Robert D Steiner

List of Publications by Citations

199 papers	7,971 citations	47 h-index	79 g-index
224 ext. papers	8,983 ext. citations	4.3 avg, IF	5.62 L-index

#	Paper	IF	Citations
199	Pompe disease diagnosis and management guideline. *Genetics in Medicine*, **2006**, 8, 267-88	8.1	376
198	Mutations in the human sterol delta7-reductase gene at 11q12-13 cause Smith-Lemli-Opitz syndrome. *American Journal of Human Genetics*, **1998**, 63, 55-62	11	363
197	Newborn screening: toward a uniform screening panel and system--executive summary. *Pediatrics*, **2006**, 117, S296-307	7.4	293
196	Enzyme replacement therapy in mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). *Journal of Pediatrics*, **2004**, 144, 574-80	3.6	235
195	Osteogenesis imperfecta. *Annual Review of Medicine*, **1992**, 43, 269-82	17.4	223
194	Long-term follow-up of endurance and safety outcomes during enzyme replacement therapy for mucopolysaccharidosis VI: Final results of three clinical studies of recombinant human N-acetylgalactosamine 4-sulfatase. *Molecular Genetics and Metabolism*, **2008**, 94, 469-475	3.7	181
193	Missense mutations in CRELD1 are associated with cardiac atrioventricular septal defects. *American Journal of Human Genetics*, **2003**, 72, 1047-52	11	163
192	Alendronate for the treatment of pediatric osteogenesis imperfecta: a randomized placebo-controlled study. *Journal of Clinical Endocrinology and Metabolism*, **2011**, 96, 355-64	5.6	157
191	The near universal presence of autism spectrum disorders in children with Smith-Lemli-Opitz syndrome. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1511-8	2.5	139
190	Risedronate in children with osteogenesis imperfecta: a randomised, double-blind, placebo-controlled trial. *Lancet, The*, **2013**, 382, 1424-32	40	129
189	Biochemical, phenotypic and neurophysiological characterization of a genetic mouse model of RSH/Smith--Lemli--Opitz syndrome. *Human Molecular Genetics*, **2001**, 10, 555-64	5.6	129
188	Agalsidase alfa and kidney dysfunction in Fabry disease. *Journal of the American Society of Nephrology: JASN*, **2009**, 20, 1132-9	12.7	127
187	Genetic Counseling and Screening of Consanguineous Couples and Their Offspring: Recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling*, **2002**, 11, 97-119	2.5	127
186	Screening and treatment for lipid disorders in children and adolescents: systematic evidence review for the US Preventive Services Task Force. *Pediatrics*, **2007**, 120, e189-214	7.4	123
185	Mutations in FKBP10, which result in Bruck syndrome and recessive forms of osteogenesis imperfecta, inhibit the hydroxylation of telopeptide lysines in bone collagen. *Human Molecular Genetics*, **2013**, 22, 1-17	5.6	117
184	Osteogenesis imperfecta: recent findings shed new light on this once well-understood condition. *Genetics in Medicine*, **2009**, 11, 375-85	8.1	113
183	Clinical course of sly syndrome (mucopolysaccharidosis type VII). *Journal of Medical Genetics*, **2016**, 53, 403-18	5.8	103

182	Mutations in PPIB (cyclophilin B) delay type I procollagen chain association and result in perinatal lethal to moderate osteogenesis imperfecta phenotypes. *Human Molecular Genetics*, **2011**, 20, 1595-609	5.6	102
181	Central nervous system stem cell transplantation for children with neuronal ceroid lipofuscinosis. *Journal of Neurosurgery: Pediatrics*, **2013**, 11, 643-52	2.1	100
180	Risk factors for premature ovarian failure in females with galactosemia. *Journal of Pediatrics*, **2000**, 137, 833-41	3.6	84
179	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and gene-level specification of the ACMG/AMP guidelines for sequence variant interpretation. *Human Mutation*, **2018**, 39, 1614-1622	4.7	83
178	Klippel-Trenaunay-Weber syndrome associated with a 5:11 balanced translocation. *American Journal of Medical Genetics Part A*, **1995**, 59, 492-4		79
177	2-Methylbutyryl-coenzyme A dehydrogenase deficiency: a new inborn error of L-isoleucine metabolism. *Pediatric Research*, **2000**, 47, 830-3	3.2	79
176	Epidemiology, diagnosis, and treatment of cerebrotendinous xanthomatosis (CTX). *Journal of Inherited Metabolic Disease*, **2017**, 40, 771-781	5.4	77
175	Bisphosphonate therapy for osteogenesis imperfecta. *Cochrane Database of Systematic Reviews*, **2008**, CD005088		77
174	Deficits in memory strategy use related to prefrontal dysfunction during early development: Evidence from children with phenylketonuria.. *Neuropsychology*, **2001**, 15, 221-229	3.8	73
173	Bisphosphonate therapy for osteogenesis imperfecta. *The Cochrane Library*, **2016**, 10, CD005088	5.2	72
172	Carrier frequency of the common mutation IVS8-1G>C in DHCR7 and estimate of the expected incidence of Smith-Lemli-Opitz syndrome. *Molecular Genetics and Metabolism*, **2001**, 72, 67-71	3.7	72
171	Bisphosphonate therapy for osteogenesis imperfecta. *Cochrane Database of Systematic Reviews*, **2014**, CD005088		71
170	Enzyme replacement therapy for mucopolysaccharidosis VI: evaluation of long-term pulmonary function in patients treated with recombinant human N-acetylgalactosamine 4-sulfatase. *Journal of Inherited Metabolic Disease*, **2010**, 33, 51-60	5.4	66
169	Sterol metabolism disorders and neurodevelopment-an update. *Developmental Disabilities Research Reviews*, **2013**, 17, 197-210		62
168	Smith-Lemli-Opitz syndrome. *Expert Reviews in Molecular Medicine*, **2011**, 13, e24	6.7	62
167	Ascorbate decreases Fabry cerebral hyperperfusion suggesting a reactive oxygen species abnormality: an arterial spin tagging study. *Journal of Magnetic Resonance Imaging*, **2004**, 20, 674-83	5.6	61
166	Nutrient intakes and physical growth of children with phenylketonuria undergoing nutrition therapy. *Journal of the American Dietetic Association*, **2003**, 103, 1167-73		60
165	Studies of collagen synthesis and structure in the differentiation of child abuse from osteogenesis imperfecta. *Journal of Pediatrics*, **1996**, 128, 542-7	3.6	59

164	Genetics of familial hypercholesterolemia. *Current Atherosclerosis Reports*, **2015**, 17, 491	6	58
163	Shared familial transmission of autism spectrum and attention-deficit/hyperactivity disorders. *Journal of Child Psychology and Psychiatry and Allied Disciplines*, **2014**, 55, 819-27	7.9	58
162	Ophthalmic drops causing coma in an infant. *Journal of Pediatrics*, **2001**, 138, 441-3	3.6	58
161	Multi-domain impact of elosufase alfa in Morquio A syndrome in the pivotal phase III trial. *Molecular Genetics and Metabolism*, **2015**, 114, 178-85	3.7	57
160	Apparent underdiagnosis of Cerebrotendinous Xanthomatosis revealed by analysis of ~60,000 human exomes. *Molecular Genetics and Metabolism*, **2015**, 116, 298-304	3.7	56
159	Mutation analysis and description of sixteen RSH/Smith-Lemli-Opitz syndrome patients: Polymerase chain reaction-based assays to simplify genotyping. *American Journal of Medical Genetics Part A*, **2000**, 94, 214-227		56
158	Importance of surgical history in diagnosing mucopolysaccharidosis type II (Hunter syndrome): data from the Hunter Outcome Survey. *Genetics in Medicine*, **2010**, 12, 816-22	8.1	52
157	Long-term management of patients with urea cycle disorders. *Journal of Pediatrics*, **2001**, 138, S56-60; discussion S60-1	3.6	50
156	Enzyme replacement therapy for mucopolysaccharidosis VI: Growth and pubertal development in patients treated with recombinant human N-acetylgalactosamine 4-sulfatase. *Journal of Pediatric Rehabilitation Medicine*, **2010**, 3, 89-100	1.4	50
155	Correlates of language impairment in children with galactosaemia. *Journal of Inherited Metabolic Disease*, **2008**, 31, 524-32	5.4	49
154	Cholesterol supplementation with egg yolk increases plasma cholesterol and decreases plasma 7-dehydrocholesterol in Smith-Lemli-Opitz syndrome. *American Journal of Medical Genetics Part A*, **2000**, 93, 360-5		49
153	Neurocognition across the spectrum of mucopolysaccharidosis type I: Age, severity, and treatment. *Molecular Genetics and Metabolism*, **2015**, 116, 61-8	3.7	48
152	A membrane defect in the pathogenesis of the Smith-Lemli-Opitz syndrome. *Journal of Lipid Research*, **2006**, 47, 134-43	6.3	47
151	Nonketotic hyperglycinemia: atypical clinical and biochemical manifestations. *Journal of Pediatrics*, **1996**, 128, 243-6	3.6	47
150	Hypomorphic temperature-sensitive alleles of NSDHL cause CK syndrome. *American Journal of Human Genetics*, **2010**, 87, 905-14	11	46
149	A cross-sectional multicenter study of osteogenesis imperfecta in North America - results from the linked clinical research centers. *Clinical Genetics*, **2015**, 87, 133-40	4	45
148	Treatment of Smith-Lemli-Opitz syndrome and other sterol disorders. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2012**, 160C, 285-94	3.1	45
147	Apolipoprotein E-low density lipoprotein receptor interaction affects spatial memory retention and brain ApoE levels in an isoform-dependent manner. *Neurobiology of Disease*, **2014**, 64, 150-62	7.5	44

146	Improved growth and nutrition status in children with methylmalonic or propionic acidemia fed an elemental medical food. *Molecular Genetics and Metabolism*, **2003**, 80, 181-8	3.7	44
145	Laboratory evaluation of urea cycle disorders. *Journal of Pediatrics*, **2001**, 138, S21-9	3.6	44
144	Sterol balance in the Smith-Lemli-Opitz syndrome: reduction in whole body cholesterol synthesis and normal bile acid production. *Journal of Lipid Research*, **2000**, 41, 1437-1447	6.3	44
143	Design, baseline characteristics, and early findings of the MPS VI (mucopolysaccharidosis VI) Clinical Surveillance Program (CSP). *Journal of Inherited Metabolic Disease*, **2013**, 36, 373-84	5.4	43
142	Smith-Lemli-Opitz syndrome: the first malformation syndrome associated with defective cholesterol synthesis. *Molecular Genetics and Metabolism*, **2000**, 71, 154-62	3.7	42
141	Variability in phenylalanine control predicts IQ and executive abilities in children with phenylketonuria. *Molecular Genetics and Metabolism*, **2014**, 111, 445-51	3.7	41
140	Age-related working memory impairments in children with prefrontal dysfunction associated with phenylketonuria. *Journal of the International Neuropsychological Society*, **2002**, 8, 1-11	3.1	41
139	Consensus statement from a conference for the management of patients with urea cycle disorders. *Journal of Pediatrics*, **2001**, 138, S1-5	3.6	41
138	Liquid chromatography-tandem mass spectrometry determination of plasma 24S-hydroxycholesterol with chromatographic separation of 25-hydroxycholesterol. *Analytical Biochemistry*, **2008**, 381, 151-3	3.1	40
137	Fabry disease in genetic counseling practice: recommendations of the National Society of Genetic Counselors. *Journal of Genetic Counseling*, **2002**, 11, 121-46	2.5	39
136	Sterol balance in the Smith-Lemli-Opitz syndrome. Reduction in whole body cholesterol synthesis and normal bile acid production. *Journal of Lipid Research*, **2000**, 41, 1437-47	6.3	39
135	Evaluation of miglustat as maintenance therapy after enzyme therapy in adults with stable type 1 Gaucher disease: a prospective, open-label non-inferiority study. *Orphanet Journal of Rare Diseases*, **2012**, 7, 102	4.2	38
134	Skeletal changes in epidermal nevus syndrome: does focal bone disease harbor clues concerning pathogenesis?. *American Journal of Medical Genetics, Part A*, **2005**, 139A, 67-77	2.5	38
133	Cholesterol storage defect in RSH/Smith-Lemli-Opitz syndrome fibroblasts. *Molecular Genetics and Metabolism*, **2002**, 75, 325-34	3.7	38
132	Treatment of pyruvate carboxylase deficiency with high doses of citrate and aspartate. *American Journal of Medical Genetics Part A*, **1999**, 87, 331-8		38
131	Inhibitory control in children with phenylketonuria. *Developmental Neuropsychology*, **2006**, 30, 845-64	1.8	37
130	Sibling Recurrence Risk and Cross-aggregation of Attention-Deficit/Hyperactivity Disorder and Autism Spectrum Disorder. *JAMA Pediatrics*, **2019**, 173, 147-152	8.3	37
129	A novel mutation in medium chain acyl-CoA dehydrogenase causes sudden neonatal death. *Journal of Clinical Investigation*, **1994**, 94, 1477-83	15.9	36

128	Mutations in adducin are associated with inherited cerebral palsy. *Annals of Neurology*, **2013**, 74, 805-14	9.4	35
127	Effects of dietary cholesterol and simvastatin on cholesterol synthesis in Smith-Lemli-Opitz syndrome. *Pediatric Research*, **2009**, 65, 681-5	3.2	35
126	Remarkable improvement in adult Leigh syndrome with partial cytochrome c oxidase deficiency. *Neurology*, **2003**, 60, 865-8	6.5	35
125	Clinical and molecular features of congenital disorder of glycosylation in patients with type 1 sialotransferrin pattern and diverse ethnic origins. *Journal of Pediatrics*, **2002**, 141, 695-700	3.6	35
124	Postmortem screening for fatty acid oxidation disorders by analysis of Guthrie cards with tandem mass spectrometry in sudden unexpected death in infancy. *Journal of Pediatrics*, **2002**, 141, 833-6	3.6	35
123	Cognitive, medical, and neuroimaging characteristics of attenuated mucopolysaccharidosis type II. *Molecular Genetics and Metabolism*, **2015**, 114, 170-7	3.7	34
122	"I'm fine; I'm just waiting for my disease": the new and growing class of presymptomatic patients. *Neurology*, **2011**, 77, 522-3	6.5	33
121	Deletion of a single mevalonate kinase (Mvk) allele yields a murine model of hyper-IgD syndrome. *Journal of Inherited Metabolic Disease*, **2007**, 30, 888-95	5.4	33
120	Cholesterol metabolism and suicidality in Smith-Lemli-Opitz syndrome carriers. *American Journal of Psychiatry*, **2004**, 161, 2123-6	11.9	33
119	DHCR7 nonsense mutations and characterisation of mRNA nonsense mediated decay in Smith-Lemli-Opitz syndrome. *Journal of Medical Genetics*, **2005**, 42, 350-7	5.8	32
118	Interhemispheric interaction during childhood: II. Children with early-treated phenylketonuria. *Developmental Neuropsychology*, **2000**, 18, 53-71	1.8	32
117	Profiling sterols in cerebrotendinous xanthomatosis: utility of Girard derivatization and high resolution exact mass LC-ESI-MS(n) analysis. *Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences*, **2011**, 879, 1384-92	3.2	31
116	Short/branched-chain acyl-CoA dehydrogenase deficiency due to an IVS3+3A>G mutation that causes exon skipping. *Human Genetics*, **2006**, 118, 680-90	6.3	31
115	Clinical profile of a male with Rett syndrome. *Brain and Development*, **2005**, 27 Suppl 1, S69-S71	2.2	31
114	Cholesterol supplementation does not improve developmental progress in Smith-Lemli-Opitz syndrome. *Journal of Pediatrics*, **2004**, 144, 783-91	3.6	30
113	Age-related working memory impairments in children with prefrontal dysfunction associated with phenylketonuria. *Journal of the International Neuropsychological Society*, **2002**, 8, 1-11	3.1	30
112	Ezetimibe reduces plant sterol accumulation and favorably increases platelet count in sitosterolemia. *Journal of Pediatrics*, **2015**, 166, 125-31	3.6	29
111	Assays of plasma dehydrocholesteryl esters and oxysterols from Smith-Lemli-Opitz syndrome patients. *Journal of Lipid Research*, **2013**, 54, 244-53	6.3	29

110	Age-related decline in the microstructural integrity of white matter in children with early- and continuously-treated PKU: a DTI study of the corpus callosum. *Molecular Genetics and Metabolism*, **2010**, 99 Suppl 1, S41-6	3.7	29
109	Cesarean delivery is not associated with decreased at-birth fracture rates in osteogenesis imperfecta. *Genetics in Medicine*, **2016**, 18, 570-6	8.1	27
108	A blood test for cerebrotendinous xanthomatosis with potential for disease detection in newborns. *Journal of Lipid Research*, **2014**, 55, 146-54	6.3	27
107	ESI-MS/MS quantification of 7alpha-hydroxy-4-cholesten-3-one facilitates rapid, convenient diagnostic testing for cerebrotendinous xanthomatosis. *Clinica Chimica Acta*, **2010**, 411, 43-8	6.2	27
106	Prolonged exposure to high and variable phenylalanine levels over the lifetime predicts brain white matter integrity in children with phenylketonuria. *Molecular Genetics and Metabolism*, **2015**, 114, 19-24	3.7	26
105	Newborn Screening for Krabbe Disease: the New York State Model. *Pediatric Neurology*, **2009**, 40, 253-255	2.9	26
104	Unique aspects of sequence variant interpretation for inborn errors of metabolism (IEM): The ClinGen IEM Working Group and the Phenylalanine Hydroxylase Gene. *Human Mutation*, **2018**, 39, 1569-1580	4.7	26
103	Natural History of Perinatal and Infantile Hypophosphatasia: A Retrospective Study. *Journal of Pediatrics*, **2019**, 209, 116-124.e4	3.6	25
102	Analysis of hedgehog signaling in cerebellar granule cell precursors in a conditional Nsdhl allele demonstrates an essential role for cholesterol in postnatal CNS development. *Human Molecular Genetics*, **2015**, 24, 2808-25	5.6	25
101	Age-related working memory impairments in children with prefrontal dysfunction associated with phenylketonuria. *Journal of the International Neuropsychological Society*, **2002**, 8, 1-11	3.1	25
100	Two siblings with early onset fetal akinesia deformation sequence and hydranencephaly: Further evidence for autosomal recessive inheritance of hydranencephaly, fowler type. *American Journal of Medical Genetics Part A*, **2002**, 108, 41-44		24
99	Tandem mass spectrometry in newborn screening: a primer for neonatal and perinatal nurses. *Journal of Perinatal and Neonatal Nursing*, **2004**, 18, 41-58; quiz 59-60	1.5	24
98	Iron status of children with phenylketonuria undergoing nutrition therapy assessed by transferrin receptors. *Genetics in Medicine*, **2004**, 6, 96-101	8.1	23
97	Shaken baby syndrome--a forensic pediatric response. *Pediatrics*, **1998**, 101, 321-3	7.4	23
96	A longitudinal study of emotional adjustment, quality of life and adaptive function in attenuated MPS II. *Molecular Genetics and Metabolism Reports*, **2016**, 7, 32-9	1.8	23
95	Inborn Errors of Metabolism (Metabolic Disorders). *Pediatrics in Review*, **2016**, 37, 3-15; quiz 16-7, 47	1.1	22
94	Processing speed and executive abilities in children with phenylketonuria. *Neuropsychology*, **2012**, 26, 735-43	3.8	22
93	The effects of sterol structure upon sterol esterification. *Atherosclerosis*, **2010**, 208, 155-60	3.1	22

92	Eye findings in 8 children and a spontaneously aborted fetus with RSH/Smith-Lemli-Opitz syndrome. *American Journal of Medical Genetics Part A*, **1998**, 80, 501-5		22
91	Fetal demise with Smith□emli□Opitz syndrome confirmed by tissue sterol analysis and the absence of measurable 7-dehydrocholesterol □7-reductase activity in chorionic villi **2000**, 20, 238-240		22
90	Hydrops fetalis: role of the geneticist. *Seminars in Perinatology*, **1995**, 19, 516-24	3.3	22
89	Deficits in memory strategy use related to prefrontal dysfunction during early development: evidence from children with phenylketonuria. *Neuropsychology*, **2001**, 15, 221-9	3.8	22
88	Rhabdomyolysis in the military: recognizing late-onset very long-chain acyl Co-A dehydrogenase deficiency. *Military Medicine*, **2006**, 171, 657-8	1.3	21
87	Effects of dietary cholesterol on plasma lipoproteins in Smith-Lemli-Opitz syndrome. *Pediatric Research*, **2004**, 56, 726-32	3.2	21
86	In vitro incubation with influenza virus primes human polymorphonuclear leukocyte generation of superoxide. *American Journal of Respiratory Cell and Molecular Biology*, **1991**, 4, 347-54	5.7	21
85	Executive strategic processing during verbal fluency performance in children with phenylketonuria. *Child Neuropsychology*, **2011**, 17, 105-17	2.7	20
84	Stem cell-mediated regeneration of the intervertebral disc: cellular and molecular challenge. *Neurosurgical Focus*, **2008**, 24, E21	4.2	20
83	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. *Clinical Genetics*, **2018**, 94, 502-511	4	20
82	Maternal prepregnancy body mass index and offspring attention-deficit/hyperactivity disorder: a□quasi-experimental sibling-comparison, population-based design. *Journal of Child Psychology and Psychiatry and Allied Disciplines*, **2017**, 58, 240-247	7.9	19
81	Response monitoring in children with phenylketonuria. *Neuropsychology*, **2009**, 23, 130-4	3.8	19
80	Asp187Asn mutation of gelsolin in an American kindred with familial amyloidosis, Finnish type (FAP IV). *Human Genetics*, **1995**, 95, 327-30	6.3	19
79	Intravenous Fish Oil and Pediatric Intestinal Failure-Associated Liver Disease: Changes in Plasma Phytosterols, Cytokines, and Bile Acids and Erythrocyte Fatty Acids. *Journal of Parenteral and Enteral Nutrition*, **2018**, 42, 633-641	4.2	18
78	Cellular therapy for childhood neurodegenerative disease. Part II: clinical trial design and implementation. *Neurosurgical Focus*, **2008**, 24, E23	4.2	18
77	Cellular therapy for childhood neurodegenerative disease. Part I: rationale and preclinical studies. *Neurosurgical Focus*, **2008**, 24, E22	4.2	18
76	Combination of diaphragmatic eventration and microphthalmia/anophthalmia is probably nonrandom. *American Journal of Medical Genetics Part A*, **2002**, 108, 45-50		18
75	Research challenges in central nervous system manifestations of inborn errors of metabolism. *Molecular Genetics and Metabolism*, **2011**, 102, 326-38	3.7	17

74	Update on newborn dried bloodspot testing for cerebrotendinous xanthomatosis: An available high-throughput liquid-chromatography tandem mass spectrometry method. *Molecular Genetics and Metabolism Reports*, **2016**, 7, 11-5	1.8	16
73	Growth characteristics in individuals with osteogenesis imperfecta in North America: results from a multicenter study. *Genetics in Medicine*, **2019**, 21, 275-283	8.1	15
72	Changes in plasma and urine globotriaosylceramide levels do not predict Fabry disease progression over 1 year of agalsidase alfa. *Genetics in Medicine*, **2013**, 15, 983-9	8.1	15
71	Black children deficient in galactose 1-phosphate uridyltransferase: correlation of activity and immunoreactive protein in erythrocytes and leukocytes. *Journal of Pediatrics*, **1997**, 130, 972-80	3.6	15
70	The role of evidence-based medicine and clinical trials in rare genetic disorders. *Clinical Genetics*, **2008**, 74, 197-207	4	15
69	Enhanced placental cholesterol efflux by fetal HDL in Smith-Lemli-Opitz syndrome. *Molecular Genetics and Metabolism*, **2008**, 94, 240-7	3.7	15
68	Potential for misdiagnosis due to lack of metabolic derangement in combined methylmalonic aciduria/hyperhomocysteinemia (cblC) in the neonate. *Journal of Perinatology*, **2003**, 23, 384-6	3.1	15
67	Evidence based medicine in inborn errors of metabolism: is there any and how to find it. *American Journal of Medical Genetics, Part A*, **2005**, 134A, 192-7	2.5	15
66	A useful multi-analyte blood test for cerebrotendinous xanthomatosis. *Clinical Biochemistry*, **2014**, 47, 860-3	3.5	14
65	Cerebrotendinous xanthomatosis: a treatable disease with juvenile cataracts as a presenting sign. *JAMA Ophthalmology*, **2011**, 129, 1087-8		14
64	Feedback inhibition of the cholesterol biosynthetic pathway in patients with Smith-Lemli-Opitz syndrome as demonstrated by urinary mevalonate excretion. *Journal of Lipid Research*, **2002**, 43, 1661-9	6.3	14
63	Alterations in membrane caveolae and BKCa channel activity in skin fibroblasts in Smith-Lemli-Opitz syndrome. *Molecular Genetics and Metabolism*, **2011**, 104, 346-55	3.7	13
62	Development of Clinical Domain Working Groups for the Clinical Genome Resource (ClinGen): lessons learned and plans for the future. *Genetics in Medicine*, **2019**, 21, 987-993	8.1	13
61	Prevalence estimation for monogenic autosomal recessive diseases using population-based genetic data. *Human Genetics*, **2015**, 134, 659-69	6.3	12
60	Identification of 7β24-dihydroxy-3-oxocholest-4-en-26-oic and 7β25-dihydroxy-3-oxocholest-4-en-26-oic acids in human cerebrospinal fluid and plasma. *Biochimie*, **2018**, 153, 86-98	4.6	12
59	Elevated Autophagy and Mitochondrial Dysfunction in the Smith-Lemli-Opitz Syndrome. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 431-442	1.8	12
58	Clinical utility of endurance measures for evaluation of treatment in patients with mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). *Journal of Pediatric Rehabilitation Medicine*, **2010**, 3, 119-27	1.4	12
57	Autosomal dominant transmission of acrodysostosis. *Clinical Dysmorphology*, **1992**, 1, 201???206	0.9	12

56	No evidence for mevalonate shunting in moderately affected children with Smith-Lemli-Opitz syndrome. *Journal of Inherited Metabolic Disease*, **2012**, 35, 859-69	5.4	11
55	Commentary on: "Newborn screening for Krabbe Disease: the New York state model" and "the long-term outcomes of presymptomatic infants transplanted for Krabbe disease. A report of the workshop held on July 11 and 12, 2008, Holiday Valley, New York". *Genetics in Medicine*, **2009**, 11, 411-3	8.1	11
54	Sitosterolemia Presenting as Pseudohomozygous Familial Hypercholesterolemia. *Clinical Medicine and Research*, **2016**, 14, 103-8	1.4	10
53	Smith-Lemli-Opitz syndrome and inborn errors of cholesterol synthesis: summary of the 2007 SLO/RSH Foundation scientific conference sponsored by the National Institutes of Health. *Genetics in Medicine*, **2009**, 11, 359-64	8.1	10
52	Perinatal onset mevalonate kinase deficiency. *Pediatric and Developmental Pathology*, **2011**, 14, 301-6	2.2	10
51	Increased nonsterol isoprenoids, dolichol and ubiquinone, in the Smith-Lemli-Opitz syndrome: effects of dietary cholesterol. *Journal of Lipid Research*, **2006**, 47, 2789-98	6.3	10
50	Severe subacute GM2 gangliosidosis caused by an apparently silent HEXA mutation (V324V) that results in aberrant splicing and reduced HEXA mRNA. *American Journal of Medical Genetics Part A*, **2004**, 127A, 158-66		10
49	Intestinal absorption of cholesterol by patients with Smith-Lemli-Opitz syndrome. *Pediatric Research*, **2005**, 57, 765-70	3.2	10
48	A simple PCR-based assay allows detection of a common mutation, IVS8-1G-->C, in DHCR7 in Smith-Lemli-Opitz syndrome. *Genetic Testing and Molecular Biomarkers*, **1999**, 3, 361-3		10
47	Protein status of infants with phenylketonuria undergoing nutrition management. *Journal of the American College of Nutrition*, **1999**, 18, 102-7	3.5	10
46	Corneal abnormalities in a mother and daughter with focal dermal hypoplasia (Goltz-Gorlin syndrome). *American Journal of Ophthalmology*, **1995**, 120, 256-8	4.9	10
45	Fetal demise with Smith-Lemli-Opitz syndrome confirmed by tissue sterol analysis and the absence of measurable 7-dehydrocholesterol Delta(7)-reductase activity in chorionic villi. *Prenatal Diagnosis*, **2000**, 20, 238-40	3.2	10
44	Breastfeeding infants with phenylketonuria in the United States and Canada. *Breastfeeding Medicine*, **2014**, 9, 142-8	2.1	9
43	Hearing loss in individuals with osteogenesis imperfecta in North America: Results from a multicenter study. *American Journal of Medical Genetics, Part A*, **2020**, 182, 697-704	2.5	9
42	Fructose-1,6-diphosphatase deficiency and glyceroluria: one possible etiology for GIS. *Molecular Genetics and Metabolism*, **2000**, 69, 338-40	3.7	8
41	A Multicenter Observational Cohort Study to Evaluate the Effects of Bisphosphonate Exposure on Bone Mineral Density and Other Health Outcomes in Osteogenesis Imperfecta. *JBMR Plus*, **2019**, 3, e10118	3.9	7
40	Challenging behavior in Smith-Lemli-Opitz syndrome: initial test of biobehavioral influences. *Cognitive and Behavioral Neurology*, **2013**, 26, 23-9	1.6	7
39	PHENOTYPIC VARIABILITY IN INDIVIDUALS WITH TYPE V OSTEOGENESIS IMPERFECTA WITH IDENTICAL IFITM5 MUTATIONS **2013**, 1, 37-42		7

38	Mobility in osteogenesis imperfecta: a multicenter North American study. *Genetics in Medicine*, **2019**, 21, 2311-2318	8.1	6
37	Challenges to breastfeeding infants with phenylketonuria. *Journal of Pediatric Nursing*, **2015**, 30, 219-26	2.2	6
36	Feeding impairments associated with plasma sterols in Smith-Lemli-Opitz syndrome. *Journal of Pediatrics*, **2014**, 165, 836-41.e1	3.6	6
35	Executive response monitoring and inhibitory control in children with phenylketonuria: effects of expectancy. *Developmental Neuropsychology*, **2013**, 38, 139-52	1.8	6
34	Lowered DHCR7 activity measured by ergosterol conversion in multiple cell types in Smith-Lemli-Opitz syndrome. *Molecular Genetics and Metabolism*, **2004**, 83, 175-83	3.7	6
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