hybridoma, 1985, 4, 351-360.	0.6	0
97	Will there be funds to support essential clinical genetic services?. Genetics in Medicine, 2002, 4, 103-104.	2.4	0
98	Louis J. â€œSkipâ€•Elsas II, MD, FACMG. Genetics in Medicine, 2013, 15, 88-88.	2.4	0
99	Systems to determine treatment effectiveness in newborn screening. Health Matrix, 2009, 19, 155-61.	1.5	0