FORMATO EUROPEO PER IL CURRICULUM VITAE

INFORMAZIONI PERSONALI

Nome ALBERTO BURLINA

E-mail [redacted]

Nazionalità

Data e Luogo di nascita

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 MetabolicheEreditarie*. 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.

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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.

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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

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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

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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

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Padova 09/02/2017