# Robert D Steiner

List of Publications by Year
in descending order



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|---|---|---|---|
| 204 papers | 9,990 citations | 31976 53 h-index | 48315 88 g-index |
| 224 all docs | 224 docs citations | 224 times ranked | 9840 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Real-world effectiveness of burosumab in children with X-linked hypophosphatemic rickets. Pediatric Nephrology, 2022, , 1. | 1.7 | 7 |
| 2 | ClinGenâ€™s Pediatric Actionability Working Group: Clinical actionability of secondary findings from genome-scale sequencing in children and adolescents. Genetics in Medicine, 2022, 24, 1328-1335. | 2.4 | 4 |
| 3 | Author preprint behaviour and nonâ€compliance with journal preprint policies: One biomedical journal's experience. Learned Publishing, 2021, 34, 389. | 1.7 | 0 |
| 4 | Cerebrotendinous xanthomatosis, sitosterolemia, Smith-Lemli-Opitz syndrome and the seminal contributions of Gerald Salen, MD (1935â€“2020). Journal of Clinical Lipidology, 2021, 15, 540-544. | 1.5 | 2 |
| 5 | Genetic counseling and screening of consanguineous couples and their offspring practice resource: Focused Revision. Journal of Genetic Counseling, 2021, 30, 1354-1357. | 1.6 | 1 |
| 6 | 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 |
| 7 | Remediation of ABCG5-Linked Macrothrombocytopenia With Ezetimibe Therapy. Frontiers in Genetics, 2021, 12, 769699. | 2.3 | 1 |
| 8 | 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 |
| 9 | Porencephaly and Intracranial Calcifications in a Neonate. Pediatrics in Review, 2020, 41, 543-545. | 0.4 | 0 |
| 10 | 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 |
| 11 | 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 |
| 12 | Mobility in osteogenesis imperfecta: a multicenter North American study. Genetics in Medicine, 2019, 21, 2311-2318. | 2.4 | 15 |
| 13 | Natural History of Perinatal and Infantile Hypophosphatasia: A Retrospective Study. Journal of Pediatrics, 2019, 209, 116-124.e4. | 1.8 | 39 |
| 14 | 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 |
| 15 | Genetics in Medicine at Twenty. Genetics in Medicine, 2019, 21, 38-40. | 2.4 | 0 |
| 16 | Sibling Recurrence Risk and Cross-aggregation of Attention-Deficit/Hyperactivity Disorder and Autism Spectrum Disorder. JAMA Pediatrics, 2019, 173, 147. | 6.2 | 59 |
| 17 | Cover Image, Volume 39, Issue 11. Human Mutation, 2018, 39, i. | 2.5 | 0 |
| 18 | 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 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | 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 |
| 20 | A multicenter study to evaluate pulmonary function in osteogenesis imperfecta. Clinical Genetics, 2018, 94, 502-511. | 2.0 | 33 |
| 21 | 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 |
| 22 | Bardetâ€“Biedl syndrome: A model for translational research in rare diseases. European Journal of Molecular and Clinical Medicine, 2017, 2, 102. | 0.1 | 5 |
| 23 | Lipid and sterol gene sequence variation in autism and correlates with neurodevelopmental status: A pilot study. European Journal of Molecular and Clinical Medicine, 2017, 2, 137. | 0.1 | 1 |
| 24 | 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 |
| 25 | Thyroid Hormone Status in Sitosterolemia Is Modified by Ezetimibe. Journal of Pediatrics, 2017, 188, 198-204.e1. | 1.8 | 5 |
| 26 | Effect of ezetimibe on low- and high-density lipoprotein subclasses in sitosterolemia. Atherosclerosis, 2017, 260, 27-33. | 0.8 | 9 |
| 27 | Normal IQ is possible in Smithâ€Lemliâ€Opitz syndrome. American Journal of Medical Genetics, Part A, 2017, 173, 2097-2100. | 1.2 | 7 |
| 28 | 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 |
| 29 | Epidemiology, diagnosis, and treatment of cerebrotendinous xanthomatosis (CTX). Journal of Inherited Metabolic Disease, 2017, 40, 771-781. | 3.6 | 127 |
| 30 | Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418. | 3.2 | 133 |
| 31 | 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 |
| 32 | 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 |
| 33 | Sitosterolemia Presenting as Pseudohomozygous Familial Hypercholesterolemia. Clinical Medicine and Research, 2016, 14, 103-108. | 0.8 | 14 |
| 34 | Bisphosphonate therapy for osteogenesis imperfecta. The Cochrane Library, 2016, 2016, CD005088. | 2.8 | 173 |
| 35 | A Pilot Study of the Association of Markers of Cholesterol Synthesis with Disturbed Sleep in Smith-Lemli-Opitz Syndrome. Journal of Developmental and Behavioral Pediatrics, 2016, 37, 424-430. | 1.1 | 4 |
| 36 | Inborn Errors of Metabolism (Metabolic Disorders). Pediatrics in Review, 2016, 37, 3-17. | 0.4 | 30 |

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|---|---|---|---|
| 37 | Cesarean delivery is not associated with decreased at-birth fracture rates in osteogenesis imperfecta. Genetics in Medicine, 2016, 18, 570-576. | 2.4 | 39 |
| 38 | Commitment to Breastfeeding in the Context of Phenylketonuria. JOGNN - Journal of Obstetric, Gynecologic, and Neonatal Nursing, 2015, 44, 726-736. | 0.5 | 8 |
| 39 | Apparent underdiagnosis of Cerebrotendinous Xanthomatosis revealed by analysis of ~60,000 human exomes. Molecular Genetics and Metabolism, 2015, 116, 298-304. | 1.1 | 79 |
| 40 | 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 |
| 41 | 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 |
| 42 | Genetics of Familial Hypercholesterolemia. Current Atherosclerosis Reports, 2015, 17, 491. | 4.8 | 68 |
| 43 | Cognitive, medical, and neuroimaging characteristics of attenuated mucopolysaccharidosis type II. Molecular Genetics and Metabolism, 2015, 114, 170-177. | 1.1 | 43 |
| 44 | Neurocognition across the spectrum of mucopolysaccharidosis type I: Age, severity, and treatment. Molecular Genetics and Metabolism, 2015, 116, 61-68. | 1.1 | 59 |
| 45 | Prevalence estimation for monogenic autosomal recessive diseases using population-based genetic data. Human Genetics, 2015, 134, 659-669. | 3.8 | 27 |
| 46 | 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 |
| 47 | 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 |
| 48 | Ezetimibe Reduces Plant Sterol Accumulation and Favorably Increases Platelet Count in Sitosterolemia. Journal of Pediatrics, 2015, 166, 125-131. | 1.8 | 38 |
| 49 | Challenges to Breastfeeding Infants With Phenylketonuria. Journal of Pediatric Nursing, 2015, 30, 219-226. | 1.5 | 13 |
| 50 | Bisphosphonate therapy for osteogenesis imperfecta. , 2014, , CD005088. | | 93 |
| 51 | Elevated autophagy and mitochondrial dysfunction in the Smithâ€“Lemliâ€“Opitz Syndrome. Molecular Genetics and Metabolism Reports, 2014, 1, 431-442. | 1.1 | 17 |
| 52 | Breastfeeding Infants with Phenylketonuria in the United States and Canada. Breastfeeding Medicine, 2014, 9, 142-148. | 1.7 | 17 |
| 53 | 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 |
| 54 | A blood test for cerebrotendinous xanthomatosis with potential for disease detection in newborns. Journal of Lipid Research, 2014, 55, 146-154. | 4.2 | 38 |

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|---|---|---|---|
| 55 | A useful multi-analyte blood test for cerebrotendinous xanthomatosis. Clinical Biochemistry, 2014, 47, 860-863. | 1.9 | 20 |
| 56 | 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 |
| 57 | Feeding Impairments Associated with Plasma Sterols in Smith-Lemli-Opitz Syndrome. Journal of Pediatrics, 2014, 165, 836-841.e1. | 1.8 | 7 |
| 58 | Variability in phenylalanine control predicts IQ and executive abilities in children with phenylketonuria. Molecular Genetics and Metabolism, 2014, 111, 445-451. | 1.1 | 59 |
| 59 | A US perspective on newborn screening: a powerful tool for prevention. Expert Opinion on Orphan Drugs, 2014, 2, 1151-1157. | 0.8 | 0 |
| 60 | Reducing circulating levels of plant sterols by ezetimibe favorably increases platelet counts in sitosterolemia patients (117.7). FASEB Journal, 2014, 28, 117.7. | 0.5 | 0 |
| 61 | Risedronate in children with osteogenesis imperfecta: a randomised, double-blind, placebo-controlled trial. Lancet, The, 2013, 382, 1424-1432. | 13.7 | 158 |
| 62 | 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 |
| 63 | Sterol metabolism disorders and neurodevelopmentâ€”an update. Developmental Disabilities Research Reviews, 2013, 17, 197-210. | 2.9 | 74 |
| 64 | Executive Response Monitoring and Inhibitory Control in Children With Phenylketonuria: Effects of Expectancy. Developmental Neuropsychology, 2013, 38, 139-152. | 1.4 | 6 |
| 65 | Central nervous system stem cell transplantation for children with neuronal ceroid lipofuscinosis. Journal of Neurosurgery: Pediatrics, 2013, 11, 643-652. | 1.3 | 117 |
| 66 | Prospects for stem cell therapy in neuronal ceroid lipofuscinosis. Regenerative Medicine, 2013, 8, 527-529. | 1.7 | 4 |
| 67 | Challenging Behavior in Smith-Lemli-Opitz Syndrome. Cognitive and Behavioral Neurology, 2013, 26, 23-29. | 0.9 | 8 |
| 68 | Assays of plasma dehydrocholesteryl esters and oxysterols from Smith-Lemli-Opitz syndrome patients. Journal of Lipid Research, 2013, 54, 244-253. | 4.2 | 35 |
| 69 | 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 |
| 70 | Mutations in gamma adducin are associated with inherited cerebral palsy. Annals of Neurology, 2013, 74, 805-814. | 5.3 | 44 |
| 71 | 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 |
| 72 | Plant sterol whole body pool size in sitosterolemia is modulated by ezetimibe. FASEB Journal, 2013, 27, 373.2. | 0.5 | 0 |

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|---|---|---|---|
| 73 | PHENOTYPIC VARIABILITY IN INDIVIDUALS WITH TYPE V OSTEOGENESIS IMPERFECTA WITH IDENTICAL IFITM5 MUTATIONS. The Journal of Rare Disorders, 2013, 1, 37-42. | 1.5 | 7 |
| 74 | Processing speed and executive abilities in children with phenylketonuria.. Neuropsychology, 2012, 26, 735-743. | 1.3 | 29 |
| 75 | 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 |
| 76 | 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 |
| 77 | 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 |
| 78 | Assessment of whole body cholesterol pool size in Smithâ€Łemliâ€•Opitz syndrome children using liquid chromatography tandem mass spectrometry. FASEB Journal, 2012, 26, 242.1. | 0.5 | 0 |
| 79 | Executive Strategic Processing During Verbal Fluency Performance in Children with Phenylketonuria. Child Neuropsychology, 2011, 17, 105-117. | 1.3 | 27 |
| 80 | Research challenges in central nervous system manifestations of inborn errors of metabolism. Molecular Genetics and Metabolism, 2011, 102, 326-338. | 1.1 | 22 |
| 81 | 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 |
| 82 | Cerebrotendinous Xanthomatosis: A Treatable Disease With Juvenile Cataracts as a Presenting Sign. JAMA Ophthalmology, 2011, 129, 1087. | 2.4 | 18 |
| 83 | 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 |
| 84 | â€œI'm fine; I'm just waiting for my diseaseâ€• Neurology, 2011, 77, 522-523. | 1.1 | 42 |
| 85 | 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 |
| 86 | 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 |
| 87 | Smithâ€“Lemliâ€“Opitz syndrome. Expert Reviews in Molecular Medicine, 2011, 13, e24. | 3.9 | 78 |
| 88 | Perinatal Onset Mevalonate Kinase Deficiency. Pediatric and Developmental Pathology, 2011, 14, 301-306. | 1.0 | 15 |
| 89 | 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 |
| 90 | Hypomorphic Temperature-Sensitive Alleles of NSDHL Cause CK Syndrome. American Journal of Human Genetics, 2010, 87, 905-914. | 6.2 | 64 |

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|---|---|---|---|
| 91 | 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 |
| 92 | 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 |
| 93 | 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 |
| 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 callosumâ€†. Molecular Genetics and Metabolism, 2010, 99, S41-S46. | 1.1 | 35 |
| 95 | The effects of sterol structure upon sterol esterification. Atherosclerosis, 2010, 208, 155-160. | 0.8 | 23 |
| 96 | 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 |
| 97 | Effects of Dietary Cholesterol and Simvastatin on Cholesterol Synthesis in Smith-Lemli-Opitz Syndrome. Pediatric Research, 2009, 65, 681-685. | 2.3 | 43 |
| 98 | 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 |
| 99 | 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 |
| 100 | Osteogenesis imperfecta: Recent findings shed new light on this once well-understood condition. Genetics in Medicine, 2009, 11, 375-385. | 2.4 | 141 |
| 101 | Agalsidase Alfa and Kidney Dysfunction in Fabry Disease. Journal of the American Society of Nephrology: JASN, 2009, 20, 1132-1139. | 6.1 | 148 |
| 102 | Plasma Plant Sterol Levels Do Not Reflect Cholesterol Absorption in Children with Smith-Lemli-Opitz Syndrome. Journal of Pediatrics, 2009, 154, 557-561.e1. | 1.8 | 6 |
| 103 | Lysosomal Disease Networkâ€™s â€œWORLD Symposium 2009â€• Molecular Genetics and Metabolism, 2009, 96, S3-S5. | 1.1 | 0 |
| 104 | Newborn Screening for Krabbe Disease: the New York State Model. Pediatric Neurology, 2009, 40, 253-255. | 2.1 | 26 |
| 105 | Response monitoring in children with phenylketonuria.. Neuropsychology, 2009, 23, 130-134. | 1.3 | 20 |
| 106 | Correlates of language impairment in children with galactosaemia. Journal of Inherited Metabolic Disease, 2008, 31, 524-532. | 3.6 | 59 |
| 107 | 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 |
| 108 | The role of evidenceâ€based medicine and clinical trials in rare genetic disorders. Clinical Genetics, 2008, 74, 197-207. | 2.0 | 16 |

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|---|---|---|---|
| 109 | Enhanced placental cholesterol efflux by fetal HDL in Smithâ€“Lemliâ€“Opitz syndrome. Molecular Genetics and Metabolism, 2008, 94, 240-247. | 1.1 | 16 |
| 110 | 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 |
| 111 | Bisphosphonate therapy for osteogenesis imperfecta. , 2008, , CD005088. | | 87 |
| 112 | Stem cellâ€“mediated regeneration of the intervertebral disc: cellular and molecular challenges. Neurosurgical Focus, 2008, 24, E21. | 2.3 | 26 |
| 113 | Cellular therapy for childhood neurodegenerative disease. Part II: clinical trial design and implementation. Neurosurgical Focus, 2008, 24, E23. | 2.3 | 20 |
| 114 | Cellular therapy for childhood neurodegenerative disease. Part I: rationale and preclinical studies. Neurosurgical Focus, 2008, 24, E22. | 2.3 | 21 |
| 115 | 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 |
| 116 | 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 |
| 117 | Effects of dietary cholesterol and simvastatin on cholesterol absorption and synthesis (CAS) in Smithâ€Lemliâ€Opitz syndrome (SLOS). FASEB Journal, 2007, 21, A340. | 0.5 | 1 |
| 118 | Pompe disease diagnosis and management guideline. Genetics in Medicine, 2006, 8, 267-288. | 2.4 | 473 |
| 119 | Rhabdomyolysis in the Military: Recognizing Late-Onset Very Long-Chain Acyl Co-A Dehydrogenase Deficiency. Military Medicine, 2006, 171, 657-658. | 0.8 | 26 |
| 120 | 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 |
| 121 | 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 |
| 122 | Newborn Screening: Toward a Uniform Screening Panel and Systemâ€”Executive Summary. Pediatrics, 2006, 117, S296-S307. | 2.1 | 386 |
| 123 | Inhibitory Control in Children With Phenylketonuria. Developmental Neuropsychology, 2006, 30, 845-864. | 1.4 | 41 |
| 124 | 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 |
| 125 | A membrane defect in the pathogenesis of the Smith-Lemli-Opitz syndrome. Journal of Lipid Research, 2006, 47, 134-143. | 4.2 | 56 |
| 126 | Clinical profile of a male with Rett syndrome. Brain and Development, 2005, 27, S69-S71. | 1.1 | 36 |

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|---|---|---|---|
| 127 | 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 |
| 128 | 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 |
| 129 | Intestinal Absorption of Cholesterol by Patients with Smith-Lemli-Opitz Syndrome. Pediatric Research, 2005, 57, 765-770. | 2.3 | 12 |
| 130 | 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 |
| 131 | Diagnosis and Treatment of Children with Suspected Metabolic Disease. , 2005, , 1866-1875. | | 3 |
| 132 | Lysosomal Storage Disorders. , 2005, , 1007-1012. | | 0 |
| 133 | Iron status of children with phenylketonuria undergoing nutrition therapy assessed by transferrin receptors. Genetics in Medicine, 2004, 6, 96-101. | 2.4 | 29 |
| 134 | Cholesterol Metabolism and Suicidality in Smith-Lemli-Opitz Syndrome Carriers. American Journal of Psychiatry, 2004, 161, 2123-2126. | 7.2 | 35 |
| 135 | Effects of Dietary Cholesterol on Plasma Lipoproteins in Smith-Lemli-Opitz Syndrome. Pediatric Research, 2004, 56, 726-732. | 2.3 | 24 |
| 136 | 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 |
| 137 | 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 |
| 138 | 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 |
| 139 | Cholesterol supplementation does not improve developmental progress in Smith-Lemli-Opitz syndrome. Journal of Pediatrics, 2004, 144, 783-791. | 1.8 | 45 |
| 140 | Enzyme replacement therapy in mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). Journal of Pediatrics, 2004, 144, 574-580. | 1.8 | 267 |
| 141 | Tandem Mass Spectrometry in Newborn Screening. Journal of Perinatal and Neonatal Nursing, 2004, 18, 41-60. | 0.7 | 26 |
| 142 | Not so rare: errors of metabolism during the neonatal period. Newborn and Infant Nursing Reviews, 2003, 3, 143-155. | 0.4 | 3 |
| 143 | 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 |
| 144 | Expanding the limits of the Fryns syndrome. American Journal of Medical Genetics Part A, 2003, 122A, 89-90. | 2.4 | 0 |

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|---|---|---|---|
| 145 | 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 |
| 146 | Metabolic disease and sudden unexpected death. Journal of Pediatrics, 2003, 142, 357. | 1.8 | 0 |
| 147 | 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 |
| 148 | Missense Mutations in CRELD1 Are Associated with Cardiac Atrioventricular Septal Defects. American Journal of Human Genetics, 2003, 72, 1047-1052. | 6.2 | 189 |
| 149 | Remarkable improvement in adult Leigh syndrome with partial cytochrome c oxidase deficiency. Neurology, 2003, 60, 865-868. | 1.1 | 41 |
| 150 | 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 |
| 151 | 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 |
| 152 | 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 |
| 153 | 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 |
| 154 | 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 |
| 155 | Cholesterol storage defect in RSH/Smithâ€“Lemliâ€“Opitz syndrome fibroblasts. Molecular Genetics and Metabolism, 2002, 75, 325-334. | 1.1 | 52 |
| 156 | Combination of diaphragmatic eventration and microphthalmia/anophthalmia is probably nonrandom. American Journal of Medical Genetics Part A, 2002, 108, 45-50. | 2.4 | 23 |
| 157 | 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 |
| 158 | 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 |
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