

# Alberto B Burlina

## List of Publications by Year in descending order

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Version: 2024-02-01

80  
papers

5,196  
citations

117625

34  
h-index

91884

69  
g-index

86  
all docs

86  
docs citations

86  
times ranked

4501  
citing authors

#	ARTICLE	IF	CITATIONS
1	Suggested guidelines for the diagnosis and management of urea cycle disorders. Orphanet Journal of Rare Diseases, 2012, 7, 32.	2.7	596
2	Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130.	2.7	482
3	Diagnosis and management of glutaric aciduria type I " revised recommendations. Journal of Inherited Metabolic Disease, 2011, 34, 677-694.	3.6	327
4	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
5	Key European guidelines for the diagnosis and management of patients with phenylketonuria. Lancet Diabetes and Endocrinology, 2017, 5, 743-756.	11.4	272
6	Inborn errors of metabolism in the Italian pediatric population: A national retrospective survey. Journal of Pediatrics, 2002, 140, 321-329.	1.8	196
7	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
8	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
9	Phenylketonuria. Nature Reviews Disease Primers, 2021, 7, 36.	30.5	174
10	Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency. Human Mutation, 2008, 29, 167-175.	2.5	158
11	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	6.2	138
12	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up. Journal of Inherited Metabolic Disease, 2017, 40, 171-176.	3.6	132
13	Diagnostic tests for Niemann-Pick disease type C (NP-C): A critical review. Molecular Genetics and Metabolism, 2016, 118, 244-254.	1.1	114
14	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
15	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
16	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
17	Mitochondrial Short-Chain L-3-Hydroxyacyl-Coenzyme A Dehydrogenase Deficiency: A New Defect of Fatty Acid Oxidation. Pediatric Research, 1996, 39, 185-188.	2.3	71
18	Fluctuations in phenylalanine concentrations in phenylketonuria: A review of possible relationships with outcomes. Molecular Genetics and Metabolism, 2013, 110, 418-423.	1.1	69

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19	Long-term Follow-up and Outcome of Phenylketonuria Patients on Sapropterin: A Retrospective Study. <i>Pediatrics</i> , 2013, 131, e1881-e1888.	2.1	68
20	Clinical and biochemical characterization of four patients with mutations in ECHS1. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 79.	2.7	68
21	Living with phenylketonuria in adulthood: The PKU ATTITUDE study. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 16, 39-45.	1.1	67
22	Long term clinical history of an Italian cohort of infantile onset Pompe disease treated with enzyme replacement therapy. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 32.	2.7	65
23	Mutations in the GLA Gene and LysoGb3: Is It Really Anderson-Fabry Disease?. <i>International Journal of Molecular Sciences</i> , 2018, 19, 3726.	4.1	63
24	Plasma and dried blood spot lysosphingolipids for the diagnosis of different sphingolipidoses: a comparative study. <i>Clinical Chemistry and Laboratory Medicine</i> , 2019, 57, 1863-1874.	2.3	60
25	Up to date knowledge on different treatment strategies for phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2011, 104, S19-S25.	1.1	59
26	Development and psychometric validation of measures to assess the impact of phenylketonuria and its dietary treatment on patients' and parents' quality of life: the phenylketonuria " quality of life (PKU-QOL) questionnaires. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 59.	2.7	52
27	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 661-672.	3.6	52
28	Newborn screening in mucopolysaccharidoses. <i>Italian Journal of Pediatrics</i> , 2018, 44, 126.	2.6	50
29	Implementation of Second-Tier Tests in Newborn Screening for Lysosomal Disorders in North Eastern Italy. <i>International Journal of Neonatal Screening</i> , 2019, 5, 24.	3.2	45
30	The neurological and psychological phenotype of adult patients with early-treated phenylketonuria: A systematic review. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 209-219.	3.6	42
31	Brain MRI diffusion-weighted imaging in patients with classical phenylketonuria. <i>Neuroradiology</i> , 2009, 51, 803-812.	2.2	39
32	Mast Cells Contain Large Quantities of Sensitive <i>N</i> -Acetylaspartate. <i>Journal of Neurochemistry</i> , 1997, 69, 1314-1317.	3.9	38
33	High level of oxysterols in neonatal cholestasis: a pitfall in analysis of biochemical markers for Niemann-Pick type C disease. <i>Clinical Chemistry and Laboratory Medicine</i> , 2016, 54, 1221-1229.	2.3	38
34	Quality of Life (QoL) assessment in a cohort of patients with Phenylketonuria. <i>BMC Public Health</i> , 2014, 14, 1243.	2.9	35
35	Management of adult patients with phenylketonuria: survey results from 24 countries. <i>European Journal of Pediatrics</i> , 2015, 174, 119-127.	2.7	32
36	Efficacy and safety of empagliflozin in glycogen storage disease type Ib: Data from an international questionnaire. <i>Genetics in Medicine</i> , 2022, 24, 1781-1788.	2.4	29

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37	Efficacy, safety and population pharmacokinetics of sapropterin in PKU patients <math>\leq 4</math> years: results from the SPARK open-label, multicentre, randomized phase IIIb trial. Orphanet Journal of Rare Diseases, 2017, 12, 47.	2.7	26
38	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
39	Health-related quality of life in paediatric patients with intoxication-type inborn errors of metabolism: Analysis of an international data set. Journal of Inherited Metabolic Disease, 2021, 44, 215-225.	3.6	22
40	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
41	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
42	Nutrition, Microbiota and Role of Gut-Brain Axis in Subjects with Phenylketonuria (PKU): A Review. Nutrients, 2020, 12, 3319.	4.1	20
43	Newborn Screening for Fabry Disease in Northeastern Italy: Results of Five Years of Experience. Biomolecules, 2021, 11, 951.	4.0	20
44	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
45	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
46	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
47	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
48	Dysbiosis, Host Metabolism, and Non-communicable Diseases: Triologue in the Inborn Errors of Metabolism. Frontiers in Physiology, 2021, 12, 716520.	2.8	15
49	Aromatic L-amino Acid Decarboxylase (AADC) deficiency: results from an Italian modified Delphi consensus. Italian Journal of Pediatrics, 2021, 47, 13.	2.6	15
50	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
51	Long-term efficacy and safety of sapropterin in patients who initiated sapropterin at <math>\leq 4</math> years 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
52	Large Neutral Amino Acid Therapy Increases Tyrosine Levels in Adult Patients with Phenylketonuria: A Long-Term Study. Nutrients, 2019, 11, 2541.	4.1	12
53	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
54	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

#	ARTICLE	IF	CITATIONS
55	Italian national consensus statement on management and pharmacological treatment of phenylketonuria. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 476.	2.7	12
56	The management of phenylketonuria in adult patients in Italy: a survey of six specialist metabolic centers. <i>Current Medical Research and Opinion</i> , 2021, 37, 411-421.	1.9	11
57	A New Approach to Objectively Evaluate Inherited Metabolic Diseases for Inclusion on Newborn Screening Programmes. <i>International Journal of Neonatal Screening</i> , 2022, 8, 25.	3.2	10
58	Application of a Novel Algorithm for Expanding Newborn Screening for Inherited Metabolic Disorders across Europe. <i>International Journal of Neonatal Screening</i> , 2022, 8, 20.	3.2	10
59	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
60	Whole-blood alpha-D-galactosidase A activity for the identification of Fabry's patients. <i>Clinical Biochemistry</i> , 2011, 44, 916-921.	1.9	8
61	Issues with European guidelines for phenylketonuria – Authors' reply. <i>Lancet Diabetes and Endocrinology</i> , 2017, 5, 683-684.	11.4	8
62	N-Acetylglutamate Synthase Deficiency Due to a Recurrent Sequence Variant in the N-acetylglutamate Synthase Enhancer Region. <i>Scientific Reports</i> , 2018, 8, 15436.	3.3	7
63	Maternal germline mosaicism in Fabry disease. <i>Neurological Sciences</i> , 2019, 40, 1279-1281.	1.9	7
64	Retrospective analysis of 19 patients with 6-Pyruvoyl Tetrahydropterin Synthase Deficiency: Prolactin levels inversely correlate with growth. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 380-389.	1.1	7
65	Survey of Italian pediatricians' perspectives and knowledge about neonatal screening. <i>Italian Journal of Pediatrics</i> , 2015, 41, 41.	2.6	6
66	Food triggers and inherited metabolic disorders: a challenge to the pediatrician. <i>Italian Journal of Pediatrics</i> , 2018, 44, 18.	2.6	5
67	Neonatal Screening for MPS Disorders in Latin America: A Survey of Pilot Initiatives. <i>International Journal of Neonatal Screening</i> , 2020, 6, 90.	3.2	5
68	Oxygen and nitrite reduction by heme-deficient sulphite oxidase in a patient with mild sulphite oxidase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 748-757.	3.6	5
69	The potential role of gut microbiota and its modulators in the management of propionic and methylmalonic acidemia. <i>Expert Opinion on Orphan Drugs</i> , 2018, 6, 683-692.	0.8	4
70	Changing Characteristics of Late-Onset Pompe Disease Patients in Italy: Data from the Pompe Registry. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, S36-S37.	2.6	2
71	Newborn screening of mucopolysaccharidosis type I. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 2022, 59, 257-277.	6.1	2
72	Disorders of Phenylalanine and Tetrahydrobiopterin Metabolism. , 2022, , 331-351.		2

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73	The long-term treatment of a patient with type 1 diabetes mellitus and glutaric aciduria type 1: the effect of insulin. <i>European Journal of Pediatrics</i> , 2016, 175, 1123-1128.	2.7	1
74	Inherited hyperammonemias: a Contemporary view on pathogenesis and diagnosis. <i>Expert Opinion on Orphan Drugs</i> , 2018, 6, 105-116.	0.8	1
75	A new strategy of desensitization in mucopolysaccharidosis type II disease treated with idursulfase therapy: A case report and review of the literature. <i>Molecular Genetics and Metabolism Reports</i> , 2022, 31, 100878.	1.1	1
76	Bone disease in early detected Gaucher Type I disease: A case report. <i>JIMD Reports</i> , 2022, 63, 414-419.	1.5	1
77	Newborn Screening and High Risk Screening Population for Neurological Inherited Metabolic Diseases. , 2018, , 29-41.		0
78	Atherogenic lipid profile in patients with Niemann-Pick disease type B: What treatment strategies?. <i>Journal of Clinical Lipidology</i> , 2022, , .	1.5	0
79	Changing Characteristics of Late-Onset Pompe Disease Patients in Italy: Data from the Pompe Registry. <i>Journal of Neuromuscular Diseases</i> , 2015, 2, S36-S37.	2.6	0
80	Experience of the NPC Brazil Network with a Comprehensive Program for the Screening and Diagnosis of Niemann-Pick Disease Type C. <i>International Journal of Neonatal Screening</i> , 2022, 8, 39.	3.2	0