# Alberto B Burlina

List of Publications by Year
in descending order

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| 80 | 5,196 | 117625 | 91884 |
|---|---|---|---|
| papers | citations | 34 h-index | 69 g-index |

| 86 | 86 | 86 | 4501 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Newborn screening of mucopolysaccharidosis type I. Critical Reviews in Clinical Laboratory Sciences, 2022, 59, 257-277. | 6.1 | 2 |
| 2 | Atherogenic lipid profile in patients with Niemann-Pick disease type B: What treatment strategies?. Journal of Clinical Lipidology, 2022, , . | 1.5 | 0 |
| 3 | Disorders of Phenylalanine and Tetrahydrobiopterin Metabolism. , 2022, , 331-351. | | 2 |
| 4 | A New Approach to Objectively Evaluate Inherited Metabolic Diseases for Inclusion on Newborn Screening Programmes. International Journal of Neonatal Screening, 2022, 8, 25. | 3.2 | 10 |
| 5 | Application of a Novel Algorithm for Expanding Newborn Screening for Inherited Metabolic Disorders across Europe. International Journal of Neonatal Screening, 2022, 8, 20. | 3.2 | 10 |
| 6 | Efficacy and safety of empagliflozin in glycogen storage disease type Ib: Data from an international questionnaire. Genetics in Medicine, 2022, 24, 1781-1788. | 2.4 | 29 |
| 7 | A new strategy of desensitization in mucopolysaccharidosis type II disease treated with idursulfase therapy: A case report and review of the literature. Molecular Genetics and Metabolism Reports, 2022, 31, 100878. | 1.1 | 1 |
| 8 | Experience of the NPC Brazil Network with a Comprehensive Program for the Screening and Diagnosis of Niemann-Pick Disease Type C. International Journal of Neonatal Screening, 2022, 8, 39. | 3.2 | 0 |
| 9 | Bone disease in early detected Gaucher Type I disease: A case report. JIMD Reports, 2022, 63, 414-419. | 1.5 | 1 |
| 10 | Healthâ€related quality of life in paediatric patients with intoxicationâ€type inborn errors of metabolism: Analysis of an international data set. Journal of Inherited Metabolic Disease, 2021, 44, 215-225. | 3.6 | 22 |
| 11 | The management of phenylketonuria in adult patients in Italy: a survey of six specialist metabolic centers. Current Medical Research and Opinion, 2021, 37, 411-421. | 1.9 | 11 |
| 12 | Phenylketonuria. Nature Reviews Disease Primers, 2021, 7, 36. | 30.5 | 174 |
| 13 | Detection of 3-O-methyldopa in dried blood spots for neonatal diagnosis of aromatic L-amino-acid decarboxylase deficiency: The northeastern Italian experience. Molecular Genetics and Metabolism, 2021, 133, 56-62. | 1.1 | 16 |
| 14 | Newborn Screening for Fabry Disease in Northeastern Italy: Results of Five Years of Experience. Biomolecules, 2021, 11, 951. | 4.0 | 20 |
| 15 | Long-term efficacy and safety of sapropterin in patients who initiated sapropterin at <â€‰4Âyears of age with phenylketonuria: results of the 3-year extension of the SPARK open-label, multicentre, randomised phase IIIb trial. Orphanet Journal of Rare Diseases, 2021, 16, 341. | 2.7 | 13 |
| 16 | Dysbiosis, Host Metabolism, and Non-communicable Diseases: Trialogue in the Inborn Errors of Metabolism. Frontiers in Physiology, 2021, 12, 716520. | 2.8 | 15 |
| 17 | Aromatic L-amino Acid Decarboxylase (AADC) deficiency: results from an Italian modified Delphi consensus. Italian Journal of Pediatrics, 2021, 47, 13. | 2.6 | 15 |
| 18 | Italian national consensus statement on management and pharmacological treatment of phenylketonuria. Orphanet Journal of Rare Diseases, 2021, 16, 476. | 2.7 | 12 |

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|---|---|---|---|
| 19 | The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250. | 6.2 | 138 |
| 20 | Neonatal Screening for MPS Disorders in Latin America: A Survey of Pilot Initiatives. International Journal of Neonatal Screening, 2020, 6, 90. | 3.2 | 5 |
| 21 | The Impact of a Slow-Release Large Neutral Amino Acids Supplement on Treatment Adherence in Adult Patients with Phenylketonuria. Nutrients, 2020, 12, 2078. | 4.1 | 14 |
| 22 | Retrospective analysis of 19 patients with 6-Pyruvoyl Tetrahydropterin Synthase Deficiency: Prolactin levels inversely correlate with growth. Molecular Genetics and Metabolism, 2020, 131, 380-389. | 1.1 | 7 |
| 23 | Report of Five Years of Experience in Neonatal Screening for Mucopolysaccharidosis Type I and Review of the Literature. International Journal of Neonatal Screening, 2020, 6, 85. | 3.2 | 12 |
| 24 | Nutrition, Microbiota and Role of Gut-Brain Axis in Subjects with Phenylketonuria (PKU): A Review. Nutrients, 2020, 12, 3319. | 4.1 | 20 |
| 25 | Oxygen and nitrite reduction by hemeâ€deficient sulphite oxidase in a patient with mild sulphite oxidase deficiency. Journal of Inherited Metabolic Disease, 2020, 43, 748-757. | 3.6 | 5 |
| 26 | The combined use of enzyme activity and metabolite assays as a strategy for newborn screening of mucopolysaccharidosis type I. Clinical Chemistry and Laboratory Medicine, 2020, 58, 2063-2072. | 2.3 | 12 |
| 27 | Implementation of Second-Tier Tests in Newborn Screening for Lysosomal Disorders in North Eastern Italy. International Journal of Neonatal Screening, 2019, 5, 24. | 3.2 | 45 |
| 28 | Large Neutral Amino Acid Therapy Increases Tyrosine Levels in Adult Patients with Phenylketonuria: A Long-Term Study. Nutrients, 2019, 11, 2541. | 4.1 | 12 |
| 29 | The neurological and psychological phenotype of adult patients with earlyâ€treated phenylketonuria: A systematic review. Journal of Inherited Metabolic Disease, 2019, 42, 209-219. | 3.6 | 42 |
| 30 | Plasma and dried blood spot lysosphingolipids for the diagnosis of different sphingolipidoses: a comparative study. Clinical Chemistry and Laboratory Medicine, 2019, 57, 1863-1874. | 2.3 | 60 |
| 31 | Suggested guidelines for the diagnosis and management of urea cycle disorders: First revision. Journal of Inherited Metabolic Disease, 2019, 42, 1192-1230. | 3.6 | 277 |
| 32 | Maternal germline mosaicism in Fabry disease. Neurological Sciences, 2019, 40, 1279-1281. | 1.9 | 7 |
| 33 | Inherited hyperammonemias: a Contemporary view on pathogenesis and diagnosis. Expert Opinion on Orphan Drugs, 2018, 6, 105-116. | 0.8 | 1 |
| 34 | Long term clinical history of an Italian cohort of infantile onset Pompe disease treated with enzyme replacement therapy. Orphanet Journal of Rare Diseases, 2018, 13, 32. | 2.7 | 65 |
| 35 | Pyridoxine-dependent epilepsies: an observational study on clinical, diagnostic, therapeutic and prognostic features in a pediatric cohort. Metabolic Brain Disease, 2018, 33, 261-269. | 2.9 | 18 |
| 36 | Newborn screening for lysosomal storage disorders by tandem mass spectrometry in North East Italy. Journal of Inherited Metabolic Disease, 2018, 41, 209-219. | 3.6 | 114 |

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|---|---|---|---|
| 37 | Newborn screening in mucopolysaccharidoses. Italian Journal of Pediatrics, 2018, 44, 126. | 2.6 | 50 |
| 38 | Mutations in the GLA Gene and LysoGb3: Is It Really Anderson-Fabry Disease?. International Journal of Molecular Sciences, 2018, 19, 3726. | 4.1 | 63 |
| 39 | N-Acetylglutamate Synthase Deficiency Due to a Recurrent Sequence Variant in the N-acetylglutamate Synthase Enhancer Region. Scientific Reports, 2018, 8, 15436. | 3.3 | 7 |
| 40 | The potential role of gut microbiota and its modulators in the management of propionic and methylmalonic acidemia. Expert Opinion on Orphan Drugs, 2018, 6, 683-692. | 0.8 | 4 |
| 41 | Living with phenylketonuria in adulthood: The PKU ATTITUDE study. Molecular Genetics and Metabolism Reports, 2018, 16, 39-45. | 1.1 | 67 |
| 42 | Brain malformations associated to Aldh7a1 gene mutations: Report of a novel homozygous mutation and literature review. European Journal of Paediatric Neurology, 2018, 22, 1042-1053. | 1.6 | 18 |
| 43 | Food triggers and inherited metabolic disorders: a challenge to the pediatrician. Italian Journal of Pediatrics, 2018, 44, 18. | 2.6 | 5 |
| 44 | Newborn Screening and High Risk Screening Population for Neurological Inherited Metabolic Diseases. , 2018, , 29-41. | | 0 |
| 45 | Key European guidelines for the diagnosis and management of patients with phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 743-756. | 11.4 | 272 |
| 46 | Efficacy, safety and population pharmacokinetics of sapropterin in PKU patients <4Âyears: results from the SPARK open-label, multicentre, randomized phase IIIb trial. Orphanet Journal of Rare Diseases, 2017, 12, 47. | 2.7 | 26 |
| 47 | Issues with European guidelines for phenylketonuria â€“ Authors' reply. Lancet Diabetes and Endocrinology,the, 2017, 5, 683-684. | 11.4 | 8 |
| 48 | International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and followâ€up. Journal of Inherited Metabolic Disease, 2017, 40, 171-176. | 3.6 | 132 |
| 49 | Diagnosis of sphingolipidoses: a new simultaneous measurement of lysosphingolipids by LC-MS/MS. Clinical Chemistry and Laboratory Medicine, 2017, 55, 403-414. | 2.3 | 78 |
| 50 | Diagnostic tests for Niemann-Pick disease type C (NP-C): A critical review. Molecular Genetics and Metabolism, 2016, 118, 244-254. | 1.1 | 114 |
| 51 | Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. Journal of Inherited Metabolic Disease, 2016, 39, 661-672. | 3.6 | 52 |
| 52 | Clinical experience with N-carbamylglutamate in a single-centre cohort of patients with propionic and methylmalonic aciduria. Molecular Genetics and Metabolism Reports, 2016, 8, 34-40. | 1.1 | 20 |
| 53 | The long-term treatment of a patient with type 1 diabetes mellitus and glutaric aciduria type 1: the effect of insulin. European Journal of Pediatrics, 2016, 175, 1123-1128. | 2.7 | 1 |
| 54 | High level of oxysterols in neonatal cholestasis: a pitfall in analysis of biochemical markers for Niemann-Pick type C disease. Clinical Chemistry and Laboratory Medicine, 2016, 54, 1221-1229. | 2.3 | 38 |

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|---|---|---|---|
| 55 | Diagnostic and management practices for phenylketonuria in 19 countries of the South and Eastern European Region: survey results. European Journal of Pediatrics, 2016, 175, 261-272. | 2.7 | 23 |
| 56 | Clinical and biochemical characterization of four patients with mutations in ECHS1. Orphanet Journal of Rare Diseases, 2015, 10, 79. | 2.7 | 68 |
| 57 | Assessment of the impact of phenylketonuria and its treatment on quality of life of patients and parents from seven European countries. Orphanet Journal of Rare Diseases, 2015, 10, 80. | 2.7 | 98 |
| 58 | Survey of Italian pediatriciansâ€™ perspectives and knowledge about neonatal screening. Italian Journal of Pediatrics, 2015, 41, 41. | 2.6 | 6 |
| 59 | Changing Characteristics of Late-Onset Pompe Disease Patients in Italy: Data from the Pompe Registry. Journal of Neuromuscular Diseases, 2015, 2, S36-S37. | 2.6 | 2 |
| 60 | Management of adult patients with phenylketonuria: survey results from 24 countries. European Journal of Pediatrics, 2015, 174, 119-127. | 2.7 | 32 |
| 61 | Development and psychometric validation of measures to assess the impact of phenylketonuria and its dietary treatment on patientsâ€™ and parentsâ€™ quality of life: the phenylketonuria â€“ quality of life (PKU-QOL) questionnaires. Orphanet Journal of Rare Diseases, 2015, 10, 59. | 2.7 | 52 |
| 62 | The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. Journal of Inherited Metabolic Disease, 2015, 38, 1041-1057. | 3.6 | 186 |
| 63 | The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074. | 3.6 | 175 |
| 64 | Changing Characteristics of Late-Onset Pompe Disease Patients in Italy: Data from the Pompe Registry. Journal of Neuromuscular Diseases, 2015, 2, S36-S37. | 2.6 | 0 |
| 65 | Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130. | 2.7 | 482 |
| 66 | Quality of Life (QoL) assessment in a cohort of patients with Phenylketonuria. BMC Public Health, 2014, 14, 1243. | 2.9 | 35 |
| 67 | Long-term Follow-up and Outcome of Phenylketonuria Patients on Sapropterin: A Retrospective Study. Pediatrics, 2013, 131, e1881-e1888. | 2.1 | 68 |
| 68 | Fluctuations in phenylalanine concentrations in phenylketonuria: A review of possible relationships with outcomes. Molecular Genetics and Metabolism, 2013, 110, 418-423. | 1.1 | 69 |
| 69 | Application of the WHOQOL-100 for the assessment of quality of life of adult patients with inherited metabolic diseases. Molecular Genetics and Metabolism, 2012, 106, 25-30. | 1.1 | 20 |
| 70 | Suggested guidelines for the diagnosis and management of urea cycle disorders. Orphanet Journal of Rare Diseases, 2012, 7, 32. | 2.7 | 596 |
| 71 | Up to date knowledge on different treatment strategies for phenylketonuria. Molecular Genetics and Metabolism, 2011, 104, S19-S25. | 1.1 | 59 |
| 72 | Impact of Metabolic Control on Bone Quality in Phenylketonuria and Mild Hyperphenylalaninemia. Journal of Pediatric Gastroenterology and Nutrition, 2011, 52, 345-350. | 1.8 | 15 |

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|---|---|---|---|
| 73 | Diagnosis and management of glutaric aciduria type I â€“ revised recommendations. Journal of Inherited Metabolic Disease, 2011, 34, 677-694. | 3.6 | 327 |
| 74 | Whole-blood alpha-D-galactosidase A activity for the identification of Fabry's patients. Clinical Biochemistry, 2011, 44, 916-921. | 1.9 | 8 |
| 75 | Brain MRI diffusion-weighted imaging in patients with classical phenylketonuria. Neuroradiology, 2009, 51, 803-812. | 2.2 | 39 |
| 76 | Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency. Human Mutation, 2008, 29, 167-175. | 2.5 | 158 |
| 77 | Inborn errors of metabolism in the Italian pediatric population: A national retrospective survey. Journal of Pediatrics, 2002, 140, 321-329. | 1.8 | 196 |
| 78 | Long-chain L 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency does not appear to be the primary cause of lipid myopathy in patients with Bannayan-Riley-Ruvalcaba syndrome (BRRS). , 1999, 83, 3-5. | | 9 |
| 79 | Mast Cells Contain Large Quantities of Secretagogueâ€Sensitive Nâ€Acetylaspartate. Journal of Neurochemistry, 1997, 69, 1314-1317. | 3.9 | 38 |
| 80 | Mitochondrial Short-Chain L-3-Hydroxyacl-Coenzyme A Dehydrogenase Deficiency: A New Defect of Fatty Acid Oxidation. Pediatric Research, 1996, 39, 185-188. | 2.3 | 71 |