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	The Progress and Future of US Newborn Screening. International Journal of Neonatal Screening, 2022, 8, 41.	3.2	14
2	Ethical Issues Surrounding Newborn Screening. International Journal of Neonatal Screening, 2021, 7, 3.	3.2	9
3	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
4	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
5	MOVRâ€"NeuroMuscular ObserVational Research, a unified data hub for neuromuscular diseases. Genetics in Medicine, 2019, 21, 536-538.	2.4	6
6	Including ELSI research questions in newborn screening pilot studies. Genetics in Medicine, 2019, 21, 525-533.	2.4	21
7	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
8	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
9	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
10	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
11	Treatment Algorithm for Infants Diagnosed with Spinal Muscular Atrophy through Newborn Screening. Journal of Neuromuscular Diseases, 2018, 5, 145-158.	2.6	148
12	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
13	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
14	Newborn screening for spinal muscular atrophy: Anticipating an imminent need. Seminars in Perinatology, 2015, 39, 217-229.	2.5	51
15	Appropriateness of Newborn Screening for Î±1â€Antitrypsin Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2014, 58, 199-203.	1.8	20
16	Fifty years of newborn screening. Molecular Genetics and Metabolism, 2014, 113, 4-5.	1.1	3
17	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
18	Louis J. â€œSkipâ€•Elsas II, MD, FACMG. Genetics in Medicine, 2013, 15, 88-88.	2.4	0

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19	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
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	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
22	Strategies for Implementing Screening for Critical Congenital Heart Disease. Pediatrics, 2011, 128, e1259-e1267.	2.1	344
23	A Disservice to Advances in Newborn Genetic Screening. Journal of Health and Social Behavior, 2011, 52, 277-278.	4.8	5
24	Quality improvement of newborn screening in real time. Genetics in Medicine, 2011, 13, 205.	2.4	3
25	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
26	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
27	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
28	Carrier testing for spinal muscular atrophy. Genetics in Medicine, 2010, 12, 621-622.	2.4	29
29	The Fate of Newborn Screening Blood Spots. Pediatric Research, 2010, 67, 237-237.	2.3	8
30	Propagation Delays in Fixed-Priority Scheduling of Periodic Tasks. , 2010, , .		1
31	Every Child Is Priceless: Debating Effective Newborn Screening Policy. Hastings Center Report, 2009, 39, 4-8.	1.0	2
32	Newborn Screening for Krabbe Disease: A Model of Cooperation. Pediatric Neurology, 2009, 40, 256-257.	2.1	2
33	Systems to determine treatment effectiveness in newborn screening. Health Matrix, 2009, 19, 155-61.	1.5	0
34	Developing a national collaborative study system for rare genetic diseases. Genetics in Medicine, 2008, 10, 325-329.	2.4	30
35	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
36	Committee Report: Advancing the current recommended panel of conditions for newborn screening. Genetics in Medicine, 2007, 9, 792-796.	2.4	30

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37	Structures for clinical followâ€up: Newborn screening. Journal of Inherited Metabolic Disease, 2007, 30, 600-605.	3.6	12
38	Pompe disease diagnosis and management guideline. Genetics in Medicine, 2006, 8, 267-288.	2.4	473
39	The high price of false positives. Molecular Genetics and Metabolism, 2006, 87, 180-183.	1.1	22
40	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
41	Introduction: Newborn screening. Mental Retardation and Developmental Disabilities Research Reviews, 2006, 12, 229-229.	3.6	5
42	Diagnostic challenges for Pompe disease: An under-recognized cause of floppy baby syndrome. Genetics in Medicine, 2006, 8, 289-296.	2.4	52
43	Executive Summary. Genetics in Medicine, 2006, 8, S1-S11.	2.4	310
44	We Need Expanded Newborn Screening. Pediatrics, 2006, 117, 1800-1805.	2.1	28
45	Newborn Screening: Toward a Uniform Screening Panel and Systemâ€"Executive Summary. Pediatrics, 2006, 117, S296-S307.	2.1	386
46	Pompe disease in infants and children. Journal of Pediatrics, 2004, 144, S35-S43.	1.8	226
47	Will there be funds to support essential clinical genetic services?. Genetics in Medicine, 2002, 4, 103-104.	2.4	0
48	Some major milestones and future directions. Genetics in Medicine, 2000, 2, 255-258.	2.4	2
49	Issues in implementing prenatal screening for cystic fibrosis: Results of a working conference. Genetics in Medicine, 1999, 1, 129-135.	2.4	15
50	Continuing Lessons from Glycogen Storage Diseases. New England Journal of Medicine, 1991, 324, 55-56.	27.0	16
51	Echocardiographic abnormalities in the mucopolysaccharide storage diseases. American Journal of Cardiology, 1988, 61, 170-176.	1.6	62
52	Hypertension in a Child With Type IA Glycogen Storage Disease. American Journal of Kidney Diseases, 1988, 11, 264-266.	1.9	6
53	Juvenile Gouty Arthritis. JAMA Pediatrics, 1985, 139, 547.	3.0	1
54	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

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55	Ectodermal manifestations in Menkes disease. Clinical Genetics, 1985, 28, 532-540.	2.0	35
56	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
57	Neurotransmitter defects and treatment of disorders of hyperphenylalaninemia. Journal of Pediatrics, 1981, 98, 729-733.	1.8	60
58	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
59	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
60	Familial recurrent rhabdomyolysis due to carnitine palmityl transferase deficiency. American Journal of Medicine, 1979, 67, 167-171.	1.5	39
61	PILI TORTI AS MARKER FOR CARRIERS OF MENKES DISEASE. Lancet, The, 1978, 311, 607-608.	13.7	28
62	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
63	MANNOSIDOSIS AND MATERNAL PENICILLAMINE THERAPY. Lancet, The, 1976, 307, 312-313.	13.7	4
64	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
65	Type IV Glycogen-storage Disease: Light-microscopic, Electron-microscopic, and Enzymatic Study. American Journal of Clinical Pathology, 1976, 66, 702-709.	0.7	43
66	Hepatic Adenomata With Type 1 Glycogen Storage Disease. JAMA - Journal of the American Medical Association, 1976, 236, 1481.	7.4	119
67	Hepatic adenomata with type 1 glycogen storage disease. JAMA - Journal of the American Medical Association, 1976, 236, 1481-1484.	7.4	68
68	The Iduronidase-Deficient Mucopolysaccharidoses: Clinical and Roentgenographic Features. Pediatrics, 1976, 57, 111-122.	2.1	38
69	Enzymatic and biochemical characterization of the avian glycogen body. Comparative Biochemistry and Physiology Part B: Comparative Biochemistry, 1975, 50, 525-530.	0.2	7
70	Prenatal Diagnosis in the Prevention of Handicapping Disorders. Pediatric Clinics of North America, 1973, 20, 141-149.	1.8	5
71	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
72	Genetic Disease: The Present Status of Treatment. Hospital Practice (1995), 1972, 7, 75-84.	1.0	3

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73	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
74	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
75	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
76	Xeroderma Pigmentosum: A Rapid Sensitive Method for Prenatal Diagnosis. Science, 1971, 174, 147-150.	12.6	75
77	Histidinemia in a Negro Child. JAMA Pediatrics, 1971, 122, 212.	3.0	0
78	Phenylketonuria in the General Population. New England Journal of Medicine, 1970, 282, 1486-1488.	27.0	3
79	THE METABOLISM OF TRYPTOPHAN IN MONGOLISM. Annals of the New York Academy of Sciences, 1970, 171, 578-586.	3.8	1
80	Ultraviolet-Absorbing Compounds in Urine of Normal Newborns and Young Children. Clinical Chemistry, 1970, 16, 702-706.	3.2	8
81	Infantile Metachromatic Leukodystrophy. New England Journal of Medicine, 1970, 282, 1336-1340.	27.0	90
82	Insulin Secretion in Type I Glycogen Storage Disease. Diabetes, 1969, 18, 755-758.	0.6	38
83	The simultaneous occurrence of histidinemia and congenital hypoplastic anemia. Journal of Pediatrics, 1969, 75, 878-880.	1.8	12
84	Excessive production of uric acid in type I glycogen storage disease. Journal of Pediatrics, 1968, 72, 488-496.	1.8	72
85	Relationships between glycogen storage disease and tophaceous gout. American Journal of Medicine, 1967, 42, 58-66.	1.5	98
86	The diagnostic value ofserum enzyme measurements. Journal of Pediatrics, 1966, 68, 121-134.	1.8	14
87	The effects of colchicine on the metabolic accompaniments of phagocytosis. Arthritis and Rheumatism, 1965, 8, 749-751.	6.7	2
88	The interrelationship of glycogen storage disease and gout. Arthritis and Rheumatism, 1965, 8, 780-785.	6.7	66
89	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
90	Turnover of Ribosomal RNA in Rat Liver. Science, 1965, 149, 1093-1095.	12.6	132

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91	Mammalian enzyme induction by hydrocortisone. Journal of Molecular Biology, 1964, 9, 100-108.	4.2	79
92	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
93	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
94	X-ray diffraction studies of the tophaceous deposits in gout. Arthritis and Rheumatism, 1963, 6, 97-103.	6.7	39
95	Biochemistry of Uric Acid and Its Relation to Gout. New England Journal of Medicine, 1963, 268, 712-716.	27.0	144
96	PROPERTIES OF ACATALASIC CELLS GROWING IN VITRO. Journal of Experimental Medicine, 1962, 115, 313-328.	8.5	58
97	The old and new concepts of acute gouty arthritis. Arthritis and Rheumatism, 1962, 5, 616-623.	6.7	71
98	THE RENAL EXCRETION OF URIC ACID IN GOUT. Journal of Clinical Investigation, 1962, 41, 1094-1098.	8.2	101
99	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