Robert D Steiner

List of Publications by Year
in descending order

204	9,990	31974	48312
papers	citations	53 h-index	88 g-index

224	224	224	9840
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	Mutations in the Human Sterol Î”7-Reductase Gene at 11q12-13 Cause Smith-Lemli-Opitz Syndrome. American Journal of Human Genetics, 1998, 63, 55-62.	6.2	405
3	Newborn Screening: Toward a Uniform Screening Panel and Systemâ€”Executive Summary. Pediatrics, 2006, 117, S296-S307.	2.1	386
4	Enzyme replacement therapy in mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). Journal of Pediatrics, 2004, 144, 574-580.	1.8	267
5	Osteogenesis Imperfecta. Annual Review of Medicine, 1992, 43, 269-282.	12.2	244
6	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.	1.1	198
7	Missense Mutations in CRELD1 Are Associated with Cardiac Atrioventricular Septal Defects. American Journal of Human Genetics, 2003, 72, 1047-1052.	6.2	189
8	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.	1.6	184
9	Alendronate for the Treatment of Pediatric Osteogenesis Imperfecta: A Randomized Placebo-Controlled Study. Journal of Clinical Endocrinology and Metabolism, 2011, 96, 355-364.	3.6	184
10	Bisphosphonate therapy for osteogenesis imperfecta. The Cochrane Library, 2016, 2016, CD005088.	2.8	173
11	The near universal presence of autism spectrum disorders in children with Smithâ€“Lemliâ€“Opitz syndrome. American Journal of Medical Genetics, Part A, 2006, 140A, 1511-1518.	1.2	162
12	Screening and Treatment for Lipid Disorders in Children and Adolescents: Systematic Evidence Review for the US Preventive Services Task Force. Pediatrics, 2007, 120, e189-e214.	2.1	160
13	Risedronate in children with osteogenesis imperfecta: a randomised, double-blind, placebo-controlled trial. Lancet, The, 2013, 382, 1424-1432.	13.7	158
14	Biochemical, phenotypic and neurophysiological characterization of a genetic mouse model of RSH/Smith-Lemli-Opitz syndrome. Human Molecular Genetics, 2001, 10, 555-564.	2.9	150
15	Agalsidase Alfa and Kidney Dysfunction in Fabry Disease. Journal of the American Society of Nephrology: JASN, 2009, 20, 1132-1139.	6.1	148
16	Osteogenesis imperfecta: Recent findings shed new light on this once well-understood condition. Genetics in Medicine, 2009, 11, 375-385.	2.4	141
17	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.	2.9	135
18	Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418.	3.2	133

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19	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and geneâ€level specification of the ACMG/AMP guidelines for sequence variant interpretation. Human Mutation, 2018, 39, 1614-1622.	2.5	132
20	Epidemiology, diagnosis, and treatment of cerebrotendinous xanthomatosis (CTX). Journal of Inherited Metabolic Disease, 2017, 40, 771-781.	3.6	127
21	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, 1595-1609.	2.9	118
22	Central nervous system stem cell transplantation for children with neuronal ceroid lipofuscinosis. Journal of Neurosurgery: Pediatrics, 2013, 11, 643-652.	1.3	117
23	Klippel-Trenaunay-Weber syndrome associated with a 5:11 balanced translocation. American Journal of Medical Genetics Part A, 1995, 59, 492-494.	2.4	99
24	Risk factors for premature ovarian failure in females with galactosemia. Journal of Pediatrics, 2000, 137, 833-841.	1.8	99
25	Bisphosphonate therapy for osteogenesis imperfecta. , 2014, , CD005088.		93
26	2-Methylbutyryl-Coenzyme A Dehydrogenase Deficiency: A New Inborn Error of L-Isoleucine Metabolism. Pediatric Research, 2000, 47, 830-833.	2.3	92
27	Bisphosphonate therapy for osteogenesis imperfecta. , 2008, , CD005088.		87
28	Carrier Frequency of the Common Mutation IVS8-1G>C in DHCR7 and Estimate of the Expected Incidence of Smithâ€“Lemliâ€“Opitz Syndrome. Molecular Genetics and Metabolism, 2001, 72, 67-71.	1.1	84
29	Enzyme replacement therapy for mucopolysaccharidosis VI: evaluation of longâ€term pulmonary function in patients treated with recombinant human Nâ€acetylgalactosamine 4â€sulfatase. Journal of Inherited Metabolic Disease, 2010, 33, 51-60.	3.6	80
30	Apparent underdiagnosis of Cerebrotendinous Xanthomatosis revealed by analysis of ~60,000 human exomes. Molecular Genetics and Metabolism, 2015, 116, 298-304.	1.1	79
31	Studies of collagen synthesis and structure in the differentiation of child abuse from osteogenesis imperfecta. Journal of Pediatrics, 1996, 128, 542-547.	1.8	78
32	Smithâ€“Lemliâ€“Opitz syndrome. Expert Reviews in Molecular Medicine, 2011, 13, e24.	3.9	78
33	Deficits in memory strategy use related to prefrontal dysfunction during early development: Evidence from children with phenylketonuria.. Neuropsychology, 2001, 15, 221-229.	1.3	75
34	Sterol metabolism disorders and neurodevelopmentâ€”an update. Developmental Disabilities Research Reviews, 2013, 17, 197-210.	2.9	74
35	Ophthalmic drops causing coma in an infant. Journal of Pediatrics, 2001, 138, 441-443.	1.8	72
36	Ascorbate decreases Fabry cerebral hyperperfusion suggesting a reactive oxygen species abnormality: An arterial spin tagging study. Journal of Magnetic Resonance Imaging, 2004, 20, 674-683.	3.4	71

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37	Shared familial transmission of autism spectrum and attentionâ€deficit/hyperactivity disorders. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2014, 55, 819-827.	5.2	70
38	Nutrient intakes and physical growth of children with phenylketonuria undergoing nutrition therapy. Journal of the American Dietetic Association, 2003, 103, 1167-1173.	1.1	69
39	Genetics of Familial Hypercholesterolemia. Current Atherosclerosis Reports, 2015, 17, 491.	4.8	68
40	Apolipoprotein Eâ€“low density lipoprotein receptor interaction affects spatial memory retention and brain ApoE levels in an isoform-dependent manner. Neurobiology of Disease, 2014, 64, 150-162.	4.4	67
41	Treatment of Smithâ€“Lemliâ€“Opitz syndrome and other sterol disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 285-294.	1.6	65
42	Multi-domain impact of elosulfase alfa in Morquio A syndrome in the pivotal phase III trial. Molecular Genetics and Metabolism, 2015, 114, 178-185.	1.1	65
43	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, 360-365.	2.4	64
44	Hypomorphic Temperature-Sensitive Alleles of NSDHL Cause CK Syndrome. American Journal of Human Genetics, 2010, 87, 905-914.	6.2	64
45	Importance of surgical history in diagnosing mucopolysaccharidosis type II (Hunter syndrome): Data from the Hunter Outcome Survey. Genetics in Medicine, 2010, 12, 816-822.	2.4	63
46	Long-term management of patients with urea cycle disorders. Journal of Pediatrics, 2001, 138, S56-S61.	1.8	62
47	Age-related working memory impairments in children with prefrontal dysfunction associated with phenylketonuria. Journal of the International Neuropsychological Society, 2002, 8, 1-11.	1.8	62
48	Correlates of language impairment in children with galactosaemia. Journal of Inherited Metabolic Disease, 2008, 31, 524-532.	3.6	59
49	Variability in phenylalanine control predicts IQ and executive abilities in children with phenylketonuria. Molecular Genetics and Metabolism, 2014, 111, 445-451.	1.1	59
50	Neurocognition across the spectrum of mucopolysaccharidosis type I: Age, severity, and treatment. Molecular Genetics and Metabolism, 2015, 116, 61-68.	1.1	59
51	A crossâ€sectional multicenter study ofÂosteogenesis imperfecta in North AmericaÂâ€“Âresults from the linked clinical research centers. Clinical Genetics, 2015, 87, 133-140.	2.0	59
52	Sibling Recurrence Risk and Cross-aggregation of Attention-Deficit/Hyperactivity Disorder and Autism Spectrum Disorder. JAMA Pediatrics, 2019, 173, 147.	6.2	59
53	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.	2.4	58
54	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.	0.5	58

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55	Design, baseline characteristics, and early findings of the MPS VI (mucopolysaccharidosis VI) Clinical Surveillance Program (CSP). Journal of Inherited Metabolic Disease, 2013, 36, 373-384.	3.6	57
56	Nonketotic hyperglycinemia: Atypical clinical and biochemical manifestations. Journal of Pediatrics, 1996, 128, 243-246.	1.8	56
57	Laboratory evaluation of urea cycle disorders. Journal of Pediatrics, 2001, 138, S21-S29.	1.8	56
58	A membrane defect in the pathogenesis of the Smith-Lemli-Opitz syndrome. Journal of Lipid Research, 2006, 47, 134-143.	4.2	56
59	Improved growth and nutrition status in children with methylmalonic or propionic acidemia fed an elemental medical food. Molecular Genetics and Metabolism, 2003, 80, 181-188.	1.1	55
60	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.	4.2	53
61	Smith-Lemli-Opitz Syndrome: The First Malformation Syndrome Associated with Defective Cholesterol Synthesis. Molecular Genetics and Metabolism, 2000, 71, 154-162.	1.1	52
62	Cholesterol storage defect in RSH/Smithâ€“Lemliâ€“Opitz syndrome fibroblasts. Molecular Genetics and Metabolism, 2002, 75, 325-334.	1.1	52
63	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.	2.5	50
64	Treatment of pyruvate carboxylase deficiency with high doses of citrate and aspartate. American Journal of Medical Genetics Part A, 1999, 87, 331-338.	2.4	49
65	Consensus statement from a Conference for the Management of Patients With Urea Cycle Disorders. Journal of Pediatrics, 2001, 138, S1-S5.	1.8	47
66	Fabry Disease in Genetic Counseling Practice: Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 2002, 11, 121-146.	1.6	45
67	Cholesterol supplementation does not improve developmental progress in Smith-Lemli-Opitz syndrome. Journal of Pediatrics, 2004, 144, 783-791.	1.8	45
68	Liquid chromatographyâ€“tandem mass spectrometry determination of plasma 24S-hydroxycholesterol with chromatographic separation of 25-hydroxycholesterol. Analytical Biochemistry, 2008, 381, 151-153.	2.4	44
69	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.	2.7	44
70	Mutations in gamma adducin are associated with inherited cerebral palsy. Annals of Neurology, 2013, 74, 805-814.	5.3	44
71	Effects of Dietary Cholesterol and Simvastatin on Cholesterol Synthesis in Smith-Lemli-Opitz Syndrome. Pediatric Research, 2009, 65, 681-685.	2.3	43
72	Cognitive, medical, and neuroimaging characteristics of attenuated mucopolysaccharidosis type II. Molecular Genetics and Metabolism, 2015, 114, 170-177.	1.1	43

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73	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.	4.2	43
74	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.	1.2	42
75	Deletion of a single mevalonate kinase (Mvk) allele yields a murine model of hyperâ€IgD syndrome. Journal of Inherited Metabolic Disease, 2007, 30, 888-895.	3.6	42
76	â€œI'm fine; I'm just waiting for my diseaseâ€. Neurology, 2011, 77, 522-523.	1.1	42
77	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.	1.8	41
78	Remarkable improvement in adult Leigh syndrome with partial cytochrome c oxidase deficiency. Neurology, 2003, 60, 865-868.	1.1	41
79	Inhibitory Control in Children With Phenylketonuria. Developmental Neuropsychology, 2006, 30, 845-864.	1.4	41
80	A novel mutation in medium chain acyl-CoA dehydrogenase causes sudden neonatal death.. Journal of Clinical Investigation, 1994, 94, 1477-1483.	8.2	41
81	Short/branched-chain acyl-CoA dehydrogenase deficiency due to an IVS3+3A>G mutation that causes exon skipping. Human Genetics, 2006, 118, 680-690.	3.8	40
82	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, 833-836.	1.8	39
83	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.	1.1	39
84	Cesarean delivery is not associated with decreased at-birth fracture rates in osteogenesis imperfecta. Genetics in Medicine, 2016, 18, 570-576.	2.4	39
85	Natural History of Perinatal and Infantile Hypophosphatasia: A Retrospective Study. Journal of Pediatrics, 2019, 209, 116-124.e4.	1.8	39
86	Deficits in memory strategy use related to prefrontal dysfunction during early development: Evidence from children with phenylketonuria.. Neuropsychology, 2001, 15, 221-229.	1.3	39
87	A blood test for cerebrotendinous xanthomatosis with potential for disease detection in newborns. Journal of Lipid Research, 2014, 55, 146-154.	4.2	38
88	Ezetimibe Reduces Plant Sterol Accumulation and Favorably Increases Platelet Count in Sitosterolemia. Journal of Pediatrics, 2015, 166, 125-131.	1.8	38
89	Profiling sterols in cerebrotendinous xanthomatosis: Utility of Girard derivatization and high resolution exact mass LCâ€“ESI-MSn analysis. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2011, 879, 1384-1392.	2.3	37
90	Clinical profile of a male with Rett syndrome. Brain and Development, 2005, 27, S69-S71.	1.1	36

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91	DHCR7 nonsense mutations and characterisation of mRNA nonsense mediated decay in Smith-Lemli-Opitz syndrome. Journal of Medical Genetics, 2005, 42, 350-357.	3.2	36
92	Age-related working memory impairments in children with prefrontal dysfunction associated with phenylketonuria. Journal of the International Neuropsychological Society, 2002, 8, 1-11.	1.8	35
93	Cholesterol Metabolism and Suicidality in Smith-Lemli-Opitz Syndrome Carriers. American Journal of Psychiatry, 2004, 161, 2123-2126.	7.2	35
94	Age-related decline in the microstructural integrity of white matter in children with early- and continuously-treated PKU: A DTI study of the corpus callosumâˆ†. Molecular Genetics and Metabolism, 2010, 99, S41-S46.	1.1	35
95	Assays of plasma dehydrocholesteryl esters and oxysterols from Smith-Lemli-Opitz syndrome patients. Journal of Lipid Research, 2013, 54, 244-253.	4.2	35
96	Interhemispheric Interaction During Childhood: II. Children With Early-Treated Phenylketonuria. Developmental Neuropsychology, 2000, 18, 53-71.	1.4	34
97	Growth characteristics in individuals with osteogenesis imperfecta in North America: results from a multicenter study. Genetics in Medicine, 2019, 21, 275-283.	2.4	34
98	ESI-MS/MS quantification of 7Î±-hydroxy-4-cholesten-3-one facilitates rapid, convenient diagnostic testing for cerebrotendinous xanthomatosis. Clinica Chimica Acta, 2010, 411, 43-48.	1.1	33
99	A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511.	2.0	33
100	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, 2808-2825.	2.9	32
101	A longitudinal study of emotional adjustment, quality of life and adaptive function in attenuated MPS II. Molecular Genetics and Metabolism Reports, 2016, 7, 32-39.	1.1	32
102	Age-related working memory impairments in children with prefrontal dysfunction associated with phenylketonuria. Journal of the International Neuropsychological Society, 2002, 8, 1-11.	1.8	31
103	Fetal demise with Smith-Lemli-Opitz syndrome confirmed by tissue sterol analysis and the absence of measurable 7-dehydrocholesterol ?7-reductase activity in chorionic villi. , 2000, 20, 238-240.		30
104	Inborn Errors of Metabolism (Metabolic Disorders). Pediatrics in Review, 2016, 37, 3-17.	0.4	30
105	Shaken Baby Syndrome--A Forensic Pediatric Response. Pediatrics, 1998, 101, 321-321.	2.1	29
106	Iron status of children with phenylketonuria undergoing nutrition therapy assessed by transferrin receptors. Genetics in Medicine, 2004, 6, 96-101.	2.4	29
107	Processing speed and executive abilities in children with phenylketonuria.. Neuropsychology, 2012, 26, 735-743.	1.3	29
108	In Vitro Incubation with Influenza Virus Primes Human Polymorphonuclear Leukocyte Generation of Superoxide. American Journal of Respiratory Cell and Molecular Biology, 1991, 4, 347-354.	2.9	27

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109	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.	2.4	27
110	Executive Strategic Processing During Verbal Fluency Performance in Children with Phenylketonuria. Child Neuropsychology, 2011, 17, 105-117.	1.3	27
111	Prevalence estimation for monogenic autosomal recessive diseases using population-based genetic data. Human Genetics, 2015, 134, 659-669.	3.8	27
112	Intravenous Fish Oil and Pediatric Intestinal Failureâ€“Associated Liver Disease: Changes in Plasma Phytosterols, Cytokines, and Bile Acids and Erythrocyte Fatty Acids. Journal of Parenteral and Enteral Nutrition, 2017, 42, 014860711770919.	2.6	27
113	Eye findings in 8 children and a spontaneously aborted fetus with RSH/Smith-Lemli-Opitz syndrome. , 1998, 80, 501-505.		26
114	Tandem Mass Spectrometry in Newborn Screening. Journal of Perinatal and Neonatal Nursing, 2004, 18, 41-60.	0.7	26
115	Rhabdomyolysis in the Military: Recognizing Late-Onset Very Long-Chain Acyl Co-A Dehydrogenase Deficiency. Military Medicine, 2006, 171, 657-658.	0.8	26
116	Stem cellâ€“mediated regeneration of the intervertebral disc: cellular and molecular challenges. Neurosurgical Focus, 2008, 24, E21.	2.3	26
117	Newborn Screening for Krabbe Disease: the New York State Model. Pediatric Neurology, 2009, 40, 253-255.	2.1	26
118	Hydrops fetalis: Role of the geneticist. Seminars in Perinatology, 1995, 19, 516-524.	2.5	25
119	Asp187Asn mutation of gelsolin in an American kindred with familial amyloidosis, Finnish type (FAP IV). Human Genetics, 1995, 95, 327-30.	3.8	25
120	Maternal prepregnancy body mass index and offspring attentionâ€deficit/hyperactivity disorder: aÂquasiâ€experimental siblingâ€comparison, populationâ€based design. Journal of Child Psychology and Psychiatry and Allied Disciplines, 2017, 58, 240-247.	5.2	25
121	Effects of Dietary Cholesterol on Plasma Lipoproteins in Smith-Lemli-Opitz Syndrome. Pediatric Research, 2004, 56, 726-732.	2.3	24
122	Combination of diaphragmatic eventration and microphthalmia/anophthalmia is probably nonrandom. American Journal of Medical Genetics Part A, 2002, 108, 45-50.	2.4	23
123	The effects of sterol structure upon sterol esterification. Atherosclerosis, 2010, 208, 155-160.	0.8	23
124	Research challenges in central nervous system manifestations of inborn errors of metabolism. Molecular Genetics and Metabolism, 2011, 102, 326-338.	1.1	22
125	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.	2.7	22
126	Cellular therapy for childhood neurodegenerative disease. Part I: rationale and preclinical studies. Neurosurgical Focus, 2008, 24, E22.	2.3	21

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127	Changes in plasma and urine globotriaosylceramide levels do not predict Fabry disease progression over 1 year of agalsidase alfa. Genetics in Medicine, 2013, 15, 983-989.	2.4	21
128	Cellular therapy for childhood neurodegenerative disease. Part II: clinical trial design and implementation. Neurosurgical Focus, 2008, 24, E23.	2.3	20
129	Response monitoring in children with phenylketonuria.. Neuropsychology, 2009, 23, 130-134.	1.3	20
130	A useful multi-analyte blood test for cerebrotendinous xanthomatosis. Clinical Biochemistry, 2014, 47, 860-863.	1.9	20
131	Potential for Misdiagnosis Due to Lack of Metabolic Derangement in Combined Methylmalonic Aciduria/Hyperhomocysteinemia (cblC) in the Neonate. Journal of Perinatology, 2003, 23, 384-386.	2.0	18
132	Alterations in membrane caveolae and BKCa channel activity in skin fibroblasts in Smithâ€“Lemliâ€“Opitz syndrome. Molecular Genetics and Metabolism, 2011, 104, 346-355.	1.1	18
133	Cerebrotendinous Xanthomatosis: A Treatable Disease With Juvenile Cataracts as a Presenting Sign. JAMA Ophthalmology, 2011, 129, 1087.	2.4	18
134	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, 11-15.	1.1	18
135	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, 1661-1669.	4.2	17
136	Elevated autophagy and mitochondrial dysfunction in the Smithâ€“Lemliâ€“Opitz Syndrome. Molecular Genetics and Metabolism Reports, 2014, 1, 431-442.	1.1	17
137	Breastfeeding Infants with Phenylketonuria in the United States and Canada. Breastfeeding Medicine, 2014, 9, 142-148.	1.7	17
138	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.	2.4	17
139	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.	1.2	17
140	Autosomal dominant transmission of acrodysostosis. Clinical Dysmorphology, 1992, 1, 201???206.	0.3	16
141	Black children deficient in galactose 1-phosphate uridyltransferase: Correlation of activity and immunoreactive protein in erythrocytes and leukocytes. Journal of Pediatrics, 1997, 130, 972-980.	1.8	16
142	The role of evidenceâ€based medicine and clinical trials in rare genetic disorders. Clinical Genetics, 2008, 74, 197-207.	2.0	16
143	Enhanced placental cholesterol efflux by fetal HDL in Smithâ€“Lemliâ€“Opitz syndrome. Molecular Genetics and Metabolism, 2008, 94, 240-247.	1.1	16
144	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, 192-197.	1.2	15

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145	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, 359-364.	2.4	15
146	Perinatal Onset Mevalonate Kinase Deficiency. Pediatric and Developmental Pathology, 2011, 14, 301-306.	1.0	15
147	Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318.	2.4	15
148	Protein Status of Infants with Phenylketonuria Undergoing Nutrition Management. Journal of the American College of Nutrition, 1999, 18, 102-107.	1.8	14
149	Clinical utility of endurance measures for evaluation of treatment in patients with mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). Journal of Pediatric Rehabilitation Medicine, 2010, 3, 119-127.	0.5	14
150	Sitosterolemia Presenting as Pseudohomozygous Familial Hypercholesterolemia. Clinical Medicine and Research, 2016, 14, 103-108.	0.8	14
151	Identification of 7Î±,24-dihydroxy-3-oxocholest-4-en-26-oic and 7Î±,25-dihydroxy-3-oxocholest-4-en-26-oic acids in human cerebrospinal fluid and plasma. Biochimie, 2018, 153, 86-98.	2.6	14
152	Challenges to Breastfeeding Infants With Phenylketonuria. Journal of Pediatric Nursing, 2015, 30, 219-226.	1.5	13
153	Expert opinion on diagnosing, treating and managing patients with cerebrotendinous xanthomatosis (CTX): a modified Delphi study. Orphanet Journal of Rare Diseases, 2021, 16, 353.	2.7	13
154	A Simple PCR-Based Assay Allows Detection of a Common Mutation, IVS8-1Gâ†'C, in DHCR7 in Smith-Lemli-Opitz Syndrome. Genetic Testing and Molecular Biomarkers, 1999, 3, 361-363.	1.7	12
155	Intestinal Absorption of Cholesterol by Patients with Smith-Lemli-Opitz Syndrome. Pediatric Research, 2005, 57, 765-770.	2.3	12
156	Commentary on: â€œNewborn screening for Krabbe Disease: the New York state modelâ€•and â€œThe 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 Yorkâ€• Genetics in Medicine, 2009, 11, 411-413.	2.4	12
157	No evidence for mevalonate shunting in moderately affected children with Smithâ€Łemliâ€Opitz syndrome. Journal of Inherited Metabolic Disease, 2012, 35, 859-869.	3.6	12
158	Corneal Abnormalities in a Mother and Daughter With Focal Dermal Hypoplasia (Goltz-Gorlin) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 222	3.3	11
159	Increased nonsterol isoprenoids, dolichol and ubiquinone, in the Smith-Lemli-Opitz syndrome: effects of dietary cholesterol. Journal of Lipid Research, 2006, 47, 2789-2798.	4.2	11
160	Severe subacute GM2 gangliosidosis caused by an apparently silentHEXA mutation (V324V) that results in aberrant splicing and reducedHEXA mRNA. American Journal of Medical Genetics Part A, 2004, 127A, 158-166.	2.4	10
161	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.	2.3	10
162	Fructose-1,6-diphosphatase Deficiency and Glyceroluria: One Possible Etiology for GIS. Molecular Genetics and Metabolism, 2000, 69, 338-340.	1.1	9

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163	Effect of ezetimibe on low- and high-density lipoprotein subclasses in sitosterolemia. Atherosclerosis, 2017, 260, 27-33.	0.8	9
164	Lowered DHCR7 activity measured by ergosterol conversion in multiple cell types in Smithâ€“Lemliâ€“Opitz syndrome. Molecular Genetics and Metabolism, 2004, 83, 175-183.	1.1	8
165	Challenging Behavior in Smith-Lemli-Opitz Syndrome. Cognitive and Behavioral Neurology, 2013, 26, 23-29.	0.9	8
166	Commitment to Breastfeeding in the Context of Phenylketonuria. JOGNN - Journal of Obstetric, Gynecologic, and Neonatal Nursing, 2015, 44, 726-736.	0.5	8
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