FORMATO EUROPEO PER IL CURRICULUM VITAE

INFORMAZIONI PERSONALI

Nome	ALBERTO BURLINA
E-mail	alberto.burlina@unipd.it
Nazionalità	Italiana
Data e Luogo di nascita	02/07/1955 Motta di Livenza (Tv)

ESPERIENZA LAVORATIVA

12/02/1980 - 29/02/1980 e 01/09/1980 - 30/04/1982
Ospedale Policlinico di Verona
Istituto di Clinica Pediatrica dell'Università di Verona
Tirocinio pratico ospedaliero
Medico Frequentatore

01/05/1982 - 30/04/1989
Ospedale Policlinico di Verona
Centro Regionale per le Malattie Metaboliche Neonatali (disciplina Pediatrica) annesso alla Clinica Pediatrica
Assistente Ospedaliero di ruolo a tempo pieno

01/05/1989 - 29/12/1993
Azienda Ospedaliera di Padova
Dipartimento di Pediatria
Aiuto Corresponsabile Ospedaliero con rapporto di lavoro a tempo pieno

30/12/1993 – 05/12/1996
Azienda Ospedaliera di Padova
Dipartimento di Pediatria
Primo livello Dirigenziale Fascia Sub A, Aiuto Corresponsabile Ospedaliero con rapporto di lavoro a tempo pieno

6/12/1996 – 30/05/2004
Azienda Ospedaliera di Padova
Dipartimento di Pediatria
Dirigente Medico primo livello, Aiuto Corresponsabile Ospedaliero con rapporto di lavoro a tempo unico

01/05/2004 - oggi Azienda Ospedaliera di Padova – Università di Padova

Dirigente Medico secondo livello, Direttore dell' Unità Operativa Complessa Malattie Metaboliche Ereditarie, Dipartimento di Pediatria –
Direttore del Centro Regionale Malattie Metaboliche Ereditarie della Regione Veneto
Direttore del Programma Regionale Screening Neonatale Allargato per le malattie metaboliche ereditarie

ISTRUZIONE E FORMAZIONE

1973
Liceo Scientifico Statale di Conegliano (TV)
Diploma di maturità scientifica con il massimo dei punti

1979
Università di Padova, sede di Verona
Laurea in Medicina e Chirurgia presso l'Università di Padova, sede di Verona con il massimo dei punti

1979
Università di Padova, sede di Verona
Abilitazione all'esercizio della professione di Medico e Chirurgo

1984
Università degli Studi di Verona
Specializzazione in Pediatria con il massimo dei punti e lode

1977-1978
Istituto di Anatomia ed Istologia Patologica dell'Università di Padova per l'anno accademico 1977-78
Allievo interno

1982-1983
Istituto di Chimica Biologica dell'Università di Milano
Comando di perfezionamento

1984-1985
Department of Clinical Biochemistry, Hospital for Sick Children University Of Toronto, Canada
Clinical Fellow

Ottobre- Dicembre 1986
Research Foundation, Department of Clinical Biochemistry, Hospital for Sick Children University Of Toronto, Canada
Visiting Professor

1991-1994
Center for Metabolic Diseases Baylor University Medical Center, Dallas e
Department of Pathology, Children's Medical Center of Dallas, Texas, USA

2014 : Abilitazione Scientifica Nazionale 2012 Seconda Fascia: settore concorsuale 06/G1 Pediatria generale e specialistica

2014 : Abilitazione scientifica Nazionale 2012 Prima Fascia : Settore Concorsuale 05/E1Biochimica Clinica

CAPACITÀ E COMPETENZE RELAZIONALI

APPARTENENZA A SOCIETÀ SCIENTIFICHE

E' membro delle seguenti Società Scientifiche:

- Società Italiana di Pediatria (S.I.P)
- Società Studio Malattie Metaboliche Ereditarie (SISMME) di cui è stato Presidente dal 2003-2010
- Society for the Study of Inborn Errors of Metabolism (SSIEM)
- Society of Inherited Metabolic Disorders (SIMD)

CAPACITÀ E COMPETENZE ORGANIZZATIVE

ATTIVITÀ CLINICA

Per tutta la durata della attività postlaurea, Alberto Burlina ha svolto attività clinica initerrotta di Malattie Metaboliche Ereditarie presso strutture nazionali e internazionali presso le quali ha lavorato. Periodi passati all'estero sono stati infatti associati all'ottenimento dell'abilitazione Canadese, per l'esercizio della professione medica. L'attività assistenziale è sempre stata comprensiva di cura diretta dei pazienti, di guardie diurne e notturne e di reperibilità a seconda delle circostanze. Le attività cliniche prevalenti sono state nel campo delle Malattie Metaboliche Ereditarie a presentazione acuta con particolare interesse alle patologie con forte scompenso metabolico.

Le dimensioni delle strutture presso le quali è stata svolta l'attività clinica sono tra le più grandi e complete (Verona, Padova in Italia; Toronto, Dallas negli USA; Canada) con le più moderne tecniche di assistenza e delle più complete sottospecialità pediatriche.

Dal 01/06/2004 a oggi è Direttore della Unità Operativa Complessa Malattie Metaboliche Ereditarie presso l'Azienda Ospedaliera Universitaria di Padova , del Centro Regionale per le Malattie Metaboliche Ereditarie della Regione Veneto e Direttore del programma Screening Neonatale Allargato per le malattie metaboliche ereditarie. La struttura comprende 6 posti letto dedicati ai pazienti con malattie metaboliche ereditarie, attività ambulatoriale e di Day Hospital. Oltre che da medici pediatri e un neuropsichiatra infantile, lavorano nell'equipe una psicologa e due dietiste. La struttura segue attualmente piu' di 700 pazienti con malattie metaboliche ereditarie.

L'attività e così riassumibile :approccio clinico del paziente metabolico con scompenso in forma acuta. Gestione del paziente in terapie intensive. Gestione del paziente metabolico in fase pre e post trapianto. Gestione del paziente metabolico con danno neurologico. Utilizzo di terapie enzimatiche sostitutive. Utilizzo di terapie cellulari

Competenze cliniche e di laboratorio:

Attualmente l'attività di laboratorio comprende una parte diagnostica e una di ricerca biochimica per le malattie neurologiche ereditarie. Lavorano otto persone tra biologi e tecnici ci di laboratorio

- Diagnosi di laboratorio delle patologie metaboliche con difetto degli
  - Aminoacidi
  - Acidi organici
  - Perossisomi
  - Beta ossidazione degli acidi grassi
  - Lisosomiali
  - Ciclo dell'urea
  - Galattosemia
  - Glicogenosi
- Diagnosi screening neonatale allargato per le malattie metaboliche ereditarie

ATTIVITÀ DI RICERCA

Ha sviluppato svariate linee di ricerca che si sono tradotte nelle pubblicazioni di articoli su qualificate riviste mediche pediatriche nazionali e internazionali.

La quasi totalità della problematica scientifica è stata sviluppata in Italia ed è frutto di idee originali ed iniziative ma non mancano lavori frutto di collaborazioni internazionali.

Ricerche nel campo degli *enzimi lisosomiali*. Tale attività si è svolta in collaborazione con il Dipartimento di Biochimica Medica dell'Università di Milano, direttore Prof. Guido Tettamanti, ed ha riguardato:

- valori di normalità in differenti età, con particolare riferimento alla gravidanza, al neonato a termine, con basso peso gestazionale e nella gemellarità;
- applicazioni diagnostiche nel campo delle malattie metaboliche da accumulo;
- studio degli enzimi lisosomiali, come indici di controllo metabolico, nella patologia diabetica sia in soggetti in terapia insulinica che in pancreas artificiale;
- studio degli enzimi lisosomiali nel liquor in alcune patologie neurologiche e nel prematuro con distress respiratorio.

Ricerche nel campo delle *Malattie MetabolicheEereditarie*. Gli studi condotti sulle Malattie Metaboliche Ereditarie costituiscono il nucleo fondamentale di tutta l'attività scientifica e possono essere così sintetizzate:

a) Studi epidemiologici mediante screening neonatale. Studio multicentrico dell'incidenza nella popolazione italiana della fenilchetonuria ed identificazione delle mutazioni. Studio di frequenza nella popolazione Triveneta del difetto di glucosio 6 fosfato deidrogenasi e di biotinidasi. Attivazione dello screening neonatale esteso nelle province di Belluno e Treviso

b) Studi sul metabolismo intermedio.

Aminoacidopatie.

- iperfenilalaninemia da difetto di guanosintrifosfato ciclo idrolisi I con identificazione biochimica e trattamento terapeutico, e caratterizzazione genetica;
- descrizione di nuovi quadri clinici in alcune aminoacidopatie, protocolli di dietoterapie e studio collaborativo delle mutazioni dei pazienti affetti da leucinosi;
- nuova metodica di laboratorio per il dosaggio dell'acido orotico e sua applicazione clinica per l'identificazione di soggetti affetti da difetto dell'enzima ornitincarbamiltransferasi;
- nuova metodica di dosaggio dell'omocisteina e sue applicazioni

Acidurie organiche.

- metodiche di laboratorio per l'identificazione mediante gas cromatografia-mass spettrometria di metaboliti per la diagnosi di difetti del metabolismo glucidico, difetti della catena respiratoria e di malattie neurodegenerative;
- nuove descrizioni cliniche e segnalazioni di complicanze in soggetti affetti da acidurie organiche; linee guida di trattamento e possibilità di diagnosi prenatale.

Malattie mitocondriali. Nel vasto campo delle malattie mitocondriali, le ricerche si sono svolte allo studio del metabolismo della beta-ossidazione degli acidi grassi con l'identificazione di:

- un nuovo difetto enzimatico a carico delle deidrogenasi idrossilate degli acidi grassi a corta catena;
- descrizione di quadri clinici peculiari e nuove complicazioni nel difetto della deidrogenasi idrossilata degli acidi grassi a lunga catena;
- identificazione del primo paziente italiano con difetto delle deidrogenasi degli acidi grassi a media catena, principale causa metabolica di SIDS;
- descrizione di una nuova sindrome in pazienti italiani con caratteristico quadro clinico, neuroradiologico e peculiari alterazioni biochimiche degli acidi organici.

Malattie dei carboidrati. Descrizione di una nuova malattia con iperinsulinismo ed iperammoniemia.

Nuove terapie per le Malattie Metaboliche Ereditarie.

- applicazioni della terapia enzimatica sostitutiva nelle malattie lisosmiali

- utilizzo della tetraidrobiopterina nella terapia della fenilchetonuria
- Dal 2001 progetto di trapianto cellulare con infusione di cellule epatiche in pazienti affetti da glicogenosi e nei difetti del ciclo dell'urea

Esperienze organizzative di attività scientifiche

- Revisore di pubblicazioni scientifiche per numerose riviste internazionali pediatriche quali J. of Pediatrics, Pediatric Research, Journal of Inherited Metabolic Diseases, Molecular Genetics and Metabolism

ELENCO DELLE PUBBLICAZIONI

1. Burlina A, Rizzotti P, Burlina AB.*A rapid technique for the amylase isoenzymes demonstration*. IRCS Med Sci 8:822,1980.

2. Romeo G, Menozzi P, Ferlini A, Prosperi L, Cerone R, Scalisi S, Romano C,Antonozzi I, Riva E, Piceni Sereni L, Zammarchi E, Lenzi G, Sartorio R, Andria G, Cioni M, Fois A, Burroni M, Burlina AB, Carnevale F. *Incidence of classic PKU in Italy estimated from consanguineous marriages and from neonatal screening.* Clinical Genetics 24:339-45,1983.

3. Burlina A, Zaninotto M, Marini M, Burlina AB. *Adenosine deaminase isoenzymes in serum: an improved spectrophotometric method*. IRCS Med Sci 12:931-2,1984.

4. Lombardo A, Goi G, Pistolesi E, Rocca R, Agosti A, Fabi A, Giuliani G, Burlina AB, Tettamanti G.*Behaviour of several enzymes of lysosomal origin in human plasma during pregnancy*. Clin Chim Acta 143: 253-64,1984.

5. Lombardo A, Goi G, Guagnellini E, Fabi A, Sciorelli G, Burlina AB, Tettamanti G. *Behaviour of several enzymes of lysosomal origin in human plasma during whole blood storage*. Clin Chim Acta 143: 343-53,1984.

6. Goi G, Achilli F, Caimi L, Lombardo A ,Tettamanti G, Burlina AB, Gaburro D. *Levels of some lysosomal activities in newborns. Perspectives in Inherited Metabolic Diseases* 5: 249-53,1984.

7. Burlina A, Zaninotto M, Marini M, Burlina AB. *Adenosine deaminase isoenzymes in serum: an improved spectrophotometric method.* IRCS Med Sci 12:931-2, 1984.

8. Rizzotti P, Cocco C, Burlina A Jr, Marcer V, Plebani M, Burlina A. *Macro creatine kinase type 2:a marker of myocardial damage in infants?* Clin Biochem 18,4:239-41,1985.

9. Goi G, Fabi A, Lorenzi R, Lombardo A, Tettamanti G, Burlina AB, Pinelli L, Gaburro D. *Serum enzymes of lysosomal origin as indicators of the metabolic control in diabetes: comparison with glycated hemoglobin and albumin.* Acta Diabetol Lat XXIII,2:117-25,1986.

10. Goi G, Fabi A, Lombardo A,Burlina AB, Tiby V,Visciani A, Malesani L, Tettamanti G.*Stability of enzymes of lysosomal origin in human cerebrospinal fluid.* Clin Chim Acta163: 215-24,1987.

11. Burlina AB, Goi G, Fabi A, Lombardo A, Gaburro D, Tettamanti G. *Behaviour of some lysosomal enzymes in the plasma of insulin dependent diabetic patients during artificial pancreas treatment*. Clin Biochem 20: 423-7,1987.

12. Goi G, Lombardo A, Fabi A, Burlina AB, Segalini G, Guagnellini E, Tettamanti G. *Serum enzymes of lysosomal origin as indicators of the metabolic control in non-insulin-dependent diabetics.* Acta Diabetol Lat 24, 4: 331-40,1987.

13. Goi G, Fabi A,Lombardo A, Burlina AB, Tettamanti G, Montalbetti N, Cavalleri M and Halberg F. *Circardian and circannual rhythms of several enzymes of lysosomal origin in plasma.* Clin Chim Acta 176, 1-8, 1988.

14. Burlina AB, Sherwood JW, Marchioro MV, Dalla Bernardina B, Gaburro D. *Neonatal screening for biotinidase deficiency in North East Italy*. Eur J Pediat 147: 317-8,1988.

15. Colamaria V, Burlina AB, Gaburro D, Saudubray JM, Merino RG and Dalla Bernardina B. *Biotin-Responsive Infantile Encephalopathy: EEG-Polygraphic Study of a Case*. Epilepsia, 24-29, 1989.

16. Burlina AB , Poletto M, Shin YS, Zacchello F. *Clinic and biochimical observation on three cases of fructose 1,6-dyphosphatase deficiency.* J Inher Metab Dis 13: 263-266, 1990.

17. Burlina AB, Sherwood WG, Zacchello F. *Partial biotinidase deficiency associated with Coffin Siris syndrome*. Eur J Pediatr 149: 628-629,1990.

18. Burlina AB, Zacchello F, Dionisi-Vici C, Bertini E, Sabetta G,Bennett MJ, Hale D, Schmidt E., Rinaldo P. *Biochemical evidence of a branched chain acyl CoA oxidation defect in three patients: a new clinical phenotype.* The Lancet, vol. 331, 1506-1507, 1991.

19. Dionisi-Vici C, Burlina AB, Bertini E, Bachmann C, Mazziotta MC, Zacchello F, Sabetta G, Hale D. *Progressive*

neuropathy and recurrent myoglobinuria in a child with long-chain 3-hydroxyacyl-coenzyme a dehydrogenase deficiency. J Pediatr 118, 5, 744-746, 1991.

20. Burlina AB, Bachmann C, Wermuth B, Bordugo A, Ferrari V, Colombo JP, Zacchello F. *Partial N-acetyl glutamate synthetase deficiency: a new case with uncontrollable movement disordens.* J. Inher Metab. Dis. 15 395-398,1992.

21. Burlina AB, Ferrari V, Dionisi Vici C, Bordugo A, Zacchello F, Tuchman M. *Allopurinol challenge test in children*.J Inher Metab Dis 15 707-712,1992

22. Bertini E, Dionisi-Vici C, Garavaglia B, Burlina AB, Sabatelli M, Rimoldi M, Bartulli A, Sabetta G, and Di Donato S. *Peripheral sensory-motor polyneuropathy, pigmentary retinopathy, and fatal cardiomyopathy in long-chain 3-Hydroxy-acyl-CoA dehydrogenase deficiency.* Eur J Pediatr 151: 121-126, 1992.

23. Mazziotta MRM, Ricci E, Bertini E, Dionisi-Vici C, Servidei S, Burlina AB, Sabetta G, Bartulli A, Manfredi G, Silvestri G, Moraes CT, Di Mauro S. *Fatal infantile liver failure associated with mitochondrial DNA depletion.* J Pediatr 121: 896-901, 1992.

24. Bennett MJ, Sherwood WG, Gibson KM, Burlina AB. *Secondary inhibition of multiple NAD-requiring dehydrogenases in respiratory chain complex I deficiency: possible metabolic markers for the primary defect.* J Inher Metab Dis 6: 537-538, 1993.

25. Burlina AB, Milanesi O, Biban P, Bordugo A, Garavaglia B, Zacchello F, Di Mauro S *Beneficial effect of sodium dicloracitate in muscle cytochrome a oxidase deficiency*. Eur J Pediatr 6: 537-538, 1993.

26. Goi G, Burlina AB, Bairati C, Bordugo A, Zanardo V, Zacchello F, Tettamanti G, Lombardo A. *Enzymes of lysosomal origin in plasma of twin neonates.* Clin Chim Acta. 214, 61-71, 1993.

27. Goi G, Caputo D,Bairati C, Lombardo A, Burlina AB, Ferrante P, Cazzullo CC, Tettamanti G. *Enzymes of lysosomal origin in the Cerebrospinal fluid and plasma of patients with multiple sclerosis.* Eur Neurol 33: 1-4, 1993.

28. Drigo P, Burlina AB, Battistella PA. *Subdural hematoma and glutaric aciduria type 1*. Brain & Develop 15, 460-561, 1993.

29. Burlina AB, Dionisi-Vici C, Bennett MJ, Gibson KM, Juidei S, Bertini E, Hale D, Schmidt-Sommerfeld E, Sabetta G, Zacchello F, Rinaldo P. *A new syndrome with ethylmalonic aciduria and normal fatty acid oxidation in fibroblasts*. J Pediatr 124, 79-86, 1994.

30. Burlina AB, Rinaldo P. *Encephalopathy, petechiae, and acrocyanosis with ethylmalonic aciduria associated with muscle cytochrome c oxidase deficiency* J Pediatr 125,843-845,1994 (Letter)

31. Burlina AP, Cavazza A, Ferrari V, Erhard P, Kunnecke K, Sceeling J, Burlina AB. *Detection of increased urinary N-acetyl aspartylglutamate in Canavan disease.* Eur J Pediatr, 52, 538-539, 1994.

32. Burlina AP, Skaper S, Mazza MR, Ferrari V, Leon A and Burlina AB. *N-acetylaspartylglutamate selectively inhibits neuronal responses to N-methyl D-aspartic acid in vitro. J* Neurochem 63, 1174-1177, 1994.

33. Ponzone A, Ferrari S, Spada M, Blau N, Piovan S, Burlina A. *Combined phenylalanine-tetrahydrobiopterin loading test in GTP cyclohydrolase deficiency.* Eur J Pediatr 52, 611-612, 1994.(Letter)

34. Bennett MJ, Weinberger MJ, Sherwood WG and Burlina AB. *Secondary 3 hydroxy dicarboxylic aciduria mimicking long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency.* J Inher Metab Dis 17, 3, 283-286, 1994.

35. Salamino F, Di Lisa F, Burlina AB, Menabò R, Barbato R, De Tullio R, Silprandi N. *Involment of erythrocyte calpain in glycine amd carnitine treated isovaleric acidemia.* Pediatr Res 36: 182-186, 1994.

36. Parrella T, Surrey S, Iolascone A, Sartore M, Heidenreich R, Framond G, Ponzone A, Guardamagna O, Burlina AB, Cerone R, Parini R, Dionisi-Vici C, Rappeport E and Fortina P. *Maple syrupe urine disease (MSUD) screening for known mutations in italian patients.* J Inher Metab Dis 17, 652-660,1994.

37. Blau N, Ichinase H, Nagatsu T, Heizmann CW, Zacchello F and Burlina AB. *A misseuse mutation in a patient with guanosine triphosphate cyclohydrolase i deficiency missed in the newborn screening program*. J Pediatr 126,401-405,1995.

38. chinose H, Ohye T, Matsuda Y, Hori T, Blau N, Burlina AB, Rouse B, Matalon R, Fujita K and Nagatsu T. *Characterization of mouse and human GTP cyclohydrolase i genes.* J Biol Chem 270, 17,10062-10071,1995.

39. Burlina AB, Bennett MJ, Gregersen N, Dalla Barba B, Zacchello F. *Medium-chain acyl coa dehydrogenase deficiency presenting in the neonatal period: the first italian case.* Eur J Pediatr 154,950-941,1995.(Letter)

40. Burlina AB, Dionisi-Vici C, Piovan S, Saponara I, Bartuli A, Sabetta G and Zacchello F. *Acute pancreatitis in propionic acidemia.* J Inher Metab Dis, 18, 169-172, 1995.

41. Mitchell GA, Jakobs C, Gibson KM, Robert MF, Burlina AB, Dionisi-Vici C and Dallaire L. *Molecular prenatal diagnosis of 3-hydroxy-3-methylglutaryl coa lyase deficiency.* Prenat Diagn 15,725-729,1995.

42. Dianzani I, Giarmattosio S, De Santis SC, Alliaudi C, Cattaurio P, Dionisi-Vici C, Burlina A.B. Burroni M,Sebastio G, Carnevale F, Guzzetta V, Marra E, Camaschella C, Ponzone A. *Characterization of phenylaketonuria alleles in the italian population.* Eur J Hum Genet 3,294-302,1995.

43. Hoffmann GF, Bohles H, Burlina AB, Duran M, Herewig J, Lenhert W, Leonard J,Mantau A, Plecko-Starting K, Superti Furga A, Tretz FK and Christensen E. *Early signs and course of disease of glutaryl-coa dehydrogenase deficiency*. J Inher Metab Dis 18,173-176,1995.

44. Lombardo A, Bairati C, Goi G, Roggi C, Maccarini L,Bollini D, Burlina AB *Plasma lysosomal glycohydrolases in a general population.* Clin Chim Acta 247, 39-49,1996

45. Bennett MJ, Weinberger MJ, Kobouri JA, Rinaldo P, Burlina AB *Mithocondrial short-chain l-3-hydroxyacyl-Coenzyme A dehydrogenase deficiency: a new defect of fatty acid oxidation* Pediatr Res 39: 185-188, 1996

46. Piva E, De Toni S, Bovo C, Bordugo A, Burlina AB, Plebani M. *NADPH oxidase activity and chemotaxis by neutrophilis in two patients with glycogen storage disease type Ib treated with recombinant human granulocyte-monocyte-colony-stimulating factor.* Hematologica 81,2:148-151, 1996

47. Burlina AB, Dermikol M, Mantau A, Piovan S, Grazian L, Zacchello F and Shin Y. *Increased plasma biotinidase activity in patients with glycogen storage disease type Ia: effect of biotin supplementation* J Inher Metab Dis.19,209-212, 1996

48. Caruso U, Adami A,Bertini E,Burlina AB,Carnevale F,Cerone R,Dionisi-Vici C,Giordano G,Leuzzi E,Parenti G,Savasta S,Uziel G and Zeviani M. *Respiratory-chain and pyruvate metabolism defects:Italian collaborative survey on 72 patients* J. Inher.Metab.Dis. 19:143-148,1996

49. Drigo P,Piovan S,Batistella PA,Dalla Puppa A,Burlina A:B. *Macrocephaly,subaracnoid fluid collection and glutaric aciduria type I* J. Child Neur 11,5:414-417,1996

50. Dionisi-Vici CD, Garavaglia B, Burlina AB, Bertini E, Saponara I, Sabetta G. *Hypoparathyroidism in mitochondrial trifunctional protein deficiency* J Pediatr, 129;159-162,1996

51. Simioni-P; Prandoni-P; Burlina-A; Tormene-D; Sardella-C; Ferrari-V; Benedetti-L; Girolami-A *Hyperhomocysteinemia and deep-vein thrombosis - A case-control study Thrombosis-and-Haemostasis.* 76 (6) : 883-886 1996

52. Hoffmann-GF; Athanassopoulos-S; Burlina-AB; Duran-M; deKlerk-JBC; Lehnert-W; Leonard-JV; Monavari-AA; Muller-E; Muntau-AC; Naughten-ER; PleckoStarting-B; SupertiFurga-A; Zschocke-J; Christensen-E *Clinical course, early diagnosis, treatment, and prevention of disease in glutaryl-CoA dehydrogenase deficiency* Neuropediatrics;27 (3) : 115-123 1996

53. Burlina-AP; Ferrari-V; Facci-L; Skaper-SD; Burlina-AB *Mast cells contain large quantities of secretagogue-sensitive N-acetylaspartate* Journal-of-Neurochemistry.; 69 (3) : 1314-1317 1997

54. Burlina-AB; Gibson-KM; Ruitenbeek-W; Bonafe-L; Bennett MJ. *Profound neurological phenotype in a patient presenting with disordered isoleucine and energy metabolism* Journal-of-Inherited-Metabolic-Disease. 21 (8) : 864-866,1998

55. Goi-G; Bairati-C; Massaccesi-L; Lombardo-A; Bonafe-L; Zanardo-V; Burlina-A *Lysosomal enzymes in preterm infants with bronchopulmonary dysplasia: a potential diagnostic marker* Clinica-Chimica-Acta. 278 (1) : 23-34, 1998

56. Pagani-F; Pariyarath-R; Garcia-R; Stuani-C; Burlina-AB; Ruotolo-G; Rabusin-M; Baralle *New lysosomal acid lipase gene mutants explain the phenotype of Wolman disease and cholesteryl ester storage disease* Journal-of-Lipid-Research. 39 (7) : 1382-1388,1998

57. Stanley-CA; Lieu-YK; Hsu-BYL; Burlina-AB; Greenberg-CR; Hopwood-NJ; Perlman-K; Rich-BH; Zammarchi-E; Poncz-M *Hyperinsulinism and hyperammonemia in infants with regulatory mutations of the glutamate dehydrogenase gene* New-England-Journal-of-Medicine. 338 (19) : 1352-1357,1998

58. Otto-LR; Boriack-RL; Marsh-DJ; Kum-JB; Eng-C; Burlina-AB; Bennett-MJ *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)* American-Journal-of-Medical-Genetics. 83 (1) : 3-5,1999

59. Avogaro-A; Calo-L; Piarulli-F; Miola-M; deKreutzenberg-S; Maran-A; Burlina-A; Mingardi-R; Tiengo-A; Del-Prato-S *Effect of acute ketosis on the endothelial function of type 1 diabetic patients - The role of nitric oxide Diabetes*. 48 (2) : 391-397,1999

60. Goi-G; Bairati-C; Segalini-G; Burlina-AB; Massaccesi-L; Lovagnini-A; Lombardo-A *Alterations in the activity of several glycohydrolases in red blood cell membrane from type 2 diabetes mellitus patients Metabolism*-Clinical-and-Experimental. Jul 1999; 48 (7) : 817-821

61. Arranz-JA; Riudor-E; Rodes-M; Roig-M; Climent-C; Rubio-V; Sentis-M; Burlina-A *Optimization of allopurinol challenge: Sample purification, protein intake control, and the use of orotidine response as a discriminative variable improve performance of the test for diagnosing ornithine carbamoyltransferase deficiency* Clinical-Chemistry. Jul 1999; 45 (7) : 995-1001

62. Burlina-AB; Bonafe-L; Zacchello-F *Clinical and biochemical approach to the neonate with a suspected inborn error of amino acid and organic acid metabolism* Seminars-in-Perinatology. Apr 1999; 23 (2) : 162-173

63. 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-Journal-Pediatrics. May 1999; 158 (5) : 406-409

64. Vergani L; Barile M; Angelini C; Burlina AB; Nijtmans L; Freda MP; Brizio C; Zerbetto E. Dabbeni Sala F*Riboflavin therapy - Biochemical heterogeneity in two adult lipid storage myopathies* Brain Dec 1999; 122 (12) : 2401-2411

65. 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-Ped. 1999; 158 (5) : 406-409

66. Jones-PM; Quinn-R; Fennessey-PV; Tjoa-S; Goodman-SI; Fiore-S; Burlina-AB; Rinaldo-P, Boriack-RL Bennett-MJ *Improved stable isotope dilution-gas chromatography-mass spectrometry method for serum or plasma free 3-hydroxy-fatty acids and its utility for the study of disorders of mitochondrial fatty acid beta-oxidation* Clin Chem. Feb 2000; 46 (2) : 149-155

67. Riudor E., Arranz J.A., Rodes M., Rubio V., Sentis M.,.Burlina A. *Influence of dose and age on the response of the alloparinol test for carbamoyltransferase deficiency in control infants* J Inher Metab Dis 23 (7) 662-668 2000

68. Burlina A.B., Bonafè L., Ferrari V., Suppiej A., Zacchello F., Burlina A.P. *Measurement of neurotransmitter metabolites in the cerebrospinal fluid of phenylketonuric patients under dietary treatment.* J Inher Metab Dis 23(4): 313-316. 2000

69. Cesari M., Burlina A, Narkiewicz K., Sartori M.T., Sacchetto A., Rossi G.P. *Are fasting plasma homocyst(e)ine levels heritable? A study of normotensive twins* Journal of investigative medicine 48 (5) 351-358 2000

70. Favaro A., Caregaro L., Burlina A, Santonastaso P. *Tryptophan levels, excessive exercise and nutritional status in anorexia nervosa* Psychosomatic medicine 62 (4) 535-538 2000

71. Jones P.M., Burlina A, Bennett M.J. *Quantitative measurement of total and free 3-hydroxy fatty acidsin serum or plasma samples short chain 3 - hydroxy fatty acids are not esterified* J Inher Metab Dis, 23 (7) 745-750 2000

72. . Cella G., Burlina A, Sharai A., Motta G., Girolami A., Berrettini M., Strass W. T*issue factor pathway inhibitor levels in patients with homocystinuria* Thrombosis research 98, (5) 375-381 2000

73. Di Rocco M., Barone R., Adami A., Burlina A, Carrozzi M., Dionisi -Vici C.,Gatti R., Jannetti P., Parini R., Rauchi U.,

Roccella M., Spada M., Fiumara A. *Carbohydrate-deficient glycoprotein syndromes: The Italian experience.* J Inher Metab Dis, 23 (4) 391-395 2000

74. Goi G., Bairati C., Burlina A, Massaccesi L., Monciotti C., Segalini G., Testa R., Lombardo A. *Plasma glycohydrolase levels in patients with type 1 diabetyes at onset and in subjects undergoing an intravenous glucose tolerance test Metabolism Clinical and Experimental* 49 (10): 1352-1355 2000

75. Jones P.M., Quinn R., Fennessey P.V., Tjoa S., Goodman S.I., Fiore S., Burlina A, Rinaldo P. Boriackr.I., Bennett M.J. *Improved stable isotope dilution gas cromatography mass spectropetry method for serum or plasma free 3-hydroxy-fatty acids and its utility for the study of disorders of mitochondrial fatty acid beta-oxidation* Clin Chem 46 (2), 149-155 2000

76. Bonafè L., Troxder H., Kuster T., Heizmann C.-W., Chamoles N.A. Burlina A.B., Blau N. *Evaluation of urinary acylglycines by electrospray tandem mass spectrometry in mitochondrial energy metabolism defects and organic aciduria* Molecular Genetics and Metabolism 69 (4) 302-311 2000

77. Salvi S, Santorelli Fm, Bertini E, Boldrini R, Meli C, Donati A, Burlina A.B.,Rizzo C, Di Capua M, Fariello G, Dionisi-Vici C. *Clinical and molecular findings in hyperornithinemia-hyperammonemia-homocitrullinuria syndrome.* Neurology. 11;57(5):911-4 2001

78. Bonafe L, Blau N, Burlina Ap, Romstad A, Guttler F, Burlina Ab. T*reatable neurotransmitter deficiency in mild phenylketonuria.* Neurology 11;57(5):908-11 2001

79. Burlina Ab, Ogier H, Korall H, Trefz Fk. *Long-term treatment with sodium phenylbutyrate in ornithine transcarbamylase-deficient patients*. Mol Genet Metab. 72(4):351-5 2001

80. Giaquinto C, De Romeo A, Giacomet V, Rampon O, Ruga E, Burlina A, De Rossi A, Sturkenboom M, D'elia R. *Lactic acid levels in children perinatally treated with antiretroviral agents to prevent HIV transmission.* AIDS. 25;15(8):1074-5 2001

81. Burlina AP; Baracchini C; Carollo C; Burlina AB *Propionic acidaemia with basal ganglia stroke: Treatment of acute extrapyramidal symptoms with L-DOPA* J Inher Metab Dis 596-598 2001

82. Invernizzi F.; Burlina AB.; Donadio A.; Giordano G.; Taroni F.; Garavaglia B. *Lethal neonatal presentation of carnitine palmitoyltransferase I deficiency J Inher Metab Dis*. 24 (5) : 601-602 2001

83. Burlina A.B., G. Giordano, S. Catuogno, F. Zacchello. *The role of tandem mass spectrometry in the diagnosis of metabolic diseases: the experience in the Veneto area*. Ital J Pediatr 2001; 27: 766-773 766

84. Cavalleri-F; Berardi-A; Burlina-AB; Ferrari-F; Mavilla-L *Diffusion-weighted MRI of maple syrup urine disease encephalopathy Neuroradiology-*. JUN 2002; 44 (6) : 499-502

85. Burlina-AP; Edini-C; Burlina-AB *Treatment of extrapyramidal symptoms in a patient with homozygous homocystinuria* J Inher Metab Dis. 2002; 25 (2) : 135-136

86. Muraca-M; Gerunda-G; Neri-D; Vilei-MT; Granato-A; Feltracco-P; Meroni-M; Giron-G; Burlina-AB *Hepatocyte transplantation as a treatment for glycogen storage disease type 1a Lancet-*. JAN 26 2002; 359 (9303) : 317-318

87. Dionisi-Vici-C; Rizzo-C; Burlina-AB; Caruso-U; Sabetta-G; Uziel-G; Abeni-D *Inborn errors of metabolism in the Italian pediatric population: A national retrospective survey* J Ped. 2002; 140 (3) : 321-327

88. Hellerud-C; Burlina-A; Gabelli-C; Ellis-JR; Nyholm-PG; *Lindstedt-S Glycerol metabolism and the determination of triglycerides - Clinical, biochemical and molecular findings in six subjects*Clin-Chem-Lab-Med. 2003; 41 (1) : 46-55

89. 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 Inher Metab Dis 2003; 26 (5) : 417-422

90. Zannolli R, Micheli V, Mazzei MA, Sacco P, Piomboni P, Bruni E, Miracco C, de Santi MM, Terrosi Vagnoli P, Volterrani L, Pellegrini L, Livi W, Lucani B, Gonnelli S, Burlina AB, Jacomelli G, Macucci F, Pucci L, Fimiani M, Swift JA, Zappella M, Morgese G. *Hereditary xanthinuria type II associated with mental delay, autism, cortical renal cysts, nephrocalcinosis, osteopenia, and hair and teeth defect*s. J Med Genet. 2003 Nov;40(11):e121

91. Rugolotto S, Prioli MD, Toniolo D, Pellegrino P, Catuogno S, Burlina AB. *Long-term treatment of Barth syndrome*

with pantothenic acid: a retrospective study. Mol Genet Metab. 2003 Dec;80(4):408-11.

92. Zanardo V, Caroni G, Burlina A. *Higher homocysteine concentrations in women undergoing caesarean section under general anesthesia.* Thromb Res. 2003;112(1-2):33-6.

93. Zannolli R, Buoni S, Macucci F, Miracco C, de Santi MM, Piomboni P, Bruni E, Malandrini A, Galluzzi P, Hadjistilianou T, Medaglini S, Mazzei MA, Sacco P, Terrosi-Vagnoli P, Volterrani L, Molinelli M, Burlina AB, Swift JA, Fimiani M. *Hypertelorism, ptosis, and myopia associated with drug-resistant epilepsy, mental delay, growth deficiency, ectodermal defects, and osteopenia*. Am J Med Genet A. 2004 Jun 1;127A(2):212-6.

94. Burlina AB. *Hepatocyte transplantation for inborn errors of metabolism*. J Inherit Metab Dis. 2004;27(3):373-83.

95. Suormala T, Baumgartner MR, Coelho D, Zavadakova P, Kozich V, Koch HG, Berghaüser M, Wraith JE, Burlina A, Sewell A, Herwig J, Fowler B. *The cblD defect causes either isolated or combined deficiency of methylcobalamin and adenosylcobalamin synthesis.* J Biol Chem. 2004 Oct 8;279(41):42742-9. Epub 2004 Aug 2.

96. Boltshauser E, Schmitt B, Wevers RA, Engelke U, Burlina AB, Burlina AP. F*ollow-up of a child with hypoacetylaspartia*. Neuropediatrics. 2004 Aug;35(4):255-8.

97. Baldas V, Not T, Tommasini A, Ansaldi F, Demarini S, Sblattero D, Marzari R, Torelli L, Burlina A, Tiribelli C, Ventura A.*Anti-transglutaminase antibodies and age.*Clin Chem. 2004 Oct;50(10):1856-60.

98. 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(6):911-5.

99. Cesari M, Zanchetta M, Burlina A, Pedon L, Maiolino G, Sticchi D, Pessina AC, Rossi GP. *Hyperhomocysteinemia is inversely related with left ventricular ejection fraction and predicts cardiovascular mortality in high-risk coronary artery disease hypertensives.* Arterioscler Thromb Vasc Biol. 2005 Jan;25(1):115-21. Epub 2004 Nov 4.

100. Maran A, Crepaldi C, Avogaro A, Catuogno S, Burlina A, Poscia A, Tiengo A. *Continuous glucose monitoring in conditions other than diabetes*. Diabetes Metab Res Rev. 2004 Nov-Dec;20 Suppl 2:S50-5.

101. 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, Buchner 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 Oct-Dec;19(4):269-75. Italian.

102. Zaffanello M, Zamboni G, Maselli M, Gandini A, Camilot M, Maffeis C, Burlina AB, Tatò L. *Genetic analysis carried out on blood-spots of phenylalanine hydroxylase-deficient newborns detected by northeastern Italian neonatal screening.* Genet Test. 2005 Summer;9(2):133-7.

103. Melis D, Parenti G, Gatti R, Casa RD, Parini R, Riva E, Burlina AB, Vici CD, Di Rocco M, Furlan F, Torcoletti M, Papadia F, Donati A, Benigno V, Andria G. *Efficacy of ACE-inhibitor therapy on renal disease in glycogen storage disease type 1: a multicentre retrospective study*. Clin Endocrinol (Oxf). 2005 Jul;63(1):19-25.

104. Goi G, Massaccesi L, Burlina AP, Baquero Herrera CJ, Lombardo A, Tettamanti G, Burlina AB. *Lysosomal leukocyte beta-D-glucuronidase during enzyme replacement therapy in Fabry disease.* Biochim Biophys Acta. 2005 Sep 25;1741(3):300-6.

105. Dardis A, Zampieri S, Filocamo M, Burlina A, Bembi B, Pittis MG. *Functional in vitro characterization of 14 SMPD1 mutations identified in Italian patients affected by Niemann Pick Type B disease*. Hum Mutat. 2005 Aug;26(2):164.

106. Tiranti V, Briem E, Lamantea E, Mineri R, Papaleo E, De Gioia L, Forlani F, Rinaldo P, Dickson P, Abu-Libdeh B, Cindro-Heberle L, Owaidha M, Jack RM, Christensen E, Burlina A, Zeviani M. *ETHE1 mutations are specific to ethylmalonic encephalopathy*. J Med Genet. 2006 Apr;43(4):340-6. Epub 2005 Sep 23.

107. Zannolli R, Buoni S, Macucci F, Santi MM, Miracco F, Pierluigi M, Mogni M, Piomboni P, Massafra MR, Galluzzi P, Livi W, Cuccia A, Margollicci MA, Pucci L, Sacco P, Molinelli M, Burlina AB, Swift JA, Fimiani M, Zappella M, Miracco C. *Global developmental delay, osteopenia and ectodermal defect: a new syndrome.* Brain Dev. 2006 Apr;28(3):155-61. Epub 2005 Dec 20.

108. Muraca M, Burlina AB. *Liver and liver cell transplantation for glycogen storage disease type IA*. Acta Gastroenterol

Belg. 2005 Oct-Dec;68(4):469-72.

109. Cesaro S, Zignol M, Burlina AB, Tridello G, Visintin G, Messina C. *Assessment of nephrotoxicity of high-cumulative dose of liposomal amphotericin B in a pediatric patient who underwent allogeneic bone marrow transplantation.* Pediatr Transplant. 2006 Mar;10(2):255-8.

110. Burlina AB, Peduto A, Di Palma A, Bellizzi A, Sperlì D, Morrone A, Burlina AP. *An unusual clinical and biochemical presentation of ornithine transcarbamylase deficiency in a male patient.* J Inherit Metab Dis. 2006 Feb;29(1):179-81.

111. Rossi GP, Maiolino G, Seccia TM, Burlina A, Zavattiero S, Cesari M, Sticchi D, Pedon L, Zanchetta M, Pessina AC. *Hyperhomocysteinemia predicts total and cardiovascular mortality in high-risk women.* J Hypertens. 2006 May;24(5):851-9.

112. Kölker S, Garbade SF, Greenberg CR, Leonard JV, Saudubray JM, Ribes A, Kalkanoglu HS, Lund AM, Merinero B, Wajner M, Troncoso M, Williams M, Walter JH, Campistol J, Martí-Herrero M, Caswill M, Burlina AB, Lagler F, Maier EM, Schwahn B, Tokatli A, Dursun A, Coskun T, Chalmers RA, Koeller DM, Zschocke J, Christensen E, Burgard P, Hoffmann GF.*Natural history, outcome, and treatment efficacy in children and adults with glutaryl-CoA dehydrogenase deficiency.* Pediatr Res. 2006 Jun;59(6):840-7. Epub 2006 Apr 26.

113. 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;576:283-7; discussion 361-3.

114. Burlina AP, Ferrari V, Burlina AB, Ermani M, Boespflug-Tanguy O, Bertini E; Clinical European Network on Brain Dysmyelinating Disease. *N-acetylaspartylglutamate (NAAG) in Pelizaeus-Merzbacher disease. Adv Exp Med Biol.* 2006;576:353-9; discussion 361-3.

115. Valente L, Tiranti V, Marsano RM, Malfatti E, Fernandez-Vizarra E, Donnini C, Mereghetti P, De Gioia L, Burlina A, Castellan C, Comi GP, Savasta S, Ferrero I, Zeviani M. I*nfantile encephalopathy and defective mitochondrial DNA translation in patients with mutations of mitochondrial elongation factors* EFG1 and EFTu. Am J Hum Genet. 2007 Jan;80(1):44-58. Epub 2006 Nov 15. Erratum in: Am J Hum Genet. 2007 Mar;80(3):580.

116. 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 RA, Wilcken B, Hoffmann GF, Burgard P. *Guideline for the diagnosis and management of glutaryl-CoA dehydrogenase deficiency (glutaric aciduria type I).*J Inherit Metab Dis. 2007 Feb;30(1):5-22. Epub 2007 Jan 3.

117. Trevisson E, Salviati L, Baldoin MC, Toldo I, Casarin A, Sacconi S, Cesaro L, Basso G, Burlina AB.*Argininosuccinate lyase deficiency: mutational spectrum in Italian patients and identification of a novel ASL pseudogene.* Hum Mutat. 2007 Jul;28(7):694-702.

118. 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 Apr;30(2):153-8. Epub 2007 Feb 27.

119. 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 gen*e. Eur J Paediatr Neurol. 2008 Jul;12(4):348-50. Epub 2007 Sep 18.

120. Zurflüh MR, Zschocke J, Lindner M, Feillet F, Chery C, Burlina A, Stevens RC, Thöny B, Blau N. *Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency.* Hum Mutat. 2008 Jan;29(1):167-75.

121. Regis S, Biancheri R, Bertini E, Burlina A, Lualdi S, Bianco MG, Devescovi R, Rossi A, Uziel G, Filocamo M. *Genotype-phenotype correlation in five Pelizaeus-Merzbacher disease patients with PLP1 gene duplications.* Clin Genet. 2008 Mar;73(3):279-87. Epub 2007 Jan 9.

122. 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 May;255(5):738-44. Epub 2008 Feb 26.

123. Zwickler T, Lindner M, Aydin HI, Baumgartner MR, Bodamer OA, Burlina AB, Das AM, DeKlerk JB, Gökcay G, Grünewald S, Guffon N, Maier EM, Morava E, Geb S, Schwahn B, Walter JH, Wendel U, Wijburg FA, Müller E, Kölker S, Hörster F. *Diagnostic work-up and management of patients with isolated methylmalonic acidurias in European metabolic centres.* J Inherit Metab Dis. 2008 Jun;31(3):361-7. Epub 2008 May 27.

124. Mineri R, Rimoldi M, Burlina AB, Koskull S, Perletti C, Heese B, von Döbeln U, Mereghetti P, Di Meo I, Invernizzi F, Zeviani M, Uziel G, Tiranti V. I*dentification of new mutations in the ETHE1 gene in a cohort of 14 patients presenting with ethylmalonic encephalopathy.* J Med Genet. 2008 Jul;45(7):473-8.

125. Trevisson E, Salviati L, Baldoin MC, Casarin A, Basso G, Burlina A. *Gene symbol: ASL. Disease: Argininosuccinate lyase deficiency.* Hum Genet. 2008 Oct;124(3):303.

126. Fowler B, Burlina A, Kozich V, Vianey-Saban C. *Quality of analytical performance in inherited metabolic disorders: the role of ERNDIM.* J Inherit Metab Dis. 2008 Dec;31(6):680-9. Epub 2008 Nov 21. Erratum in: J Inherit Metab Dis. 2009 Feb;32(1):127.

127. Burlina A, Blau N. E*ffect of BH(4) supplementation on phenylalanine tolerance.* J Inherit Metab Dis. 2009 Feb;32(1):40-5. Epub 2008 Dec 9.

128. Bortot B, Barbi E, Biffi S, Lunazzi G, Bussani R, Burlina A, Norbedo S, Ventura A, Carrozzi M, Severini GM.*Two novel POLG mutations causing hepatic mitochondrial DNA depletion with recurrent hypoketotic hypoglycaemia and fatal liver dysfunction.* Dig Liver Dis. 2009 Jul;41(7):494-9. Epub 2009 Feb 4.

129. Meyburg J, Das AM, Hoerster F, Lindner M, Kriegbaum H, Engelmann G, Schmidt J, Ott M, Pettenazzo A, Luecke T, Bertram H, Hoffmann GF, Burlina A. *One liver for four children: first clinical series of liver cell transplantation for severe neonatal urea cycle defects.*Transplantation. 2009 Mar 15;87(5):636-41.

130. Cermàkovà I, Pettenazzo A, Perrone S, Burlina A, Zannin ME. *Grade III lipaemia retinalis in a newborn.* Acta Ophthalmol. 2010 Jun;88(4):e141-2. Epub 2009 Jun 26.

131. Hörster F, Garbade SF, Zwickler T, Aydin HI, Bodamer OA, Burlina AB, Das AM, De Klerk JB, Dionisi-Vici C, Geb S, Gökcay G, Guffon N, Maier EM, Morava E, Walter JH, Schwahn B, Wijburg FA, Lindner M, Grünewald S, Baumgartner MR, Kölker S. *Prediction of outcome in isolated methylmalonic acidurias: combined use of clinical and biochemical parameters.* J Inherit Metab Dis. 2009 Oct;32(5):630-9. Epub 2009 Jul 31. Erratum in: J Inherit Metab Dis. 2009 Dec;32(6):762-3.

132. 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 Dec;51(12):803-12. Epub 2009 Aug 4.

133. Trevisson E, Burlina A, Doimo M, Pertegato V, Casarin A, Cesaro L, Navas P, Basso G, Sartori G, Salviati L.*Functional complementation in yeast allows molecular characterization of missense argininosuccinate lyase mutations.* J Biol Chem. 2009 Oct 16;284(42):28926-34. Epub 2009 Aug 24.

134. Leuzzi V, Carducci CA, Carducci CL, Pozzessere S, Burlina A, Cerone R, Concolino D, Donati MA, Fiori L, Meli C, Ponzone A, Porta F, Strisciuglio P, Antonozzi I, Blau N. *Phenotypic variability, neurological outcome and genetics background of 6-pyruvoyl-tetrahydropterin synthase deficiency*. Clin Genet. 2010 Mar;77(3):249-57. Epub 2009 Jan 3

135. Willemsen MA, Verbeek MM, Kamsteeg EJ, de Rijk-van Andel JF, Aeby A, Blau N, Burlina A, Donati MA, Geurtz B, Grattan-Smith PJ, Haeussler M, Hoffmann GF, Jung H, de Klerk JB, van der Knaap MS, Kok F, Leuzzi V, de Lonlay P, Megarbane A, Monaghan H, Renier WO, Rondot P, Ryan MM, Seeger J, Smeitink JA, Steenbergen-Spanjers GC, Wassmer E, Weschke B, Wijburg FA, Wilcken B, Zafeiriou DI, Wevers RA. *Tyrosine hydroxylase deficiency: a treatable disorder of brain catecholamine biosynthesis.* Brain. 2010 Jun;133(Pt 6):1810-22. Epub 2010 Apr 29.

136. Brun L, Ngu LH, Keng WT, Ch'ng GS, Choy YS, Hwu WL, Lee WT, Willemsen MA, Verbeek MM, Wassenberg T, Régal L, Orcesi S, Tonduti D,Accorsi P, Testard H, Abdenur JE, Tay S, Allen GF, Heales S, Kern I, Kato M, Burlina A, Manegold C, Hoffmann GF, Blau N. C*linical and biochemical features of aromatic L-amino acid decarboxylase deficiency.* Neurology. 2010 Jul 6;75(1):64-71. Epub 2010 May 26. Erratum in: Neurology. 2010 Aug 10;75(6):576.

137. Viscomi C, Burlina AB, Dweikat I, Savoiardo M, Lamperti C, Hildebrandt T, Tiranti V, Zeviani M. *Combined treatment with oral metronidazole and N-acetylcysteine is effective in ethylmalonic encephalopathy*. Nat Med. 2010 Aug;16(8):869-71. Epub 2010 Jul 25.

138. Del Rizzo M, Fanin M, Cerutti A, Cazzorla C, Milanesi O, Nascimbeni AC, Angelini C, Giordano L, Bordugo A, Burlina AB. *Long-term follow-up results in enzyme replacement therapy for Pompe disease: a case report.* J Inherit Metab Dis. 2010 Sep 10.

139. Parolin G, Drigo P, Toldo I, Boniver C, Gatta M, Burlina A, Laverda AM, Sartori S. *Pre- and postprandial*

electroencephalography in glucose transporter type 1 deficiency syndrome: an illustrative case to discuss the concept of carbohydrate responsiveness. J Child Neurol. 2011 Jan;26(1):103-8.

140. Porta F, Mussa A, Zanin A, Greggio NA, Burlina A, Spada M. I*mpact of metabolic control on bone quality in phenylketonuria and mild hyperphenylalaninemia*. J Pediatr Gastroenterol Nutr. 2011 Mar;52(3):345-50.

141. Veggiotti P, Burlina A, Coppola G, Cusmai R, De Giorgis V, Guerrini R, Tagliabue A, Dalla Bernardina B. *The ketogenic diet for Dravet syndrome and other epileptic encephalopathies: an Italian consensus.* Epilepsia. 2011 Apr;52 Suppl 2:83-9.

142. Burlina AP, Sims KB, Politei JM, Bennett GJ, Baron R, Sommer C, Møller AT, 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 May 27;11:61.

143. Kölker S, Christensen E, Leonard JV, Greenberg CR, Boneh A, Burlina AB, Burlina AP, Dixon M, Duran M, García Cazorla A, Goodman SI, Koeller DM, Kyllerman M, Mühlhausen C, Müller E, Okun JG, Wilcken B, Hoffmann GF, Burgard P.*Diagnosis and management of glutaric aciduria type I--revised recommendations.* J Inherit Metab Dis. 2011 Jun;34(3):677-94.

144. 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 Jul;44(10-11):916-21.

145. Belmatoug N, Burlina A, Giraldo P, Hendriksz CJ, Kuter DJ, Mengel E, Pastores GM. *Gastrointestinal disturbances and their management in miglustat-treated patients*. J Inherit Metab Dis. 2011 Oct;34(5):991-1001.

146. Bélanger-Quintana A, Burlina A, Harding CO, Muntau AC. *Up to date knowledge on different treatment strategies for phenylketonuria.* Mol Genet Metab. 2011;104 Suppl:S19-25.

147. 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 Apr;3(2):155-64.

148. Giordano C, Viscomi C, Orlandi M, Papoff P, Spalice A, Burlina A, Di Meo I, Tiranti V, Leuzzi V, d'Amati G, Zeviani M. *Morphologic evidence of diffuse vascular damage in human and in the experimental model of ethylmalonic encephalopathy.* J Inherit Metab Dis. 2012 May;35(3):451-8.

149. Doimo M, Trevisson E, Sartori G, Burlina A, Salviati L. *Yeast complementation is sufficiently sensitive to detect the residual activity of ASL alleles associated with mild forms of argininosuccinic aciduria.* J Inherit Metab Dis. 2012 May;35(3):557-8.

150. 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 May;35(3):413-7.

151. Häberle J, Boddaert N, Burlina A, Chakrapani A, Dixon M, Huemer M, Karall D, Martinelli D, Crespo PS, Santer R, Servais A, Valayannopoulos V, Lindner M, Rubio V, Dionisi-Vici C. *Suggested guidelines for the diagnosis and management of urea cycle disorders.*Orphanet J Rare Dis. 2012 May 29;7:32.

152. 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 May;106(1):25-30.

153. Ghezzi D, Baruffini E, Haack TB, Invernizzi F, Melchionda L, Dallabona C, Strom TM, Parini R, Burlina AB, Meitinger T, Prokisch H, Ferrero I, Zeviani M. *Mutations of the mitochondrial-tRNA modifier MTO1 cause hypertrophic cardiomyopathy and lactic acidosis.* Am J Hum Genet. 2012 Jun 8;90(6):1079-87.

154. 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 Jul;106(3):330-44.

155. Di Meo I, Auricchio A, Lamperti C, Burlina A, Viscomi C, Zeviani M. *Effective AAV-mediated gene therapy in a mouse model of ethylmalonic encephalopathy.* EMBO Mol Med. 2012 Sep;4(9):1008-14.

156. Jumbo-Lucioni PP, Garber K, Kiel J, Baric I, Berry GT, Bosch A, Burlina A, Chiesa A, Pico ML, Estrada SC,

Henderson H, Leslie N, Longo N, Morris AA, Ramirez-Farias C, Schweitzer-Krantz S, Silao CL, Vela-Amieva M, Waisbren S, Fridovich-Keil JL. *Diversity of approaches to classic galactosemia around the world: a comparison of diagnosis, intervention, and outcomes.*J Inherit Metab Dis. 2012 Nov;35(6):1037-49.

157. Piva E, Pelloso M, Ciubotaru D, Penello L, Burlina A, Plebani M.T*he role of automated analyzers in detecting abnormal granulation of leucocytes in lysosomal storage diseases: Maroteaux-Lamy disease.*Am J Hematol. 2012 Dec 19.

158. Doimo M, Desbats MA, Baldoin MC, Lenzini E, Basso G, Murphy E, Graziano C, Seri M, Burlina A, Sartori G, Trevisson E, Salviati L. Functional *Analysis of Missense Mutations of OAT, Causing Gyrate Atrophy of Choroid and Retina.Hum Mutat*. 2013 Jan;34(1):229-36.

159. Sechi A, Deroma L, Lapolla A, Paci S, Melis D, Burlina A, Carubbi F, Rigoldi M, Di Rocco M. *Fertility and pregnancy in women affected by glycogen storage disease type I, results of a multicenter Italian study*. J Inherit Metab Dis. 2013 Jan;36(1):83-9.

160. Faletra F, Snider K, Shyng SL, Bruno I, Athanasakis E, Gasparini P, Dionisi-Vici C, Ventura A, Zhou Q, Stanley CA, Burlina A. *Co-inheritance of two ABCC8 mutations causing an unresponsive congenital hyperinsulinism: Clinical and functional characterization of two novel ABCC8 mutations*. Gene. 2013 Mar 1;516(1):122-5.

161. Zanco C, Cazzorla C, Bordugo A, Giordano L, Manara R, Burlina AB. *Metabolic stroke in a late-onset form of isolated sulfite oxidase deficiency. Del Rizzo M, Burlina AP, Sass JO, Beermann F.* Mol Genet Metab.; 108(4):263-6, 2013.

162. Cerone R, Andria G, Giovannini M, Leuzzi V, Riva E, Burlina A.. *Testing for tetrahydrobiopterin responsiveness in patients with hyperphenylalaninemia due to phenylalanine hydroxylase deficiency*. Adv Ther. 30(3): 212-28, 2013.

163. Faletra F, Athanasakis E, Morgan A, Biarnés X, Fornasier F, Parini R, Furlan F, Boiani A, Maiorana A, Dionisi-Vici C, Giordano L, Burlina A, Ventura A, Gasparini P.*Congenital hyperinsulinism: clinical and molecular analysis of a large Italian cohort.* Gene, 25;521(1):160-5, 2013.

164. Keil S, Anjema K, van Spronsen FJ, Lambruschini N, Burlina A, Bélanger-Quintana A, Couce ML, Feillet F, Cerone R, Lotz-Havla AS, Muntau AC, Bosch AM, Meli CA, Billette de Villemeur T, Kern I, Riva E, Giovannini M, Damaj L, Leuzzi V, Blau N. Pediatrics. *Long-term follow-up and outcome of phenylketonuria patients on sapropterin: a retrospective study.* Pediatrics,131(6):e1881-8, 2013.

165. Baruffini E, Dallabona C, Invernizzi F, Yarham JW, Melchionda L, Blakely EL, Lamantea E, Donnini C, Santra S, Vijayaraghavan S, Roper HP, Burlina A, Kopajtich R, Walther A, Strom TM, Haack TB, Prokisch H, Taylor RW, Ferrero I, Zeviani M, Ghezzi D. *MTO1 mutations are associated with hypertrophic cardiomyopathy and lactic acidosis and cause respiratory chain deficiency in humans and yeast.* Hum Mutat, 34(11):1501-9, 2013.

166. Cleary M, Trefz F, Muntau AC, Feillet F, van Spronsen FJ, Burlina A, Bélanger-Quintana A, Giżewska M, Gasteyger C, Bettiol E, Blau N, MacDonald A. *Fluctuations in phenylalanine concentrations in phenylketonuria: a review of possible relationships with outcomes.* Mol Genet Metab; 110(4):418-23, 2013.

167. Fischer S, Huemer M, Baumgartner M, Deodato F, Ballhausen D, Boneh A, Burlina AB, Cerone R, Garcia P, Gökçay G, Grünewald S, Häberle J, Jaeken J, Ketteridge D, Lindner M, Mandel H, Martinelli D, Martins EG, Schwab KO, Gruenert SC, Schwahn BC, Sztriha L, Tomaske M, Trefz F, Vilarinho L, Rosenblatt DS, Fowler B, Dionisi-Vici C. J *Clinical presentation and outcome in a series of 88 patients with the cblC defect.* Inherit Metab Dis.; 37(5):831-40, 2014.

168. Camp KM, Parisi MA, Acosta PB, Berry GT, Bilder DA, Blau N, Bodamer OA, Brosco JP, Brown CS, Burlina AB, Burton BK, Chang CS, Coates PM, Cunningham AC, Dobrowolski SF, Ferguson JH, Franklin TD, Frazier DM, Grange DK, Greene CL, Groft SC, Harding CO, Howell RR, Huntington KL, Hyatt-Knorr HD, Jevaji IP, Levy HL, Lichter-Konecki U, Lindegren ML, Lloyd-Puryear MA, Matalon K, MacDonald A, McPheeters ML, Mitchell JJ, Mofidi S, Moseley KD, Mueller CM, Mulberg AE, Nerurkar LS, Ogata BN, Pariser AR, Prasad S, Pridjian G, Rasmussen SA, Reddy UM, Rohr FJ, Singh RH, Sirrs SM, Stremer SE, Tagle DA, Thompson SM, Urv TK, Utz JR, van Spronsen F, Vockley J, Waisbren SE, Weglicki LS, White DA, Whitley CB, Wilfond BS, Yannicelli S, Young JM. *Phenylketonuria Scientific Review Conference: state of the science and future research needs.* Mol Genet Metab; 112(2): 87-122, 2014.

169. Garbade SF, Greenberg CR, Demirkol M, Gökçay G, Ribes A, Campistol J, Burlina AB, Burgard P, Kölker S. *Unravelling the complex MRI pattern in glutaric aciduria type I using statistical models-a cohort study in 180 patients.*

J Inherit Metab; 37(5): 763-73, 2014.

170. Picca S, Dionisi-Vici C, Bartuli A, De Palo T, Papadia F, Montini G, Materassi M, Donati MA, Verrina E, Schiaffino MC, Pecoraro C, Iaccarino E, Vidal E, Burlina A, Emma F. *Short-term survival of hyperammonemic neonates treated with dialysis.* Pediatr Nephrol.; 4, 2014.

171. Baumgartner MR, Hörster F, Dionisi-Vici C, Haliloglu G, Karall D, Chapman KA, Huemer M, Hochuli M, Assoun M, Ballhausen D, Burlina A, Fowler B, Grünert SC, Grünewald S, Honzik T, Merinero B, Pérez-Cerdá C, Scholl-Bürgi S, Skovby F, Wijburg F, MacDonald A, Martinelli D, Sass JO, Valayannopoulos V, Chakrapani A. *Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia.* Orphanet J Rare Dis. 2;9:130, 2014.

172. Barone R, Carrozzi M, Parini R, Battini R, Martinelli D, Elia M, Spada M, Lilliu F, Ciana G, Burlina A, Leuzzi V, Leoni M, Sturiale L, Matthijs G, Jaeken J, Di Rocco M, Garozzo D, Fiumara A. *A nationwide survey of PMM2-CDG in Italy: high frequency of a mild neurological variant associated with the L32R mutation.* J Neurol.; 262(1):154-64, 2015.

173. 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; 4, 14-1243, 2014.

174. Trefz FK, van Spronsen FJ, MacDonald A, Feillet F, Muntau AC, Belanger-Quintana A, Burlina A, Demirkol M, Giovannini M, Gasteyger C. *Management of adult patients with phenylketonuria: survey results from 24 countries.* Eur J Pediatr.; 174(1):119-27, 2015.

175. Desbats MA, Vetro A, Limongelli I, Lunardi G, Casarin A, Doimo M, Spinazzi M, Angelini C, Cenacchi G, Burlina A, Rodriguez Hernandez MA, Chiandetti L, Clementi M, Trevisson E, Navas P, Zuffardi O, Salviati L. *Primary coenzyme Q10 deficiency presenting as fatal neonatal multiorgan failure.* Eur J Hum Genet.; 1, 110-1038, 2015.

176. 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 uridyltransferase (GALT) gene.* Gene.; 559(2):112-8, 2015.

177. Crow YJ, Chase DS, Lowenstein Schmidt J, Szynkiewicz M, Forte GM, Gornall HL, Oojageer A, Anderson B, Pizzino A, Helman G, Abdel-Hamid MS, Abdel-Salam GM, Ackroyd S, Aeby A, Agosta G, Albin C, Allon-Shalev S, Arellano M, Ariaudo G, Aswani V, Babul-Hirji R, Baildam EM, Bahi-Buisson N, Bailey KM, Barnerias C, Barth M, Battini R, Beresford MW, Bernard G, Bianchi M, Billette de Villemeur T, Blair EM, Bloom M, Burlina AB, Luisa Carpanelli M, Carvalho DR, Castro-Gago M, Cavallini A, Cereda C, Chandler KE, Chitayat DA, Collins AE, Sierra Corcoles C, Cordeiro NJ, Crichiutti G, Dabydeen L, Dale RC, D Arrigo S, De Goede CG, De Laet C, De Waele LM, Denzler I, Desguerre I, Devriendt K, Di Rocco M, Fahey MC, Fazzi E, Ferrie CD, Figueiredo A, Gener B, Goizet C, Gowrinathan NR, Gowrishankar K, Hanrahan D, Isidor B, Kara B, Khan N, King MD, Kirk EP, Kumar R, Lagae L, Landrieu P, Lauffer H, Laugel V, Piana RL, Lim MJ, Lin JP, Linnankivi T, Mackay MT, Marom DR, Marques Lourenço C, McKee SA, Moroni I, Morton JE, Moutard ML, Murray K, Nabbout R, Nampoothiri S, Nunez-Enamorado N, Oades PJ, Olivieri I, Ostergaard JR, Pérez-Dueñas B, Prendiville JS, Ramesh V, Rasmussen M, Régal L, Ricci F, Rio M, Rodriguez D, Roubertie A, Salvatici E, Segers KA, Sinha GP, Soler D, Spiegel R, Stödberg TI, Straussberg R, Swoboda KJ, Suri M, Tacke U, Tan TY, Te Water Naude J, Wee Teik K, Mary Thomas M, Till M, Tonduti D, Maria Valente E, Noel Van Coster R, van der Knaap MS, Vassallo G, Vijzelaar R, Vogt J, Wallace GB, Wassmer E, Webb HJ, Whitehouse WP, Whitney RN, Zaki MS, Zuberi SM, Livingston JH, Rozenberg F, Lebon P, Vanderver A, Orcesi S, Rice GI. *Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1.* Am J Med Genet A.; 2015, 167(2):296-312, 2015.

178. Melis D, Cozzolino M, Minopoli G, Balivo F, Parini R, Rigoldi M, Paci S, Dionisi-Vici C, Burlina A, Andria G, Parenti G. *Progression of Renal Damage in Glycogen Storage Disease Type I Is Associated to Hyperlipidemia: A Multicenter Prospective Italian Study.* J Pediatr.; 1, (29) , 2015.

179. Wortmann SB, van Hasselt PM, Barić I, Burlina A, Darin N, Hörster F, Coker M, Kalkan Ucar S, Krumina Z, Naess K, Ngu LH, Pronicka E, Riordan G, Santer R, Wassmer E, Zschocke J, Schiff M, de Meirleir L, Alowain MA, Smeitink JA, Morava E, Kozicz T, Wevers RA, Wolf NI, Willemsen MA. *Eyes on MEGDEL: Distinctive Basal Ganglia Involvement in Dystonia Deafness Syndrome*. Neuropediatrics; 2, 2015.

Padova 09/02/2017