# Susan A Berry

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/1423075/publications.pdf

Version: 2024-02-01

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|---|---|---|---|
| 94 papers | 4,836 citations | 147801 31 h-index | 106344 65 g-index |
| 96 all docs | 96 docs citations | 96 times ranked | 5385 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Cockayne syndrome: Review of 140 cases. American Journal of Medical Genetics Part A, 1992, 42, 68-84. | 2.4 | 749 |
| 2 | Phenylalanine hydroxylase deficiency: diagnosis and management guideline. Genetics in Medicine, 2014, 16, 188-200. | 2.4 | 486 |
| 3 | Survival after Treatment with Phenylacetate and Benzoate for Urea-Cycle Disorders. New England Journal of Medicine, 2007, 356, 2282-2292. | 27.0 | 320 |
| 4 | Increased MECP2 gene copy number as the result of genomic duplication in neurodevelopmentally delayed males. Genetics in Medicine, 2006, 8, 784-792. | 2.4 | 245 |
| 5 | Mutations in transcriptional regulator ATRX establish the functional significance of a PHD-like domain. Nature Genetics, 1997, 17, 146-148. | 21.4 | 196 |
| 6 | Klippel-Trenaunay syndrome. , 1998, 79, 319-326. | | 190 |
| 7 | A Common Mutation Is Associated with a Mild, Potentially Asymptomatic Phenotype in Patients with Isovaleric Acidemia Diagnosed by Newborn Screening. American Journal of Human Genetics, 2004, 75, 1136-1142. | 6.2 | 145 |
| 8 | Ten novelFBN2mutations in congenital contractural arachnodactyly: Delineation of the molecular pathogenesis and clinical phenotype. Human Mutation, 2002, 19, 39-48. | 2.5 | 114 |
| 9 | Growth Hormone Induction of Hepatic Serine Protease Inhibitor 2.1 Transcription Is Mediated by a Stat5-related Factor Binding Synergistically to Two Î³-Activated Sites. Journal of Biological Chemistry, 1995, 270, 24903-24910. | 3.4 | 113 |
| 10 | Newborn screening 50 years later: access issues faced by adults with PKU. Genetics in Medicine, 2013, 15, 591-599. | 2.4 | 97 |
| 11 | Exome sequencing identifies ACSF3 as a cause of combined malonic and methylmalonic aciduria. Nature Genetics, 2011, 43, 883-886. | 21.4 | 89 |
| 12 | Jarchoâ€Łevin syndrome: Four new cases and classification of subtypes. American Journal of Medical Genetics Part A, 1991, 40, 264-270. | 2.4 | 86 |
| 13 | Expanded newborn screening identifies maternal primary carnitine deficiency. Molecular Genetics and Metabolism, 2007, 90, 441-445. | 1.1 | 86 |
| 14 | Ammonia control and neurocognitive outcome among urea cycle disorder patients treated with glycerol phenylbutyrate. Hepatology, 2013, 57, 2171-2179. | 7.3 | 83 |
| 15 | Returning a Research Participant's Genomic Results to Relatives: Analysis and Recommendations. Journal of Law, Medicine and Ethics, 2015, 43, 440-463. | 0.9 | 81 |
| 16 | Phase 2 comparison of a novel ammonia scavenging agent with sodium phenylbutyrate in patients with urea cycle disorders: Safety, pharmacokinetics and ammonia control. Molecular Genetics and Metabolism, 2010, 100, 221-228. | 1.1 | 78 |
| 17 | Inhibition of growth hormone action in models of inflammation. American Journal of Physiology - Cell Physiology, 2000, 279, C1906-C1917. | 4.6 | 76 |
| 18 | Prospective Diagnosis of 2-Methylbutyryl-CoA Dehydrogenase Deficiency in the Hmong Population by Newborn Screening Using Tandem Mass Spectrometry. Pediatrics, 2003, 112, 74-78. | 2.1 | 67 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Single central incisor in familial holoprosencephaly. Journal of Pediatrics, 1984, 104, 877-880. | 1.8 | 63 |
| 20 | A report on stateâ€wide implementation of newborn screening for Xâ€linked Adrenoleukodystrophy. American Journal of Medical Genetics, Part A, 2019, 179, 1205-1213. | 1.2 | 56 |
| 21 | Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849. | 6.2 | 55 |
| 22 | Biallelic Mutations in FUT8 Cause a Congenital Disorder of Glycosylation with Defective Fucosylation. American Journal of Human Genetics, 2018, 102, 188-195. | 6.2 | 49 |
| 23 | Partial monosomy of chromosome 1p36.3: Characterization of the critical region and delineation of a syndrome. American Journal of Medical Genetics Part A, 1995, 59, 467-475. | 2.4 | 48 |
| 24 | Newborn Screening. Clinics in Perinatology, 2015, 42, 441-453. | 2.1 | 44 |
| 25 | Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128. | 5.3 | 42 |
| 26 | Ammonia Control in Children Ages 2 Months through 5 Years with Urea Cycle Disorders: Comparison of Sodium Phenylbutyrate and Glycerol Phenylbutyrate. Journal of Pediatrics, 2013, 162, 1228-1234.e1. | 1.8 | 40 |
| 27 | Six New Mutations in the Ornithine Transcarbamylase Gene Detected by Single-Strand Conformational Polymorphism. Pediatric Research, 1992, 32, 600-604. | 2.3 | 38 |
| 28 | Brain anomalies, retardation of mentality and growth, ectodermal dysplasia, skeletal malformations, Hirschsprung disease, ear deformity and deafness, eye hypoplasia, cleft palate, cryptorchidism, and kidney dysplasia/hypoplasia BRESEK/BRESHECK: New X-link. , 1997, 68, 386-390. | | 38 |
| 29 | Glycerol phenylbutyrate treatment in children with urea cycle disorders: Pooled analysis of short and long-term ammonia control and outcomes. Molecular Genetics and Metabolism, 2014, 112, 17-24. | 1.1 | 38 |
| 30 | A novel microdeletion/microduplication syndrome of 19p13.13. Genetics in Medicine, 2010, 12, 503-511. | 2.4 | 37 |
| 31 | Report of a patient with a constitutional missense mutation in SMARCB1, Coffinâ€“Siris phenotype, and schwannomatosis. American Journal of Medical Genetics, Part A, 2015, 167, 3186-3191. | 1.2 | 35 |
| 32 | Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106. | 3.6 | 35 |
| 33 | Regulation of Spi 2.1 and 2.2 gene expression after turpentine inflammation: discordant responses to IL-6. American Journal of Physiology - Cell Physiology, 1999, 276, C1374-C1382. | 4.6 | 31 |
| 34 | What questions should newborn screening long-term follow-up be able to answer? A statement of the US Secretary for Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children. Genetics in Medicine, 2011, 13, 861-865. | 2.4 | 31 |
| 35 | Mosaicism for deletion 1p36.33 in a patient with obesity and hyperphagia. , 1997, 70, 409-412. | | 30 |
| 36 | Blood ammonia and glutamine as predictors of hyperammonemic crises in patients with urea cycle disorder. Genetics in Medicine, 2015, 17, 561-568. | 2.4 | 30 |

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|---|---|---|---|
| 37 | 3-Hydroxyisovalerylcarnitine in patients with deficiency of 3-methylcrotonyl CoA carboxylase. Clinica Chimica Acta, 1995, 240, 35-51. | 1.1 | 28 |
| 38 | Evaluation of newborn screening bloodspot-based genetic testing as second tier screen for bedside newborn hearing screening. Genetics in Medicine, 2011, 13, 1006-1010. | 2.4 | 28 |
| 39 | Yin-yang 1 and Glucocorticoid Receptor Participate in the Stat5-mediated Growth Hormone Response of the Serine Protease Inhibitor 2.1 Gene. Journal of Biological Chemistry, 2000, 275, 8114-8120. | 3.4 | 27 |
| 40 | Agenesis of the penis, scrotal raphe, and anus in one of monoamniotic twins. Teratology, 1984, 29, 173-176. | 1.6 | 26 |
| 41 | Infant with multiple congenital anomalies and deletion (9)(q34.3). American Journal of Medical Genetics Part A, 1994, 51, 140-142. | 2.4 | 26 |
| 42 | Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871. | 3.7 | 26 |
| 43 | Duplication of 7p: Further delineation of the phenotype and restriction of the critical region to the distal part of the short arm. , 1996, 61, 21-25. |  | 25 |
| 44 | Social skills in children with RASopathies: a comparison of Noonan syndrome and neurofibromatosis type 1. Journal of Neurodevelopmental Disorders, 2018, 10, 21. | 3.1 | 25 |
| 45 | Variants in TCF20 in neurodevelopmental disability: description of 27 new patients and review of literature. Genetics in Medicine, 2019, 21, 2036-2042. | 2.4 | 23 |
| 46 | Lack of IL7RÎ± expression in T cells is a hallmark of T-cell immunodeficiency in Schimke immuno-osseous dysplasia (SIOD). Clinical Immunology, 2015, 161, 355-365. | 3.2 | 22 |
| 47 | Clinical and genetic spectrum of AMPD2-related pontocerebellar hypoplasia type 9. European Journal of Human Genetics, 2018, 26, 695-708. | 2.8 | 22 |
| 48 | Outcomes of four patients with homocysteine remethylation disorders detected by newborn screening. Genetics in Medicine, 2016, 18, 162-167. | 2.4 | 21 |
| 49 | Two siblings with biotin-resistant 3-methylcrotonyl-coenzyme A carboxylase deficiency. Journal of Pediatrics, 1989, 115, 110-113. | 1.8 | 20 |
| 50 | Parental Permission for Pilot Newborn Screening Research: Guidelines From the NBSTRN. Pediatrics, 2014, 133, e410-e417. | 2.1 | 20 |
| 51 | Developing interactions with industry in rare diseases: lessons learned and continuing challenges. Genetics in Medicine, 2020, 22, 219-226. | 2.4 | 20 |
| 52 | A framework for assessing outcomes from newborn screening: on the road to measuring its promise. Molecular Genetics and Metabolism, 2016, 118, 221-229. | 1.1 | 19 |
| 53 | Differential Endocrine Regulation ofÎ±2U-Globulin Messenger Ribonucleic Acid Activity: Effect of Age at Hypophysectomy*. Endocrinology, 1986, 119, 600-605. | 2.8 | 18 |
| 54 | 221 newborn-screened neonates with medium-chain acyl-coenzyme A dehydrogenase deficiency: Findings from the Inborn Errors of Metabolism Collaborative. Molecular Genetics and Metabolism, 2016, 119, 75-82. | 1.1 | 18 |

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|---|---|---|---|
| 55 | Outcomes of cases with 3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency - Report from the Inborn Errors of Metabolism Information System. Molecular Genetics and Metabolism, 2016, 118, 15-20. | 1.1 | 18 |
| 56 | Foundation of the Newborn Screening Translational Research Network and its tools for research. Genetics in Medicine, 2019, 21, 1271-1279. | 2.4 | 18 |
| 57 | Insurance coverage of medical foods for treatment of inherited metabolic disorders. Genetics in Medicine, 2013, 15, 978-982. | 2.4 | 17 |
| 58 | Ontogenesis of Growth Hormone (GH)-Responsive Hepatic Gene Products: Lack of Correlation with Hepatic GH Receptor Content*. Endocrinology, 1986, 119, 2290-2296. | 2.8 | 16 |
| 59 | Clinical and biochemical heterogeneity in females of a large pedigree with ornithine transcarbamylase deficiency due to the R141Q mutation. , 1996, 66, 311-315. | | 16 |
| 60 | Hepatic Growth Hormone Signaling in the Late Gestation Fetal Rat*. Endocrinology, 2000, 141, 3527-3533. | 2.8 | 16 |
| 61 | Ophthalmic manifestations of Wolfâ€“Hirschhorn syndrome. Journal of AAPOS, 2004, 8, 345-348. | 0.3 | 16 |
| 62 | Safety and efficacy of glycerol phenylbutyrate for management of urea cycle disorders in patients aged 2 months to 2 years. Molecular Genetics and Metabolism, 2017, 122, 46-53. | 1.1 | 16 |
| 63 | Long-term follow-up of newborn screening patients. Genetics in Medicine, 2010, 12, S267-S268. | 2.4 | 14 |
| 64 | Medical Foods for Inborn Errors of Metabolism: History, Current Status, and Critical Need. Pediatrics, 2020, 145, . | 2.1 | 14 |
| 65 | Hormonal regulation of serum alpha1-antitrypsin and hepatic alpha1-antitrypsin mRNA in rats. Biochemical and Biophysical Research Communications, 1987, 147, 936-941. | 2.1 | 13 |
| 66 | Effects of elevated serum insulinlike growth factor-II on growth hormone and insulinlike growth factor-I mRNA and secretion. Metabolism: Clinical and Experimental, 1989, 38, 57-62. | 3.4 | 13 |
| 67 | The inborn errors of metabolism information system: A project of the Region 4 Genetics Collaborative Priority 2 Workgroup. Genetics in Medicine, 2010, 12, S215-S219. | 2.4 | 13 |
| 68 | Improving recommendations for genomic medicine: building an evolutionary process from clinical practice advisory documents to guidelines. Genetics in Medicine, 2019, 21, 2431-2438. | 2.4 | 13 |
| 69 | Severe Methylenetetrahydrofolate Reductase (MTHFR) Deficiency: A Case Report of Nonclassical Homocystinuria. Journal of Child Neurology, 2008, 23, 823-828. | 1.4 | 12 |
| 70 | Self-reported treatment-associated symptoms among patients with urea cycle disorders participating in glycerol phenylbutyrate clinical trials. Molecular Genetics and Metabolism, 2015, 116, 29-34. | 1.1 | 12 |
| 71 | Comparison of Methods of Initial Ascertainment in 58 Cases of Propionic Acidemia Enrolled in the Inborn Errors of Metabolism Information System Reveals Significant Differences in Time to Evaluation and Symptoms at Presentation. Journal of Pediatrics, 2017, 180, 200-205.e8. | 1.8 | 12 |
| 72 | Glycerol phenylbutyrate efficacy and safety from an open label study in pediatric patients under 2Âmonths of age with urea cycle disorders. Molecular Genetics and Metabolism, 2021, 132, 19-26. | 1.1 | 11 |

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|---|---|---|---|
| 73 | Substantial Induction of a New Serum Protein by Growth Hormone: Physiochemical and Physiological Characterization*. Endocrinology, 1984, 115, 1164-1170. | 2.8 | 10 |
| 74 | Newborn blood spot screening and genetic services: A survey of Minnesota primary care physicians. Genetics in Medicine, 2005, 7, 564-570. | 2.4 | 10 |
| 75 | Long-term safety and efficacy of glycerol phenylbutyrate for the management of urea cycle disorder patients. Molecular Genetics and Metabolism, 2019, 127, 336-345. | 1.1 | 10 |
| 76 | Methionine synthase deficiency: Variable clinical presentation and benefit of early diagnosis and treatment. Journal of Inherited Metabolic Disease, 2022, 45, 157-168. | 3.6 | 10 |
| 77 | Inborn Errors of Metabolism Collaborative: large-scale collection of data on long-term follow-up for newborn-screened conditions. Genetics in Medicine, 2016, 18, 1276-1281. | 2.4 | 9 |
| 78 | Pragmatic Tools for Sharing Genomic Research Results with the Relatives of Living and Deceased Research Participants. Journal of Law, Medicine and Ethics, 2018, 46, 87-109. | 0.9 | 9 |
| 79 | Emotional functioning among children with neurofibromatosis type 1 or Noonan syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2433-2446. | 1.2 | 9 |
| 80 | Treatment of mucopolysaccharidosis type II (Hunter syndrome): a Delphi derived practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2020, 22, 1735-1742. | 2.4 | 8 |
| 81 | Pharmacokinetics of glycerol phenylbutyrate in pediatric patients 2â€¯months to 2â€¯years of age with urea cycle disorders. Molecular Genetics and Metabolism, 2018, 125, 251-257. | 1.1 | 7 |
| 82 | Deletion and duplication of 11p13â€¯11p14: Reciprocal aberrations derived from a paternal insertion. American Journal of Medical Genetics, Part A, 2011, 155, 2775-2783. | 1.2 | 6 |
| 83 | Necrotizing Enterocolitis in Two Siblings and an Unrelated Infant with Overlapping Chromosome 6q25 Deletions. Molecular Syndromology, 2018, 9, 141-148. | 0.8 | 5 |
| 84 | Hepatic Growth Hormone Signaling in the Late Gestation Fetal Rat. Endocrinology, 2000, 141, 3527-3533. | 2.8 | 5 |
| 85 | Definition of a high affinity growth hormone DNA response element. Molecular and Cellular Endocrinology, 1999, 150, 151-159. | 3.2 | 4 |
| 86 | Liver transplant as a curative treatment in a pediatric patient with classic homocystinuria: A case report. American Journal of Medical Genetics, Part A, 2021, 185, 1247-1250. | 1.2 | 4 |
| 87 | Regulation of purine metabolism: A comparative study of the kinetic properties of adenylosuccinate synthetases from various sources. Comparative Biochemistry and Physiology Part B: Comparative Biochemistry, 1977, 58, 63-65. | 0.2 | 3 |
| 88 | An Exploration of Genetic Test Utilization, Genetic Counseling, and Consanguinity within the Inborn Errors of Metabolism Collaborative (IBEMC). Journal of Genetic Counseling, 2017, 26, 1238-1243. | 1.6 | 3 |
| 89 | Rare presentation of <scp>FDX2â€related</scp> disorder and untargeted global metabolomics findings. American Journal of Medical Genetics, Part A, 2022, 188, 1239-1244. | 1.2 | 3 |
| 90 | Management Principles for Acute Illness in Patients With Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency. Pediatrics, 2021, 147, . | 2.1 | 2 |

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|---|---|---|---|
| 91 | Using Long-Term Follow-Up Data to Classify Genetic Variants in Newborn Screened Conditions. Frontiers in Genetics, 0, 13, . | 2.3 | 2 |
| 92 | Growth Hormone Action in Hypothyroid Infant Rats. Pediatric Research, 2000, 47, 250-250. | 2.3 | 1 |
| 93 | Mary Ella Mascia Pierpont: Geneticist, scientist, mentor, friend (1945â€“2020). American Journal of Medical Genetics, Part A, 2021, 185, 319-323. | 1.2 | 0 |
| 94 | Cobalamin J disease detected on newborn screening: Novel variant and normal neurodevelopmental course. American Journal of Medical Genetics, Part A, 2021, 185, 1870-1874. | 1.2 | 0 |