

CURRICULUM VITAE

Alessandro P. Burlina

INFORMAZIONI LAVORATIVE

Direttore U.O.C. di Neurologia, Ospedale San Bassiano, Bassano del Grappa, AULSS
7 "Pedemontana"

STUDI UNIVERSITARI

Laurea in Medicina e Chirurgia

Università di Padova

Esame di abilitazione all'esercizio della professione medica

Università di Padova

QUALIFICAZIONI POST-UNIVERSITARIE

Specializzazione in Neurologia

Università di Padova

Dottorato di Ricerca in Scienze Neurologiche

Università di Verona

PROGETTI DI RICERCA CONDOTTI ALL'ESTERO

**Center for Neurochemistry, Nathan S. Kline Institute for Psychiatric Research,
New York University, New York, NY- USA (12 mesi)**

Prof. Abel Lajtha

Yale University, School of Medicine, Department of Neurology

New Haven, CT-USA (12 mesi)

Prof. James W. Prichard

**Biozentrum, University of Basel, Department of Biophysical Chemistry
Basel, Switzerland (15 mesi)**

Prof. Joachim Seelig

Interesse principale in ambito clinico e scientifico: malattie neurometaboliche ereditarie

PUBBLICAZIONI SCIENTIFICHE

Riviste in lingua inglese con sistema di referaggio tra pari

1. Galzigna L, Fasolato C, Bianchi M, **Burlina AP**, Previero A.
Dopamine-depleting activity of L-3,4-(dioxypheylacetyl)-phenylalanine.
Neuropsychobiology 1988; 19:180-185
2. Debler EA, Wajda I, Manigault I, **Burlina AP**, Lajtha A.
Effects of caffeine on amino acid transport in the brain.
Neurochem Int 1989; 14:55-60
3. **Burlina AP**, Sershen H, Debler EA, Lajtha A.
Uptake of acetyl-L-carnitine in the brain.
Neurochem Res 1989; 14:489-493
4. Plebani M, Masiero M, **Burlina AP**, Chiozza ML, Scanarini M, Burlina A.
Measurement of melatonin in blood by radioimmunoassay. Analytical considerations and clinical usefulness.
Child's Nerv Syst 1990; 6:220-221
5. Petroff OAC, **Burlina AP**, Black J, Prichard JW.
Metabolism of [1-¹³C] glucose in a synaptosomally enriched fraction of rat cerebrum studied by H-1/C-13 magnetic resonance spectroscopy.
Neurochem Res 1991; 16:1245-1251
6. Seelig J, **Burlina AP**.
Carbon-13 magnetic resonance in biology and medicine.
Clin Chim Acta 1992; 206:125-136
7. Petroff OAC, **Burlina AP**, Black J, Prichard JW.
Quantitative analysis of rat synaptosomes and cerebrum using high-resolution ¹H magnetic resonance spectroscopy.
Clin Chim Acta 1992; 206:137-146
8. **Burlina AP**, Skaper SD, Mazza MR, Ferrari V, Leon A, Burlina AB.
N-acetylaspartylglutamate selectively inhibits neuronal responses to N-methyl-D-aspartic acid in vitro.
J Neurochem 1994; 63:1174-1177
9. **Burlina AP**, Corazza A, Ferrari V, Erhard P, Künnecke B, Seelig J, Burlina AB.
Detection of increased urinary N-acetylaspartylglutamate in Canavan disease.
Eur J Pediatr 1994; 153:538-539
10. Hagberg G, **Burlina AP**, Mader I, Roser W, Radü EW, Seelig J.
In vivo proton MR spectroscopy of human gliomas: definition of metabolic coordinates for multi-dimensional classification.
Magn Reson Med 1995; 34:242-252

11. **Burlina AP**, Ferrari V, Facci L, Skaper SD, Burlina AB.
Mast cells contain large quantities of secretagogue-sensitive N-acetylaspartate.
J Neurochem, 1997; 69:1314-1317
12. Casolino M, De Pascale MP, Morselli A, Narici L, Picozza P, Prigione V, Sparvoli R, Adriani O, Spillantini P, Castellini G, Bartalucci S, Catena C, Conti D, Ricci M, Righi E, Spataro B, Trenta G, Durante M, Gialanella G, Grossi G, Pugliese M, Barbiellini G, Boezio M, Vacchi A, Zampa N, Sannita WG, Lopez L, Peresson M, Conforto S, **Burlina AP**, Tanzarella C, Alberici G, Casoli L, Cerdoni S, Lenti A, Galper A, Ozerov Yu, Popov A, Zemskov V, Zverev V, Alexandrov A, Avdeev S, Shabelnikov V.
Light flash observation in space: experiment ELFO.
Il Nuovo Cimento 1997; 19D:1601-1623
13. **Burlina AP**, Ferrari V, Divry P, Gradowska W, Jakobs C, Bennett MJ, Sewell AC, Dionisi-Vici C, Burlina AB.
N-acetylaspartylglutamate in Canavan disease: an adverse effector ?
Eur J Pediatr, 1999; 158:406-409
14. Burlina AB, Bonafé L, Ferrari V, Suppiej A, Zacchello F, **Burlina AP**.
Measurement of neurotransmitters in the cerebrospinal fluid of phenylketonuric patients under dietary treatment.
J Inherit Metab Dis 2000; 23:313-316.
15. **Burlina AP**, Aureli T, Bracco F, Conti F, Battistin L.
MR spectroscopy: a powerful tool for investigating brain function and neurological diseases.
Neurochem Res 2000; 25:1365-1372
16. Bonafé L, Blau N, **Burlina AP**, Romstad A, Güttler F, Burlina AB.
Treatable neurotransmitter deficiency in mild phenylketonuria.
Neurology 2001; 57:908-911
17. **Burlina AP**, Baracchini C, Carollo C, Burlina AB.
Propionic acidemia with basal ganglia stroke: treatment of acute extrapyramidal symptoms with L-DOPA.
J Inherit Metab Dis 2001; 24:596-598
18. **Burlina AP**, Edini C, Burlina AB.
Treatment of extrapyramidal symptoms in a patient with homozygous homocystinuria.
J Inherit Metab Dis 2002; 25:135-136
19. De Iuliis A, **Burlina AP**, Boschetto R, Zambenedetti P, Arslan P, Galzigna L.
Increased dopamine peroxidation in postmortem Parkinsonian brain.
Biochim Biophys Acta (BBA General Subjects) 2002; 1573:63-67
20. **Burlina AP**, Manara R, Calderone M, Catuogno S, Burlina AB.

- Diffusion-weighted imaging in the assessment of neurological damage in patients with methylmalonic aciduria.
J Inherit Metab Dis 2003; 26:417-422
21. Boltshauser E, Schmitt B, Wevers RA, Engelke U, Burlina AB, **Burlina AP**. Follow-up of a child with hypoacetylaspartia.
Neuropediatrics, 2004; 35:255-258
 22. **Burlina AP**, Zara G, Hoffmann GF, Zschocke J, Burlina AB. Management of movement disorders in glutaryl-CoA dehydrogenase deficiency: anticholinergic drugs and botulinum toxin as additional therapeutic options.
J Inherit Metab Dis 2004; 27:911-915
 23. Goi G, Massacesi L, **Burlina AP**, Baquero Herrera CJ, Lombardo A, Tettamanti G, Burlina AB. Lysosomal leukocyte β -D-glucuronidase during enzymatic therapy replacement in Fabry disease.
Biochim Biophys Acta (BBA Molecular Basis of Disease) 2005; 1741:300-306
 24. Burlina AB, Peduto A, Di Palma A, Bellizzi A, Sperli D, Morrone A, **Burlina AP**. An unusual clinical and biochemical presentation of ornithine transcarbamylase deficiency in a male patient.
J Inherit Metab Dis 2006; 29:179-181
 25. Ginsberg L, Manara R, Valentine AR, Kendall B, **Burlina AP**. Magnetic resonance imaging changes in Fabry disease.
Acta Paediatrica 2006; 95 (Suppl 451):57-62
 26. **Burlina AP**, Schmitt B, Engelke U, Wevers RA, Burlina AB, Boltshauser E. Hypoacetylaspartia: clinical and biochemical follow-up of a patient.
Adv Exp Med Biol 2006; 576:283-287
 27. **Burlina AP**, Ferrari V, Burlina AB, Ermani M, Boespflug-Tanguy O, Bertini E, and the Clinical European Network on Brain Dysmyelinating Disease N-acetylaspartylglutamate (NAAG) in Pelizaeus-Merzbacher disease.
Adv Exp Med Biol 2006; 576:353-359
 28. Kölker S, Christensen E, Leonard JV, Greenberg CR, Burlina AB, **Burlina AP**, Dixon M, Duran M, Goodman SI, Koeller DM, Müller E, Naughten ER, Neumaier-Probst E, Okun JG, Kyllerman M, Surtees R, Wilcken B, Hoffmann GF, Burgard P. Guidelines for the diagnosis and management of glutaryl-CoA dehydrogenase deficiency (glutaric aciduria type I).
J Inherit Metab Dis 2007; 30:5-22
 29. Matalon R, Michals-Matalon K, Bhatia G, Burlina AB, **Burlina AP**, Braga C, Fiori L, Giovannini M, Grechanina E, Novikov P, Grady J, Tyring SK, Guttler F.

- Double blind placebo control trial of large neutral amino acids in treatment of PKU: effect on blood phenylalanine.
J Inherit Metab Dis 2007; 30:153-158
30. Henneke M, Combes P, Diekmann S, Bertini E, Brockmann K, **Burlina AP**, Kaiser J, Ohlenbusch A, Plecko B, Rodriguez D, Boespflug-Tanguy O, Gärtner J.
GJA12 mutations are a rare cause of Pelizaeus-Merzbacher-like disease.
Neurology 2008, 70:748-754
 31. Sartori S, Burlina AB, Salviati L, Trevisson E, Toldo I, Laverda AM, **Burlina AP**.
Increased level of N-acetylaspartylglutamate (NAAG) in the CSF of a patient with Pelizaeus-Merzbacher-like disease due to mutation in the GJA12 gene.
Eur J Paediatr Neurol 2008; 12:348-350
 32. **Burlina AP**, Manara R, Caillaud C, Laissy JP, Severino M, Klein I, Burlina A, Lidove O.
The *pulvinar sign*: frequency and clinical correlations in Fabry disease.
J Neurol 2008; 255:738-744
 33. Buechner S, Moretti M, **Burlina AP**, Cei G, Manara R, Ricci R, Mignani R, Parini R, Di Vito R, Giordano GP, Simonelli P, Siciliano G, Borsini W.
Central nervous system involvement in Anderson-Fabry disease: a clinical and MRI retrospective study.
J Neurol Neurosurg Psych 2008; 79:1249-1254
 34. De Iuliis A, Arrigoni G, Andersson L, **Burlina A**, Zambenedetti P, James P, Arslan P, Vianello F.
Oxidative metabolism of dopamine: a colour reaction from human midbrain analysed by mass spectrometry.
Biochim Biophys Acta (BBA – Proteins and Proteomics) 2008; 1784:1687-1693
 35. Mazzucco S, Anzola GP, Ferrarini M, Taioli F, Olivato S, **Burlina AP**, Fabrizi GM, Rizzuto N.
Cerebral autosomal dominant arteriopathy with subcortical infarcts and Leucoencephalopathy and right-to-left shunt: lack of evidence for an association in a prevalence study.
Eur Neurol 2009; 61:46-49
 36. Manara R, **Burlina AP**, Citton V, Ermani M, Vespignani F, Carollo C, Burlina AB.
Brain MRI diffusion-weighted imaging in patients with classical phenylketonuria.
Neuroradiology 2009; 51:803-812
 37. Liguori R, Di Stasi V, Bugiardini E, Mignani R, **Burlina A**, Borsini W, Baruzzi A, Montagna P, Donadio P.
Small fiber neuropathy in female patients with Fabry disease.

- Muscle Nerve* 2010; 41:409-412
38. Salviati A, **Burlina AP**, Borsini W.
Nervous system and Fabry disease, from symptoms to diagnosis: damage evaluation and follow-up in adult patients, enzyme replacement, and support therapy.
Neurol Sci 2010; 31:299-306
 39. Watt T, **Burlina AP**, Cazzorla C, Schönfeld D, Banikazemi M, Hopkin RJ, Martins AM, Sims K, Beitner-Johnson D, O'Brien F, Feldt-Rasmussen U.
Agalsidase beta treatment is associated with improved quality of life in patients with Fabry disease: findings from the Fabry Registry.
Genet Med 2010; 12:703-712
 40. Kölker S, Christensen E, Leonard JV, Greenberg CR, Boneh A, Burlina AB, **Burlina AP**, Dixon M, Duran M, Garcia-Cazorla A, Goodman SI, Koeller DM, Kyllerman M, Mülhausen C, Müller E, Okun JG, Wilcken B, Hoffmann GF, Burgard P.
Diagnosis and management of glutaric aciduria type I – revised recommendations.
J Inher Metab Dis 2011; 34:677-694
 41. Manara R, Citton V, Nardetto L, Semplicini C, **Burlina A**, Trevisan C, Argentiero V, Baracchini C.
Spinal cord herniation: a missing piece in the pathogenesis of amyotrophic lateral sclerosis and multiple sclerosis?
Neurodegener Dis 2011; 8:381-385
 42. Massaccesi L, Burlina A, Baquero CJ, Goi G, **Burlina AP**, Tettamanti G.
Whole-blood alpha-D-galactosidase A activity for the identification of Fabry's patients.
Clin Biochem 2011; 44:916-921
 43. **Burlina AP**, Sims KB, Politei JM, Bennett GJ, Baro R, Sommer C, Torvin Møller A, Hilz MJ.
Early diagnosis of peripheral nervous system involvement in Fabry disease and treatment of neuropathic pain: the report of an expert panel.
BMC Neurol 2011; 11:61
 44. Baslow MH, **Burlina AP**.
N-acetylaspartate metabolism underlies the structural and functional units of the vertebrate brain: a bioenergetic rationale for clinical observations of changes in the neuronal biomarker "NAA" in many human brain disorders.
Bioenerg Open Access 2012; 1:102
 45. Citton V, Burlina A, Baracchini C, Gallucci M, Catalucci A, Dal Pos S, **Burlina A**, Manara R.
Apparent diffusion coefficient restriction in the white matter: going beyond acute brain territorial ischemia.
Insights Imaging 2012; 3:155-164

46. Manara R, Del Rizzo M, **Burlina AP**, Bordugo A, Citton V, Rodriguez-Pombo P, Ugarte M, Burlina AB.
Wernicke-like encephalopathy during classic maple syrup urine disease decompensation.
J Inherit Metab Dis 2012; 35:413-417
47. Cazzorla C, Del Rizzo M, Burgard P, Zanco C, Bordugo A, Burlina AB, **Burlina AP**.
Application of the WHOQOL-100 for the assessment of quality of life of adult patients with inherited metabolic diseases.
Mol Genet Metab 2012; 106:25-30
48. **Burlina AP**, Politei J, Cinque S, Soliani A, Carlier RY, Germain DP, Manara R.
The pulvinar sign in Fabry patients: the first report in female patients.
J Neurol 2012; 259:1227-1228
49. Patterson MC, Hendriksz CJ, Walterfang M, Sedel F, Vanier MT, Wijburg F, NP-C Guidelines Working Group*
Recommendations for the diagnosis and management of Niemann-Pick disease type C: an update.
Mol Genet Metab 2012; 106:330-344 (*member)
50. Linthorst GE, **Burlina AP**, Cecchi F, Cox TM, Fletcher JM, Feldt-Rasmussen U, Giugliani R, Hollak CEM, Houge G, Hughes D, Kantola I, Lachmann R, Lopez M, Ortiz A, Parini R, Rivera A, Rolfs A, Ramaswami U, Svarstad E, Tondel C, Tylki-Szymanska A, Vujkovic B, Waldek S, West M, Weidemann F, Mehta A.
Recommendations on reintroduction of agalsidase beta for patients with Fabry disease in Europe, following a period of shortage.
JIMD Rep 2013; 8:51-56
51. Del Rizzo M, **Burlina AP**, Sass JO, Beermann F, Zanco C, Cazzorla C, Bordugo A, Giordano L, Manara R, Burlina AB.
Metabolic stroke in a late-onset form of isolated sulfite oxidase deficiency.
Mol Genet Metab 2013; 108:263-266
52. Politei J, Schenone A, **Burlina A**, Blanco M, Lescano S, Szlago M.
Vertebrobasilar dolichoectasia in Fabry disease: the earliest marker of neurovascular involvement ?
J Inborn Errors Metab Screening 2014; 2:1-6
53. **Burlina A**.
Niemann-Pick type C: introduction and main clinical features
J Neurol 2014; 261 (Suppl 2):S5252-S527
54. Mosca L, Rivieri F, Tanel R, Bonfante A, **Burlina A**, Manfredini E, Primignani P, Gesu GP, Marocchi A, Penco S.

- Mutational screening of NOTCH3 gene reveals two novel mutations: complexity of CADASIL diagnosis.
J Mol Neurosci 2014; 54:723-729
55. Cazzorla C, Cegolon L, **Burlina AP**, Celato A, Massa P, Giordano L, Polo G, Daniele A, Salvatore F, Burlina AB.
Quality of life (QoL) assessment in a cohort of patients with phenylketonuria.
BMC Public Health 2014; 14:1243
 56. Kolodny E, Fellgiebel A, Hilz MJ, Sims K, Caruso P, Phan TG, Politei J, Manara R, **Burlina A**.
Cerebrovascular involvement in Fabry disease: current status of knowledge.
Stroke 2015; 46:302-313
 57. Viggiano E, Marabotti A, **Burlina AP**, Cazzorla C, D'Apice MR, Giordano L, Fasan I, Novelli G, Facchiano A, Burlina AB.
Clinical and molecular spectra in galactosemic patients from neonatal screening in northeastern Italy: structural and functional characterization of new variations in the galactose-1-phosphate uridylyltransferase (GALT) gene.
Gene 2015; 559:112-118
 58. Reunert J, Lotz-Havla AS, Polo G, Kannenberg F, Fobker M, Griese M, Mengel E, Muntau AC, Schnabel P, Sommerburg O, Borggraefe I, Dardis A, **Burlina AP**, Mall MA, Ciana G, Bembi B, Burlina AB, Marquardt T, Niemann-Pick type C-2 disease: identification by analysis of plasma cholestane-3b,5a,6b-triol and further insight into the clinical phenotype.
JIMD Rep 2015; 23:17-26
 59. Polo G, **Burlina A**, Furlan F, Kolamunnage T, Cananzi M, Giordano L, Zaninotto M, Plebani M, Burlina A.
High Level of oxysterols in neonatal cholestasis: a pitfall in analysis of Biochemical markers for Niemann-Pick type C disease.
Clin Chem Lab Med 2016; 54:1221-1229
 60. Politei J, Bouhassira D, Germain DP, Goizet C, Guerrero-Sola A, Hilz MJ, Hutton EJ, Karaa A, Liguori R, Üçeyler N, Zeltzer LK, **Burlina A**.
Pain in Fabry disease: practical recommendations for diagnosis and treatment.
CNS Neurosci Ther 2016; 22:568-576
 61. Mignani R, Pieruzzi F, Berri F, **Burlina A**, Chinaa B, Gallieni M, Pieroni M, Salviati A, Spada M.
FABry STabilization indEX (FASTEx): an innovative tool for the assessment of clinical stabilization in Fabry disease.
Clin Kidney J 2016; 9:739-747
 62. **Burlina A**, Politei.
The central nervous system involvement in Fabry disease: a review.
J Inborn Errors Metab Screening 2016; 4:1-7

63. Polo G, **Burlina AP**, Kolamunnage TB, Zampieri M, Dionisi-Vici C, Strisciuglio P, Zaninotto M, Plebani M, Burlina AB.
Diagnosis of sphingolipidoses: a new simultaneous measurement of lysosphingolipids by LC-MS/MS.
Clin Chem Lab Med 2017; 55:403-414
64. Poli L, Zedde M, Zini A, Del Sette M, Lodigiani C, Spalloni A, Di Lisi F, Toriello A, Piras V, Stilo C, Tomelleri G, Tancredi L, Paciaroni M, Silvestrelli G, Adami A, Costa P, Morotti A, De Giuli V, Caria F, Gamba M, Malferrari G, Simone AM, Musolino R, Giorli E, Banfi E, Marcheselli S, Rasura M, Pugliese N, Melis M, Bovi P, Padovani A, **Burlina A**, Pezzini A and on behalf of the Italian Project on Stroke in Young Adults (YPSYS) Investigators.
Screening for Fabry disease in patients with ischaemic stroke at young age: the Italian Project on Stroke in Young Adults.
Eur J Neurol 2017; 24:e12-e14
65. Manara R, Carlier RY, Righetto S, Citton V, Locatelli G, Colas F, Ermani M, Germain DP, **Burlina A**.
Basilar artery changes in Fabry disease.
Am J Neuroradiol 2017; 38:531-536
66. Burlina AB, Celato A, Polo G, Edini C. **Burlina AP**.
The utility of CSF for the diagnosis of primary and secondary monoamine neurotransmitter deficiencies.
EJIFCC 2017; 28:64-76
67. Liguori R, Incensi A, de Pasqua S, Mignani R, Fileccia E, Santostefano M, Biagini E, Rapezzi C, Palmieri S, Romani I, Borsini W, **Burlina A**, Bombardi R, Caprini C, Avoni P, Donadio V.
Skin globotriaosylceramide 3 deposits are specific to Fabry disease with classical mutations and associated with small fibre neuropathy.
PLOS ONE 2017; 12:e0180581
68. Busa G, **Burlina A**, Damuzzo V, Chiumente M, Palozzo AC.
Comorbidity, polytherapy, and drug interactions in a neurological context: an example of a multidisciplinary approach to promote the rational use of drugs.
J Pharm Pract 2018; 31:58-65
69. Colomba P, Zizzo C, Alessandro R, Cammarata G, Scalia S, Giordano A, Pieroni M, Sicurella L, Amico L, **Burlina A**, Duro G.
Fabry disease and multiple sclerosis misdiagnosis: the role of family history and neurological signs.
Oncotarget 2018; 9:7758-7762
70. Burlina AB, Polo G, Salviati L, Duro G, Zizzo C, Dardis A, Bembi B, Cazzorla C, Rubert L, Zordan R, Desnick RJ, **Burlina AP**.
Newborn screening for lysosomal storage disorders by tandem mass spectrometry in North East Italy.
J Inherit Metab Dis 2018; 41:209-219

71. Ortiz A, Germain DP, Desnick RJ, Politei J, Mauer M, **Burlina A**, Eng C, Hopkin RJ, Laney D, Linhart A, Waldek S, Wallace E, Weidemann F, Wilcox WR.
Fabry disease revisited: management and treatment recommendations for adult patients.
Mol Genet Metab 2018; 123:416-427
72. Hilz MJ, Arbustini E, Dagna L, Gasbarrini A, Goizet C, Lacombe D, Liguori R, Manna R, Politei J, Spada M, **Burlina A**.
Non-specific gastrointestinal features: could it be Fabry disease?
Dig Liver Dis 2018; 50:429-437
73. Wanner C, Arad M, Baron R, **Burlina A**, Elliott PM, Feldt-Rasmussen U, Fomin VV, Germain DP, Hughes DA, Jovanovic A, Kantola I, Linhart A, Mignani R, Monserrat L, Namdar M, Nowak A, Oliveira JP, Ortiz A, Pieroni M, Spada M, Tylki-Szymanska A, Tøndel C, Viana-Baptista M, Weidemann F, Hilz MJ.
European expert consensus statement on therapeutic goals in Fabry disease.
Mol Genet Metab 2018; 124:189-203
74. Germain DP, Brand E, **Burlina A**, Cecchi F, Garman SC, Kempf J, Laney DA, Linhart A, Marodi L, Nicholss K, Ortiz A, Pieruzzi F, Shankar SP, Waldek S, Wanner C, Jovanovic A.
Phenotypic characteristics of the p. Asn215Ser (p.N215S) *GLA* mutation in male and female patients with Fabry disease: a multicenter Fabry Registry study.
Mol Genet Genomic Med 2018; 6:492-503
75. Serrano Russi A, Donoghue S, Boneh A, Manara R, Burlina AB, **Burlina AP**.
Malignant brain tumors in patients with glutaric aciduria type I.
Mol Genet Metab 2018; 125:276-280
76. Burlina A, Tims S, van Spronsen F, Sperl W, **Burlina AP**, Kuhn M, Knol J, Rakhshandehroo M, Coskun T, Singh RH, MacDonald A.
The potential role of gut microbiota and its modulators in the management of propionic and methylmalonic acidemia.
Exp Opin Orphan Drugs 2018; 6:683-692
77. Duro G, Zizzo C, Cammarata G, **Burlina A**, Burlina A, Polo G, Scalia S, Oliveri R, Sciarrino S, Francofonte D, Alessandro R, Pisani A, Palladino G, Napoletano R, Tenuta M, Masarone D, Limongelli G, Riccio E, Frustaci A, Chimenti C, ferri C, Pieruzzi F, Pieroni M, Spada M, Castana C, Caserta M, Monte I, Rodolico MS, Feriozzi S, Battaglia Y, Amico L, Losi MA, Autore C, Lombardi M, Zoccali C, Testa A, Postorino M, Mignani R, Zachara E, Giordano A, Colomba P.
Int J Mol Sci 2018; 19:3726

78. Forlivesi S, Cappellari M, Baracchini C, Viaro F, Critelli A, Tamborino C, Tonello S, Guidoni SV, Bruno M, Favaretto S, **Burlina A**, Turinese E, Ferracci F, Zambito Marsala S, Bazzano S, Orlando F, Turazzini M, Ricci S, Cadaldini M, De Biasia F, Bruno S, Gaudenzi A, Morra M, Danese A, L'Erario R, Russo M, Zanette G, Idone D, Basile AM, Atzori M, Masato M, Menegazzo E, Paladin F, Tonon A, Caneve G, Bozzato G, Campagnaro A, Carella S, Nicolao P, Padoan R, Perini F, De Boni A, Adami A, Bonetti B, Bovi P.
Intravenous thrombolysis for ischemic stroke in the Veneto region: the gap between eligibility and reality.
J Thromb Thrombolysis 2019; 47:113-120
79. Lee TH, Yang JT, Lee JD, Chang KC, Peng TI, Chang TY, Huang KL, Liu CH, Ryu SJ, **Burlina AP**.
Genomic screening of Fabry disease in young stroke patients: the Taiwan experience and a review of the literature.
Eur J Neurol 2019; 26:553-555
80. **Burlina AP**, Lachmann RH, Manara R, Cazzorla C, Celato A, van Spronsen FJ, Burlina A.
The neurological and psychological phenotype of adult patients with early-treated phenylketonuria: a systematic review.
J Inherit Metab Dis 2019; 42:209-219
81. Germain DP, Arad M, **Burlina A**, Elliott PM, Falissard B, Feldt-Rasmussen U, Hilz MJ, Hughes DA, Ortiz A, Wanner C, Weidemann F, Spada M.
The effect of enzyme replacement therapy on clinical outcomes in female patients with Fabry disease – A systematic literature review by a European panel of experts.
Mol Genet Metab 2019; 126:224-235
82. Camporeale A, Pieroni M, Pieruzzi F, Lusardi P, Pica S, Spada M, Mignani R, **Burlina A**, Bandera F, Guazzi M, Graziani F, Crea F, Greiser A, Boveri S, Ambrogi F, Lombardi M.
Predictors of clinical evolution in prehypertrophic Fabry disease.
Circ Cardiovasc Imaging 2019; 12:e008424
83. Pianese L, Fortunato A, Silvestri S, Solano FG, Burlina A, **Burlina AP**, Ragno M.
Maternal germline mosaicism in Fabry disease.
Neurol Sci 2019; 40:1279-1281
84. Burlina AB, Polo G, Rubert L, Guerardi D, Cazzorla C, Duro G, Salviati L, **Burlina AP**.
Implementation of second-tier tests in newborn screening for lysosomal disorders in North Eastern Italy.
Int J Neonatal Screen 2019; 5(2):24
85. **Burlina AP**, Cazzorla C, Massa P, Polo G, Loro C, Guerardi D, Burlina AB.

Large neutral amino acid therapy increases tyrosine levels in adult patients with phenylketonuria: a long-term study.
Nutrients 2019; 11:2541

86. Polo G, **Burlina AP**, Ranieri E, Colucci F, Rubert L, Pascarella A, Duro G, Tummolo A, Padoan A, Plebani M, Burlina AB.
Plasma and dried blood spot lysosphingolipids for the diagnosis of different sphingolipidoses: a comparative study.
Clin Chem Lab Med 2019; 57:1863-1874
87. Mancuso M, Arnold M, Bersano A, **Burlina A**, Chabriat H, Debette S, Enzinger C, Federico A, Filla A, Finsterer J, Hunt D, Lesnil Oberstein S, Tournier-Lasserre E, Markus HM.
Monogenic cerebral small-vessel diseases: diagnosis and therapy. Consensus recommendations of the European Academy of Neurology.
Eur J Neurol 2020; 27:909-927
88. **Burlina AP**, Cazzorla C, Massa P, Loro C, Guerardi D, Burlina AB.
The impact of a slow-release large neutral amino acids supplement on treatment adherence in adult patients with phenylketonuria.
Nutrients 2020; 12:2078
89. Bernardini A, Camporeale A, Pieroni M, Pieruzzi F, Figliozzi S, Lusardi P, Spada M, Mignani R, **Burlina A**, Carubbi F, Battaglia Y, Graziani F, Pica S, Tondi L, Chow K, Boveri S, Olivotto I, Lombardi M.
Atrial dysfunction assessed by cardiac magnetic resonance as an early marker of Fabry cardiomyopathy.
JACC Cardiovasc Imag 2020; 13:2262-2264
90. Polo G, Guerardi D, Giuliani A, Rubert L, Cazzorla C, Salviati L, Marzollo A, Biffi A, **Burlina AP**, Burlina AB.
The combined use of enzyme activity and metabolite assays as a strategy for newborn screening of mucopolysaccharidosis type I.
Clin Chem Lab Med 2020; 58:2063-2072
91. Laney DA, Germain DP, Oliveira JP, **Burlina AP**, Cabrera GH, Hong G-R, Hopkin RJ, Niu D-M, Thomas M, Trimarchi H, Wilcox WR, Politei JM, Ortiz A.
Fabry disease and COVID-19: international expert recommendations for management based on real-world experience.
Clin Kidney J 2020; 13:913-925
92. Cappellari M, Bonetti B, Forlivesi S, Sajeve G, Naccarato M, Caruso P, Lorenzut S, Merlino G, Viaro F, Pieroni A, Giometto B, Bignamini V, Perini F, De Boni A, Morra M, Critelli A, Tamborino C, Tonello S, Guidoni SV, L'Erario R, Russo M, **Burlina A**, Turinese E, Passadore P, Zanet L, Polo A, Turazzini M, Basile AM, Atzori M, Marini B, Bruno M, Carella S, Campagnaro A, Baldi A, Corazza E, Zanette G, Idone D, Gaudenzi A, Bombardi R, Cadaldini M, Lanzafame S, Ferracci F, Zambito S, Ruzza G,

- Simonetto M, Menegazzo E, Masato M, Padoan R, Bozzato G, Paladin F, Tonon A, Bovi P.
Acute revascularization treatments for ischemic stroke in the Stroke Units of Triveneto, Northeast Italy; time to treatment and functional outcomes.
J Thromb Thrombolysis 2021; 51:159-167
93. Moreno-Martinez D, Aguiar P, Auray-Blais C, Beck M, Bichet DG, **Burlina A**, Cole D, Elliott P, Feldt-Rasmussen U, Feriozzi S, Fletcher J, Giugliani R, Jovanovic A, Kampmann C, Langeveld M, Lidove O, Linhart A, Mauer M, Moon JC, Muir A, Nowak A, Oliveira JP, Ortiz A, Pintos-Morell G, Politei J, Rozenfeld P, Schiffmann R, Svarstad E, Talbot AS, Thomas M, Tøndel C, Warnock D, West ML, Hughes DA.
Standardising clinical outcomes measures for adult clinical trials in Fabry disease: a global Delphi consensus.
Mol Genet Metab 2021; 132:234-243
94. Burlina A, Giuliani A, Polo G, Gueraldi D, Gragnaniello V, Cazzorla C, Opladen T, Hoffmann G, Blau N, **Burlina AP**.
Detection of 3-O-methyldopa in dried blood spots for neonatal diagnosis of aromatic L-amino-acid decarboxylase deficiency: the North-eastern Italian experience.
Mol Genet Metab 2021; 133:56-62
95. Gragnaniello V, **Burlina AP**, Polo G, Giuliani A, Salviati L, Duro G, Cazzorla C, Rubert L, Maines E, Germain DP, Burlina AB.
Newborn screening for Fabry disease in Northeastern Italy: results of five years of experience.
Biomolecules 2021; 11:951
96. Bersano A, Kraemer M, **Burlina A**, Mancuso M, Finsterer J, Sacco S, Salvarani C, Caputi L, Chabriat H, Lesnik Oberstein S, Federico A, Tournier Lasserre E, Hunt D, Dichgans M, Arnold M, Debette S, Markus HS.
Heritable and non-heritable uncommon causes of stroke.
J Neurol 2021; 268:2780-2807
97. Figliozzi S, Camporeale A, Boveri S, Pieruzzi F, Pieroni M, Lusardi P, Spada M, Mignani R, **Burlina A**, Graziani F, Pica S, Tondi L, Bernardini A, Chow K, Namdar M, Lombardi M.
ECG-based score estimates the probability to detect Fabry disease cardiac involvement.
Int J Cardiol 2021; 339:110-117
98. Manganotti P, Naccarato M, Scali I, Cappellari M, Bonetti B, **Burlina A**, Turinese E, Bogo S, Teatini F, Franchini E, Caneve G, Ruzza G, Gaudenzi A, Bombardi R, Bozzato G, Padoan R, Gentile C, Rana M, Turazzini M, Danese A, Brigo F, Nardone R, Quatralo R, Menegazzo E, Masato M, Novello S, Passadore P, Baldi A, Valentinis L, Baracchini C, Pieroni A, Basile AM, Semplicini C, Piffer S, Giometto B, Tonello S, Bonifatti DM, Lorenzuti S, Merlino G, Valente MR, Paladin F, Tonon A, de Luca C, Perini F, Centonze S, Bovi P.

Stroke management during the coronavirus disease 2019 (COVID-19) pandemic: experience from three regions of the north east of Italy (Veneto, Friuli-Venezia-Giulia, Trentino-Alto-Adige).
Neurol Sci 2021; 42:4599-4606

99. Camporeale A, Moroni F, Lazzeroni D, Garibaldi S, Pieroni M, Pieruzzi F, Lusardi P, Spada M, Mignani R, **Burlina A**, Carubbi F, Econimo L, Battaglia Y, Graziani F, Poca S, Chow K, Camici PG, Lombardi M.
Trabecular complexity as an early marker of cardiac involvement in Fabry disease.
Eur Heart J Cardiovasc Imaging 2022; 23:200-208
100. Tinazzi M, Fiorio M, Berardelli A, Bonetti B, Bonifati DM, **Burlina A**, Cagnin A, Calabria F, Corbetta M, Cortelli P, Giometto B, Guidoni SV, Lopiano L, Mancardi G, Marchioretto F, Pellegrini M, Teatini F, tedeschi G, Tesolin L, Turinese E, Zappia M, Marotta A.
Opinion, knowledge, and clinical experience with functional neurological disorders among Italian neurologists: results from an online survey.
J Neurol 2022; 269:2549-2559
101. Gragnaniello V, **Burlina AP**, Manara R, Cazzorla C, Rubert L, Gueraldi D, Toniolli E, Quaia E, Burlina AB.
Bone disease in early detected Gaucher type I disease: a case report.
JIMD Rep Epub Jun 26, 2022

Curatore scientifico del libro:

Neurometabolic Hereditary Diseases of Adults, Springer, 2018.

Capitoli di libri in lingua inglese

1. Bracco F, **Burlina AP**, Scarpa M, Rigo A, Battistin L.
Superoxide dismutase in aging brain and related degenerative diseases.
Adv Biosci 1993; 87: p.169-170
2. **Burlina AP**.
New biochemical markers for leukodystrophy.
In: Uziel G, Taroni F, eds. Hereditary Leukodystrophy and Neuropathies in Childhood: Biological Bases, Diagnosis and Treatment. John Libbey Eurotext. Montrouge, 2003: p.17-21
3. **Burlina AP**.
Neurological phenomena.
In: de Valk HW, Barriento Martinez ZM, eds. Practical Approach to Rare Metabolic Disorders in Adulthood. Lemma Publishers. Utrecht, 2005: p.39-48
4. Burlina A, **Burlina AP**.

Eye disorders.

In: Hoffmann G, Zschocke J, Nyhan WL, eds. Inherited Metabolic Diseases. A Clinical Approach. Springer-Verlag, Berlin Heidelberg, 2010: p.181-196

5. **Burlina A**, Manara R.
MRI and in vivo spectroscopy of the brain.
In: Blau N, Duran M, Gibson KM, Dionisi-Vici C, eds. Physician's Guide to the Diagnosis, Treatment, and Follow-up of Inherited Metabolic Diseases. Springer-Verlag Berlin Heidelberg 2014: p. 803-815
6. Burlina A, Celato A, **Burlina AP**.
Inborn errors of metabolism.
In: Sghirlanzoni A, Lauria G, Chiapparini L, eds. Prognosis of Neurological Diseases. Springer-Verlag Italia, Milano 2015: p. 217-247
7. **Burlina A**, Manara R.
Brain MRI in inherited metabolic diseases of adulthood.
In: Hollak CEM, Lachmann RH, eds. Inherited Metabolic Disease in Adults: A Clinical Guide. Oxford University Press New York, NY, USA 2016: p. 457-465
8. Burlina A, Celato A, **Burlina AP**.
Eye disorders.
In: Hoffmann GF, Zschocke J, Nyhan WL, eds. Inherited Metabolic Diseases: A Clinical Approach, 2nd edition. Springer-Verlag Berlin Heidelberg 2017: p. 319-339
9. Manara R, **Burlina AP**.
Neuroimaging of Inherited Metabolic Diseases of Adulthood
In: Burlina AP. Neurometabolic Hereditary Diseases of Adults. Springer International Publishing AG, Cham, Switzerland 2018: p. 43-66
10. **Burlina AP**, Politei J.
Fabry Disease
In: Burlina AP. Neurometabolic Hereditary Diseases of Adults. Springer International Publishing AG, Cham, Switzerland 2018: p. 67-98

Riviste in lingua italiana

1. **Burlina AP**, Galzigna L.
Preparazione, proprietà e potenzialità terapeutiche della citidin-difosfo-colina associata a liposomi.
Rivista di Neurologia 1989; 59:26-31
2. Rizzoli AA, **Burlina AP**.
Dalla malattia morale alla chimica delle passioni.
Federazione Medica 1990; 43:183-185
3. Burlina AB, **Burlina AP**, Bonafé L, Giordano G, Zacchello F.
Nuove metodologie nella diagnosi delle malattie metaboliche ereditarie.

- Prospettive in Pediatria 1998; 28:37-44*
4. Burlina AB, Catuogno S, **Burlina AP**.
La presa in carico di pazienti con errori
congeniti del metabolismo: una nuova sfida per il futuro.
Saggi Child Development & Disabilities 2002; XXVIII, 1:55-61
 5. Burlina AB, Catuogno S, **Burlina AP**.
Le terapie innovative per le malattie metaboliche ereditarie.
Prospettive in Pediatria 2002; 32:283-293
 6. Ricci R, Castorina M, Di Lillo M, Antuzzi D, Frustaci A, Parini R, Menni F,
Furlan F, Burlina A, **Burlina A**, Catuogno S, Gabrielli O, Burattini I, Borsini
W, Büchner S, Ferriozzi S, Spisni C, De Vito R, Di Rocco M, Aricò M,
Pistone G, Bongiorno AM, Morrone A, Cavicchi C, Zammarchi E.
Fabry disease in Italy: first epidemiologic and collaborative study.
Ann Ital Med Int 2004; 19:269-275
 7. **Burlina A**.
La malattia di Niemann-Pick tipo C: una malattia lisosomiale
neurodegenerativa.
J Clin Med 2012; 12(1):7-12
 8. **Burlina A**.
Omocistinuria. Inquadramento clinico e indicazioni di terapia.
La Neurologia italiana 2014; Anno X (n.1):20-25

Capitoli di libri in lingua italiana

1. **Burlina AP**, Podo F.
Tecniche spettroscopiche: spettroscopia di risonanza
magnetica nucleare.
In: Burlina A. (ed.) Medicina di Laboratorio. Principi di Tecnologia.
C.G. Edizioni Medico Scientifiche. Torino, 1994: p.333-353
2. Saladini M, **Burlina AP**, Battistin L.
Cefalee.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni
Medico Scientifiche. Torino, 1997: p.345-349
3. Saladini M, **Burlina AP**, Battistin L.
Coma.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni
Medico Scientifiche. Torino, 1997: p.352-363
4. Saladini M, **Burlina AP**, Battistin L.
Demenza.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni
Medico Scientifiche. Torino, 1997: p.364-371

5. Saladini M, **Burlina AP**, Battistin L.
Disturbi del gusto e dell'olfatto.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni Medico Scientifiche. Torino, 1997: p.407-410
6. Saladini M, **Burlina AP**, Battistin L.
Disturbi della parola e del linguaggio.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni Medico Scientifiche. Torino, 1997: p.411-414
7. Saladini M, **Burlina AP**, Battistin L.
Disturbi della sensibilità.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni Medico Scientifiche. Torino, 1997: p.415-421
8. Saladini M, **Burlina AP**, Battistin L.
Disturbi visivi e della motilità oculare.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni Medico Scientifiche. Torino, 1997: p.422-431
9. Saladini M, **Burlina AP**, Battistin L.
Dolori, spasmi e crampi muscolari.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni Medico Scientifiche. Torino, 1997: p.457-461
10. Saladini M, **Burlina AP**, Battistin L.
Epilessia.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni Medico Scientifiche. Torino, 1997: p.507-518
11. Saladini M, **Burlina AP**, Battistin L.
Paralisi, paresi e disturbi motori.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni Medico Scientifiche. Torino, 1997: p.921-925
12. Saladini M, **Burlina AP**, Battistin L.
Vertigini e capogiri.
In: Burlina A. (ed.) Guida di Laboratorio per il Medico Pratico. C.G. Edizioni Medico Scientifiche. Torino, 1997: p.1021-1030
13. **Burlina A.**
Cause rare e inusuali di ictus. Malattie metaboliche ereditarie: malattia di Fabry, omocistinuria, aciduria metilmalonica, deficit di ornitina transcarbamilasi.
In: SPREAD, Stroke Prevention and Educational Awareness Diffusion. Ictus cerebrale: linee guida italiane di prevenzione e trattamento. Pubblicazioni Catel – Hyperphar Group SpA 2007: p.540-542
14. **Burlina A.**

Cause rare e inusuali di ictus. Malattie metaboliche ereditarie: malattia di Fabry, omocistinuria, aciduria metilmalonica, deficit di ornitina transcarbamilasi.

In: SPREAD Live (www.spread.it). Stroke Prevention and Educational Awareness Diffusion. Linee Guida Italiane per la prevenzione e il trattamento dell'ictus cerebrale. Cap. 17, v. Jan 7, 2010

15. **Burlina A.**

Cause rare e inusuali di ictus. Malattie metaboliche ereditarie: malattia di Fabry, omocistinuria, aciduria metilmalonica, MELAS

In: SPREAD, Ictus cerebrale: linee guida italiane di prevenzione e trattamento. Raccomandazioni e Sintesi. VIII Edizione, 2016

Bassano del Grappa, 9 settembre 2022