

**FORMATO EUROPEO
PER IL CURRICULUM
VITAE**



INFORMAZIONI PERSONALI

Nome **ALBERTO BURLINA**

E-mail

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/E1 Biochimica 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 ininterrotta 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 più di 700 pazienti con malattie metaboliche ereditarie.

L'attività è 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 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 Metaboliche Ereditarie*. 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 lisosomiali

- 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, Foies 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 Acta 163: 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. *Circadian 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 Pediatr 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 biochemical observation on three cases of fructose 1,6-diphosphatase 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 disorders.* 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, Milanese O, Biban P, Bordugo A, Garavaglia B, Zacchello F, Di Mauro S. *Beneficial effect of sodium dicloracetate 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-acetyl aspartylglutamate 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, Piovon 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, Rapoport 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 *Mitochondrial 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 neutrophils in two patients with glycogen storage disease type Ib treated with recombinant human granulocyte-monocyte-colony-stimulating factor*. Hematologica 81,2:148-151, 1996
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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
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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
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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
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INFORMAZIONI PERSONALI

Nome Maria Teresa Carbone

ESPERIENZA LAVORATIVA

- Nome e indirizzo del datore di lavoro *2003 al 2011 ASL NA1*
- Tipo di azienda o settore *UOC Pediatria Osp. Annunziata*
- Tipo di impiego *Dirigente medico pediatra*
- Principali mansioni e responsabilità *Referente malattie rare ASL NA1*
- Nome e indirizzo del datore di lavoro *Dal 2011 a tutt'oggi AORN Santobono Pausilipon Napoli*
- Tipo di azienda o settore
- Tipo di impiego *Dirigente medico pediatra*
- Principali mansioni e responsabilità *Responsabile UOSD Malattie Metaboliche , Centro di Riferimento regionale Iperfenilalaninemie neonatali (Delibera Regione Campania 25/2019)*

ISTRUZIONE E FORMAZIONE

- Titolo di studio *Laurea in Medicina e Chirurgia , conseguita presso Seconda Università degli studi di Napoli nel 1995 con voto 110 e lode*
- Ulteriori titoli di studio e Formazione *Diploma Trinity (English Course)*
- Diploma di Perfezionamento in Nutrizione infantile*
- Diploma di Perfezionamento in Ematologia pediatrica*
- Master in Neonatologia Avanzata*
- Perfezionamento presso il Centro Regionale di Riferimento per gli screening neonatali e la diagnosi delle malattie metaboliche Istituto G. Gaslini di Genova*
- Perfezionamento presso il Centro Regionale di Riferimento per gli screening neonatali e la diagnosi delle malattie metaboliche Università di Milano, ospedale San Paolo*
- Comando p/o Osp. Bambino Gesù (gennaio 2018) finalizzato all'approfondimento dei PDTA malattie metaboliche ereditarie*

Servizio prestato all'estero
Dipartimento di patologie metaboliche e genetiche dell'Università dello
UTAH, Salt Lake City (USA)

*Specializzazione in Pediatria, conseguita presso Seconda università degli
studi di Napoli nel 2002*

*Specializzazione in Genetica medica conseguita presso Università
Federico II di Napoli nel 2011*

Master II Livello in Management sanitario, conseguita presso Università
Federico II nel marzo 2020

Master II Livello “Formazione manageriale per Direttori Struttura
Complessa e Sanitaria” del Foromez conseguita nel febbraio 2021

Altre esperienze lavorative
ed incarichi ricoperti

Socio attivo della Società Italiana di Pediatria (SIP)

Consigliere Regionale SIPPS (Società Italiana Pediatria Preventiva e
Sociale)

Referente Malattie Rare ASL Na1

Componente di varie commissioni tecniche per l'UNIAMO (Federazione
Nazionale Malattie Rare)

Componente tavoli tecnici regionali per la definizione dei percorsi Malattie
Rare

Componente della prima commissione Malattie Rare Nazionale SIP-
SIMGEPED (Società Italiana malattie genetiche e pediatriche con
disabilità)

Consigliere Nazionale SIMGEPED

Tesoriere Nazionale della Società Scientifica SIMGEPED

Socio Attivo SIMMESN (Società Nazionale Screening neonatale e
metabolismo)

Socio attivo della società internazionale e mondiale delle malattie
metaboliche ereditarie (SSIEM, ICIEM)

Coordinatore gruppo di studio “Microbiota e Malattie Metaboliche
Ereditarie” della Società Scientifica Nazionale SIMMESN

Componente CUG aziendale

Componente Comitato Scientifico Aziendale

Componente segreteria aziendale ANAAO

- Expanded Newborn Screening in Italy Using Tandem Mass Spectrometry: Two Years of National Experience. Ruoppolo M, Malvagia S, Boenzi S, et al. *Int J Neonatal Screen*. 2022 Aug 9;8(3):47. doi: 10.3390/ijns8030047.PMID: 35997437 Free PMC article.
- Possible role of tryptophan metabolism along the microbiota-gut-brain axis on cognitive & behavioral aspects in Phenylketonuria. Parolisi S, Montanari C, Borghi E, et al.; SIMMESN Working Group for Gut Microbiota in Inborn Errors of Metabolism. *Pharmacol Res*. 2023 Nov;197:106952. doi: 10.1016/j.phrs.2023.106952. Epub 2023 Oct5.PMID: 37804926 Free article. Review.
- Italian national consensus statement on management and pharmacological treatment of phenylketonuria. Burlina A, Biasucci G, Carbone MT, et al. *Orphanet J Rare Dis*. 2021 Nov 16;16(1):476. doi: 10.1186/s13023-021-02086-8.PMID: 34784942 Free PMC article.
- Are Phe-Free Protein Substitutes Available in Italy for Infants with PKU All the Same? Verduci E, Tosi M, Montanari C, et al. *Nutrients*. 2023 Dec 21;16(1):30. doi: 10.3390/nu16010030.PMID: 38201860 Free PMC article. Review.
- Dysbiosis, Host Metabolism, and Non-communicable Diseases: Trialogue in the Inborn Errors of Metabolism. Montanari C, Parolisi S, Borghi E, et al. *Front Physiol*. 2021 Sep 6;12:716520. doi: 10.3389/fphys.2021.716520. eCollection 2021.PMID: 34588993 Free PMC article. Review.
- Nutrition, Microbiota and Role of Gut-Brain Axis in Subjects with Phenylketonuria (PKU): A Review. Verduci E, Carbone MT, Borghi E, et al. *Nutrients*. 2020 Oct 29;12(11):3319. doi: 10.3390/nu12113319.PMID: 33138040 Free PMC article. Review.
- The management of phenylketonuria in adult patients in Italy: a survey of six specialist metabolic centers. Burlina A, Leuzzi V, Spada M, Carbone MT, et al. *Curr Med Res Opin*. 2021 Mar;37(3):411-421. doi: 10.1080/03007995.2020.1847717. Epub 2021 Feb 1.PMID: 33222540 Free article
- Quality of life aspects of a low protein diet using GMP in patients with phenylketonuria. Bensi G, Carbone MT, Schiaffino MC, et al. *J Int Med Res*. 2022 Sep;50(9):3000605221125524. doi: 10.1177/03000605221125524.PMID: 36146893 Free PMC article.

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- Five human phenylalanine hydroxylase proteins identified in mild hyperphenylalaninemia patients are disease-causing variants. Daniele A, Cardillo G, Pennino C, Carbone MT, Scognamiglio D, Esposito L, Correr A, Castaldo G, Zagari A, Salvatore F. *Biochim Biophys Acta*. 2008 Jun;1782(6):378-84. doi: 10.1016/j.bbdis.2008.01.012. Epub 2008 Feb 20.PMID: 18346471 Free article.

- HHH syndrome (hyperornithinaemia, hyperammonaemia, homocitrullinuria), with fulminant hepatitis-like presentation. Fecarotta S, Parenti G, Vajro P, Zuppaldi A, Della Casa R, Carbone MT, Correr A, Torre G, Riva S, Dionisi-Vici C, Santorelli FM, Andria G. *J Inher Metab Dis*. 2006 Feb;29(1):186-9. doi: 10.1007/s10545-006-0120-7.PMID: 16601889

- Molecular epidemiology of phenylalanine hydroxylase deficiency in Southern Italy: a 96% detection rate with ten novel mutations. Daniele A, Cardillo G, Pennino C, Carbone MT, Scognamiglio D, Correr A, Pignero A, Castaldo G, Salvatore F. *Ann Hum Genet*. 2007 Mar;71(Pt 2):185-93. doi: 10.1111/j.1469-1809.2006.00328.x. Epub 2006 Nov 10.PMID: 17096675

- Molecular screening of the proopiomelanocortin (POMC) gene in Italian obese children: report of three new mutations. Miraglia del Giudice E, Cirillo G, Santoro N, D'Urso L, Carbone MT, Di Toro R, Perrone L. *Int J Obes Relat Metab Disord*. 2001 Jan;25(1):61-7. doi: 10.1038/sj.ijo.0801485.PMID: 11244459

- Neonatal malpractice claims in Italy: how big is the problem and which are the causes? Fanos V, Tagliabue P, Greco L, Agostiniani R, Carbone MT, D'Agostino P, Correr A. *J Matern Fetal Neonatal Med*. 2012 May;25(5):493-7. doi: 10.3109/14767058.2011.622004.PMID: 21919821

- Transition in inherited metabolic diseases: the dietitians, pediatricians and adult physicians' point of view: the results of an Italian survey. Rossi A, Pancaldi C, Regazzi MG, et al. *Orphanet J Rare Dis.* 2025 May 22;20(1):241. doi: 10.1186/s13023-025-03755-8.PMID: 40405235 Free PMC article.
- Recurrent asthmatic bronchitis in the first years of life: a 3-year follow-up. Brigante E, Cirillo G, Aurelio G, Nocerino G, Carbone MT, de Seta L.*Pediatr Med Chir.* 1998 May-Jun;20(3):205-8.PMID: 9744014 Italian.
- Evaluation of leptin protein levels in patients with Cooley's anaemia. Miraglia Del Giudice E, Perrotta S, Carbone MT, Calabrò C, Esposito L, De Rosa C, Saviano A, Di Toro R, Perrone L.*Br J Haematol.* 1999 Jun;105(3):839-40. doi: 10.1046/j.1365-2141.1999.01484.x.PMID: 10354156 No abstract available.
- Medico-legal liability in neonatal screening. Corbetta C, Carbone MT.*Minerva Pediatr.* 2007 Oct;59(5):454-5.PMID: 17947867 Italian. No abstract available.
- Children with special health care needs attending emergency department in Italy: analysis of 3479 cases. Cianci P, D'Apolito V, Moretti A, et al; Italian Society of Pediatrics (SIP); Italian Society of Pediatric Genetic Diseases and Congenital Disabilities (SIMGePed) the Italian Society of Pediatric Emergency Medicine (SIMEUP); Italian Society For The Study Of Inborn Metabolic Disorders And Newborn Screening (SIMMENS) and Members of Italian Network.*Ital J Pediatr.* 2020 Nov 23;46(1):173. doi: 10.1186/s13052-020-00937-x.PMID: 33228805 Free PMC article.
- Diagnostic issues faced by a rare disease healthcare network during Covid-19 outbreak: data from the Campania Rare Disease Registry. Limongelli G, Iucolano S, Monda E, et al; Campania Rare Disease Network.*J Public Health (Oxf).* 2022 Aug 25;44(3):586-594. doi: 10.1093/pubmed/fdab137.PMID: 33982102 Free.
- Management of phenylketonuria in Europe: survey results from 19 countries. Blau N, Bélanger-Quintana A, Demirkol M, Feillet F, Giovannini M, MacDonald A, Trefz FK, van Spronsen F; European PKU centers.*Mol Genet Metab.* 2010 Feb;99(2):109-15. doi: 10.1016/j.ymgme.2009.09.005. Epub 2009 Sep 13.PMID: 19800826
- Children at social risk and the utilization of health services. Cirillo G, Manetti S, Siani P, Carbone MT. *Minerva Pediatr Med Chir.* 1998 May-Jun;20(3):217-8.PMID: 9744017 Italian.

- Italian national consensus statement on management and pharmacological treatment of phenylketonuria Burlina A, Biasucci G, Carbone MT, Cazzorla C, Paci S, Pochiero F, Spada M, Tummolo A, Zuvadelli J, Leuzzi V. Orphanet J Rare Dis. 2021 Nov 16;16(1):476. doi: 10.1186/s13023-021-02086-8. PMID: 34784942; PMCID: PMC8594187.
- Neonatal malpractice claims in Italy: how big is the problem and which are the causes? Fanos V, Tagliabue P, Greco L, Agostiniani R, Carbone MT, D'Agostino P, Correria A. J Matern Fetal Neonatal Med. 2012 May;25(5):493-7. doi:10.3109/14767058.2011.622004. PMID: 21919821.
- Diagnostic issues faced by a rare disease healthcare network during Covid-19 outbreak: data from the Campania Rare Disease Limongelli G, Iucolano S, Monda E, et. al. Registry. J Public Health (Oxf). 2021 May 13:fdab137.

Collaborazioni Editoriali

- Screening neonatali in Campania e fenilchetonuria: l'esperienza della dell'ospedale SS Annunziata.
- Nuove prospettive terapeutiche nel trattamento della fenilchetonuria,.
- Adolescenza e fenilchetonuria: problematiche in pazienti affetti da patologie metaboliche.

Attività didattica

Numerose attività di docenza sul tema: malattie metaboliche e rare

Diverse consulenze e contratti di collaborazione scientifica sul tema Malattie Metaboliche e Rare

CAPACITÀ E COMPETENZE PERSONALI

Acquisite nel corso della vita e della carriera ma non necessariamente riconosciute da certificati e diplomi ufficiali.

PRIMA LINGUA	Italiano
ALTRE LINGUE	Inglese

• Capacità di lettura	Ottima
• Capacità di scrittura	Ottima
• Capacità di espressione orale	Ottima

CAPACITÀ E COMPETENZE RELAZIONALI	OTTIMA
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CAPACITÀ E COMPETENZE
ARTISTICHE

Amante musica
Sport

PATENTE O PATENTI

B

Tutte le informazioni e dichiarazioni sopra riportate nel curriculum sono rese sotto la propria responsabilità ai sensi degli art. 46 e 47 del decreto del Presidente della Repubblica 28 dicembre 2000, n.445, consapevole delle sanzioni penali per dichiarazioni mendaci, falsità in atti ed uso di atti falsi ai sensi dell'art. 76 del citato decreto del Presidente della Repubblica n.445/2000.

Autorizzo il trattamento dei dati personali contenuti nel mio curriculum vitae in base al D. Lgs. 196/2003 e al Regolamento UE 2016/679

Napoli, 28/05/2025



Curriculum Vitae Europass

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Nationality Italian

Place and date of Birth Naples, 06 aprile

sex 1975 Female

Codice fiscale SCLRSI75D46F839V

Current Position **Pediatrician, full time, at the Department of Maternal and Child Health, Federico II University Hospital, Clinical Genetics Unit.**

Professional experience

November 1, 2010-today Pediatrician, full time, at the Department of Maternal and Child Health, Federico II University Hospital, Clinical Genetics Unit.

June 2007-today Diagnosis, management and follow-up of patients with genetic disorders, with particular experience in Down syndrome
Dietary and pharmacological management of patients with phenylketonuria

1 nov.2010-1 apr.2015 Physician of the Regional Coordination Center for Rare Diseases _ Campania Region

January 2017-April 2025 Coordinator of the activities of the Federico II University Hospital Rare Disease Centre

July 31, 2010-July 31, 2011 Research fellowship at the Federico II University (identification of clinical indicators of efficacy of sapropterin therapy in patients with phenylketonuria)
(Fondazione Serono)

January 22, 2008-march 23, 2008 Research fellowship at the Federico II University (Management of children with Down syndrome)
(Campania Region)

Education

1/4/2008-31/3/2010	Postdoctoral fellowship in "Developmental Medical Sciences", University of Naples Federico II (2 years)
28/1/2008	PhD in "Human Reproduction, Development and Growth" (2008) at the University of Naples Federico II; (3 years)
20/10/2004	Specialization in Paediatrics (5 years) at the University of Naples Federico II (70/70 magna cum laude) Experimental thesis on "Metabolism of sulfur amino acids and folates in Down syndrome"
26/7/1999	Degree in Medicine (1999) at the University of Naples Federico II (110/110 magna cum laude) Experimental thesis on: "Lysinuric protein intolerance: gene identification".
24/3-24/7/1997	Erasmus at the University of Nice, Hopital De L'Archet 2, Nice (FR)

Personal skills

Mother tongue	Italian
Other language	English (C1), Spanish (A2), French (A1)

Additional informations

Research Interests	Genetic disorders (dysmorphology, chromosomal disorders, multifactorial diseases) Hereditary metabolic disorders, in particular Phenylketonuria. From 2002-present, Dr Scala has been involved in the research of the metabolic and molecular mechanisms of Down syndrome and principal investigator or co-investigator in clinical trials for Down syndrome. She has a long standing experience in the clinical follow-up of subjects with Down syndrome. In the field of Phenylketonuria, dr Scala investigated the genotype-phenotype correlation in patients with hyperphenylalaninemia due to phenylalanine hydroxylase deficiency and characterized new gene mutations from a molecular and functional point of view (Daniele et al, 2009). In phenylketonuria, he studied the efficacy and safety of innovative nutritional and pharmacological approaches.
Scientific Societies	Member of the Italian Down syndrome Task Force; Member of the Italian society of Human Genetics; Member of the Italian Society of inborn Errors of Metabolism; Member of the Italian Society of Pediatrics.

Certificates
(Last 5 years)

16.10.2021 Good Clinical Practice Certificate “ICH GOOD CLINICAL PRACTICE E6 (R2)”

Advanced Course in Neurogenetics. University of Naples Federico II –
18.01.2023/20.04.2023

11.12.2023 Good Clinical Practice Certificate “ICH GOOD CLINICAL PRACTICE E6 (R2)”

Certificate on Security and Health in the Workplace. University of Naples Federico II,
25.12.2023

19-20 march 2024 – CERTIFICATE of ADVANCED LIFE SUPPORT (ALS/PALS)
valid until 20/03/2026

Publications

1. Sperandeo MP, Buoninconti A, Passariello A, **Scala I**, Adami A, Lauteala T, Mykkanen J, Andria G, Sebastio G. Feasibility of prenatal diagnosis of lysinuric protein intolerance by linkage analysis: a case report. *Prenat Diagn*. 1999 Aug;19(8):771-3. IF 2,683
2. Romano A, Passariello A, **Scala I**, Sebastio G, Titomanlio L, Del Giudica E. Approccio clinico alle macrocranie non familiari: presentazione di una casistica di 21 pazienti. *Rivista Italiana di Pediatria*, 2000, 26(1), pp. 182-188
3. Pierri NB, Lecora M, Passariello A, **Scala I**, Andria G. New case of bilateral upper limb amelia, facial clefts, and renal hypoplasia. *Am J Med Genet*. 2000 Mar 13;91(2):123-5. IF 2,3
4. **Scala I**, Parenti G, Sebastio G, Andria G. Intolleranza alle proteine con lisinuria: una malattia multisistemica curabile, ma non sempre. *Prospettive in Pediatria* 2001; 31: 133-141.
5. **Scala I**, Brancaccio R, Andria G, Sebastio G. Fattori metabolici di rischio per malformazioni congenite e sindrome di Down: il ruolo dell'omocisteina. In: Mantovani A, Stazi AV, Taruscio D. Interazioni geni e ambiente: folati e malformazioni congenite. *Rapporti ISTISAN 02/26*; pp 52-58
6. **Scala I**, Fiorito V, Andria G, Sebastio G. Hyperhomocysteinemia: should it be a pediatrician's concern? *Ital J Pediatr* 2002; 28: 453-459 IF 1,344
7. Wilcken B, Bamforth F, Li Z, Zhu H, Ritvanen A, Renlund M, Stoll C, Alembik Y, Dott B, Czeizel AE, Gelman-Kohan Z, Scarano G, Bianca S, Ettore G, Tenconi R, Bellato S, **Scala I**, Mutchinick OM, Lopez MA, de Walle H, Hofstra R, Joutchenko L, Kavteladze L, Bermejo E, Martinez-Frias ML, Gallagher M, Erickson JD, Vollset SE, Mastroiacovo P, Andria G, Botto LD. Geographical and ethnic variation of the 677C>T allele of 5,10 methylenetetrahydrofolate reductase (MTHFR): findings from over 7000 newborns from 16 areas world wide. *J Med Genet*. 2003 Aug;40(8):619-25 IF 5,703
8. **Scala I**, Sperandeo MP, Fiorito V, Andria G, Sebastio G. How many loci for the His475Tyr polymorphism of the glutamate carboxypeptidase II gene? *Am J Med Genet A*. 2003 Dec 15;123(3):314-5 IF 2,3
9. **Scala I**, Sellitto M, Salomè S, Granese B, Cestari M, Andria G. Iperomocisteinemia moderata e malformazioni congenite: la prevenzione con l'acido folico. *Occhio Clinico Pediatria*, 2004 n. 8,232-236
10. Martinez-Frias ML, Bermejo E, Rodriguez-Pinilla E, **Scala I**, Andria G, Botto L. Grupo de Trabajo del ECEMC. Frequency of the mutation 677C-T of methylenetetrahydrofolate reductase gene on a sample of 652 Spanish liveborn infants *Med Clin (Barc)*. 2004 Mar 20;122(10):361-4 IF 1,399
11. Andria G, **Scala I**, Sebastio G. Health implications of homocysteine and folates: possible preventive measures. *Nutr. Metab. Cardiovasc. Dis*. 2005; 15: 87-93 IF 3,978

12. Andria G and **Scala I**. Genetic risk factors for neural tube defects: Folic acid supplementation and prevention of birth defects. In Esposito, Gough, Guys and Savanelli: Neurogenic Bladder Dysfunctions in Pediatric patients. Diagnosis, treatment and follow-up. Springer Eds, 2014, pp 42-50.
13. **Scala I**, Granese B, Sellitto M, Salomè S, Sammartino A, Pepe A, Mastroiacovo P, Sebastio G, Andria G. Analysis of seven maternal polymorphisms of genes involved in homocysteine/folate metabolism and risk of Down syndrome offspring. **Genetics in Medicine**. 2006; 8:409-416 IF 5,56
14. **Scala I**, Granese B, Lisi A, Mastroiacovo P, Andria G. Response to Folate Gene Polymorphisms and the Risk of Down Syndrome Pregnancies in Young Italian Women by Coppedè et al., 2006: Am J Med Genet 140A:1083-1091. **Am J Med Genet A**. 2007 12:1015-1017 IF 2,3
15. **Scala I**, Sebastio G. Nuove conoscenze sulla genetica molecolare delle cromosomopatie. **Prospettive in Pediatria**, 2007;37:59-68
16. Galletti P, De Bonis ML, Sorrentino A, Raimo M, D'Angelo S, **Scala I**, Andria G, D'Aniello A, Ingrosso D, Zappia V. Accumulation of altered aspartyl residues in erythrocyte proteins from patients with Down's syndrome. **FEBS J**. 2007; 274:5263-77 IF 4,25
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biomarker for cancer: focus on CD43. *Biomed Res Int.* 2014;2014:742831. doi: 10.1155/2014/742831. Epub 2014 Feb 13.

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Can early physical therapy positively affect the onset of independent walking in infants with Down syndrome? A retrospective cohort study. *Minerva Pediatr.* 2018 Feb 19. doi: 10.23736/S0026-4946.18.05041-7. Online ahead of print.

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Diagnostic issues faced by a rare disease healthcare network during Covid-19 outbreak: data from the Campania Rare Disease Registry. *J Public Health (Oxf)*. 2021 May 13;fdab137. doi: 10.1093/pubmed/fdab137. Online ahead of print.

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43. **Scala I**, Concolino D, Nastasi A, Esposito G, Crisci D, Sestito S, Ferraro S, Albano L, Ruoppolo M, Parenti G, Strisciuglio P. Beneficial Effects of Slow-Release Large Neutral Amino Acids after a Phenylalanine Oral Load in Patients with Phenylketonuria. *Nutrients*. 2021 Nov 10;13(11):4012. doi: 10.3390/nu13114012.

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Naples, June 5, 2026