

CURRICULUM VITAE

PERSONAL INFORMATION

Name	Maria Alice Donati
Address	Professional address: Children's Hospital Meyer viale Pieraccini 24, 50139 Firenze, Italy
Telephone	+39 (0) 55 5662409
Fax	+39 (0) 55 5662849
E-mail	m.donati@meyer.it
Nationality	Italian
Date of birth	24 th August 1954

EXPERTISE

- | Principal areas of expertise | Therapeutic Area/Indication | Notes |
|------------------------------|---|--|
| | 1. Neonatal Screening | Expanded Newborn screening in Tuscany and Umbria |
| | 2. Neonatal Screening for lysosomal storage disease | Pilot Project: Newborn screening for Pompe disease, Fabry disease and MPS I |
| | 3. Pompe disease | Therapeutic trial with MYOZYME (ERT with acid maltase)
And Miglustat-chaperonic therapy
Multicenter project for Italian Guidelines |
| | 4. GM1 gangliosidosis | Miglustat in GM1 gangliosidosis chaperonic therapy |
| | 5. Mitochondrial Diseases | Collaborative study with Columbia University New York in clinical trial in MNGIE
Collaborative study with Columbia University New York in clinical trial in Timidine Kinase defect |
| | 6. Hunter disease | Hunter Registry-enzyme replacement therapy |

WORK EXPERIENCE

- Dates (from – to) 2012-
- Name and address of employer Children's Hospital Meyer viale Pieraccini 24, 50139 Firenze, Italy
 - Type of business or sector Department of Neuroscience
 - Occupation or position held Head of the Metabolic and Muscular Unit (including Clinical regional Centre Newborn Screening)
 - Main activities and responsibilities Coordinator of clinical, and research activities of *Metabolic and Muscular Unit*
- Dates (from – to) 2006-2010
- Name and address of employer Children's Hospital Meyer viale Pieraccini 24, 50139 Firenze, Italy
 - Type of business or sector Paediatric Neurology Unit
 - Occupation or position held Head of the Metabolic and Muscular Unit (including Clinical regional Centre Newborn Screening, Laboratory in Biochemical and molecular area in Metabolic and Muscular disease)
 - Main activities and responsibilities Coordinator of clinical, laboratory and research activities of *Metabolic and Muscular Unit*
- Dates (from – to) 2006-
- Name and address of employer University of Florence, Piazza San Marco 4, Florence (Italy)
 - Type of business or sector Medical School
 - Occupation or position held Professor (contract without charge) at the graduate school of Child Neurology and Psychiatry (University of Florence)
 - Main activities and responsibilities Clinical, research, teaching
- Dates (from – to) 1999-2006
- Name and address of employer Children's Hospital Meyer Via Luca Giordano 13, 50123 Firenze, Italy
 - Type of business or sector Paediatric Department
 - Occupation or position held Head of Metabolic and Muscular Unit
 - Main activities and responsibilities Coordinator of Metabolic Laboratory, Regional Screening Centre for hyperthyroidism, metabolic diseases and clinical activities about inborn errors of Metabolism and Muscular diseases
- Dates (from – to) 1996-2006
- Name and address of employer Children's Hospital Meyer Via Luca Giordano 13, 50123 Firenze, Italy
 - Type of business or sector Paediatric Department I
 - Occupation or position held Assistant
 - Main activities and responsibilities *Clinical activity in General Pediatrics and Pediatric Specialties as hereditary neuromuscular metabolic diseases, pediatric neurology and neonatology*
- Dates (from – to) 1988-1996
- Name and address of employer Children's Hospital Meyer Via Luca Giordano 13, 50123 Firenze, Italy
 - Type of business or sector Paediatric Department III
 - Occupation or position held Assistant

- Main activities and responsibilities
Activities in metabolic laboratory: set up of numerous assays for the diagnosis of metabolic diseases such as Lysosomal Storage Disorder like enzyme and metabolite assays
- Dates (from – to)
1988-
- Name and address of employer
University of Florence, Piazza San Marco 4, Florence (Italy)
- Type of business or sector
Medical School
- Occupation or position held
Professor (contract without charge) at the graduate school of Paediatric (University of Florence)
- Main activities and responsibilities
Clinical, research, teaching
- Dates (from – to)
1979-1988
- Name and address of employer
University of Florence, *Piazza San Marco 4, Florence (Italy)*
- Type of business or sector
Laboratory and diagnostic activity in the hereditary metabolic encephalopathy and neuromuscular diseases
- Occupation or position held
Volunteer assistant in Pediatrics Department
- Main activities and responsibilities
Clinical and laboratory activities for the diagnosis of Lysosomal Storage and mitochondrial Disorder: set up of numerous assays, cultured cells, skin-muscular biopsy

EDUCATION AND TRAINING

- Dates (from – to)
October 1979
- Name and type of organization providing education and training
Medical School, University of Florence, Florence, Italy, October 15, 1979
- Principal subjects/occupational skills covered
- Title of qualification awarded
MD
- Level in national classification (if appropriate)
- Dates (from – to)
November 1979
- Name and type of organization providing education and training
Ministry of Health
- Principal subjects/occupational skills covered
- Title of qualification awarded
Licence for Medical Practice
- Level in national classification (if appropriate)
- Dates (from – to)
July 1982
- Name and type of organization providing education and training
University of Florence, Florence, July 19, 1982
- Principal subjects/occupational skills covered
Postgraduate training-clinical duties
- Title of qualification awarded
Specialist of Pediatric
- Level in national classification (if appropriate)
- Dates (from – to)
1987-1988

<ul style="list-style-type: none"> • Name and type of organization providing education and training • Principal subjects/occupational skills covered • Title of qualification awarded • Level in national classification (if appropriate) 	<p>University of Florence, Florence</p> <p><i>Postgraduate training-clinical duties in Pediatric Sciences</i></p> <p>Pediatric Sciences</p>
<ul style="list-style-type: none"> • Dates (from – to) • Name and type of organization providing education and training • Principal subjects/occupational skills covered • Title of qualification awarded • Level in national classification (if appropriate) 	<p>1988-1989</p> <p>University of Siena, Siena</p> <p><i>Postgraduate training-clinical duties in Neuropediatrics</i></p> <p>Neuropediatrics</p>
<ul style="list-style-type: none"> • Dates (from – to) • Name and type of organization providing education and training • Principal subjects/occupational skills covered 	<p>19 November 2009</p> <p>PHARMANET-SERVICES GMBH</p> <p>ICH-GCP</p>
MOTHER TONGUE	Italian
OTHER LANGUAGES	
<ul style="list-style-type: none"> • Reading skills • Writing skills • Verbal skills 	<p>English</p> <p>Good</p> <p>basic</p> <p>basic</p>
ADDITIONAL INFORMATION	<p>Appointment to External Committees</p> <ul style="list-style-type: none"> - Member elected, Directory, The Italian Society of Neuropediatric (1996-1999) - Member elected, Directory, The Italian Society of Inborn Error of Metabolism (SISMME) (1999-2001) - Member of The Italian Society of Paediatrics (SIP) - Member of The Italian Society of Neuropaediatrics (SINP) - Member of The Italian Society of Inborn Error of Metabolism (SISMME) - Member of the Society for the Study of Inborn Errors of Metabolism (SSIEM)

1. Pichiecchio A, Rossi M, Cinnante C, Colafati SG, De Icco R, Parini R, Menni F, Furlan F, Burlina A, Sacchini M, **Donati MA**, Fecarotta S, Casa RD, Deodato F, Taurisano R, Di Rocco M. **Muscle MRI of classic infantile Pompe patients: Fatty substitution and edema-like changes.** Muscle Nerve. 2016 Sep 26. IF: 2.713
2. Harel T, Yoon WH, Garone C, Gu S, Coban-Akdemir Z, Eldomery MK, Posey JE, Jhangiani SN, Rosenfeld JA, Cho MT, Fox S, Withers M, Brooks SM, Chiang T, Duraine L, Erdin S, Yuan B, Shao Y, Moussallem E, Lamperti C, **Donati MA**, Smith JD, McLaughlin HM, Eng CM, Walkiewicz M, Xia F, Pippucci T, Magini P, Seri M, Zeviani M, Hirano M, Hunter JV, Srour M, Zagnoni S, Lewis RA, Muzny DM, Lotze TE, Boerwinkle E; Baylor-Hopkins Center for Mendelian Genomics.; University of Washington Center for Mendelian Genomics., Gibbs RA, Hickey SE, Graham BH, Yang Y, Buhas D, Martin DM, Potocki L, Graziano C, Bellen HJ, Lupski JR. **Recurrent De Novo and Biallelic Variation of ATAD3A, Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes.** Am J Hum Genet. 2016 Oct 6;99(4):831-845. IF: 10.794
3. Tonin R, Caciotti A, Funghini S, Pasquini E, Mooney SD, Cai B, Proncopio E, **Donati MA**, Baronio F, Bettocchi I, Cassio A, Biasucci G, Bordugo A, la Marca G, Guerrini R, Morrone A. **Clinical relevance of short-chain acyl-CoA dehydrogenase (SCAD) deficiency: Exploring the role of new variants including the first SCAD-disease-causing allele carrying a synonymous mutation.** BBA Clin. 2016 Mar 10;5:114-9.
4. Valayannopoulos V, Baruteau J, Delgado MB, Cano A, Couce ML, Del Toro M, **Donati MA**, Garcia-Cazorla A, Gil-Ortega D, Gomez-de Quero P, Guffon N, Hofstede FC, Kalkan-Ucar S, Coker M, Lama-More R, Martinez-Pardo Casanova M, Molina A, Pichard S, Papadia F, Rosello P, Plisson C, Le Mouhaer J, Chakrapani A. **Carglumic acid enhances rapid ammonia detoxification in classical organic acidurias with a favourable risk-benefit profile: a retrospective observational study.** Orphanet J Rare Dis. 2016 Mar 31;11:32. IF: 3.290
5. Mazzone ES, Coratti G, Sormani MP, Messina S, Pane M, D'Amico A, Colia G, Fanelli L, Berardinelli A, Gardani A, Lanzillotta V, D'Ambrosio P, Petillo R, Cavallaro F, Frosini S, Bello L, Bonfiglio S, De Sanctis R, Rolle E, Forcina N, Magri F, Vita G, Palermo C, **Donati MA**, Procopio E, Arnoldi MT, Baranello G, Mongini T, Pini A, Battini R, Pegoraro E, Torrente Y, Previtali SC, Bruno C, Politano L, Comi GP, D'Angelo MG, Bertini E, Mercuri E. **Timed Rise from Floor as a Predictor of Disease Progression in Duchenne Muscular Dystrophy: An Observational Study.** PLoS One. 2016 Mar 16;11(3):e0151445. IF: 3.057
6. Astrea G, Perazza S, Frosini S, Moretti E, Sacchini M, Dati E, Pecini C, Procopio E, Santorelli FM, **Donati MA**, Battini R. **Infantile-Onset Pompe Disease: The Care Beyond the Cure.** J Neuromuscul Dis. 2015;2(s1):S58-S59.
7. Sacchini M, Procopio E, Pasquini E, Pochiero F, Ombrone D, LaMarca G, Catarzi S, Morrone A, **Donati MA**. **Alpha Glucosidase Assay on Dried Blood Spot in the Early Diagnosis of Infantile Pompe Disease.** J Neuromuscul Dis. 2015;2(s1):S53.
8. Panza E, Escamilla-Honrubia JM, Marco-Marín C, Gougéard N, De Michele G, Brescia Morra V, Liguori R, Salvati L, **Donati MA**, Cusano R, Pippucci T, Ravazzolo R, Németh AH, Smithson S, Davies S, Hurst JA, Bordo D, Rubio V, Seri M. **ALDH18A1 gene mutations cause dominant spastic paraplegia SPG9: loss of function effect and plausibility of a dominant negative mechanism.** Brain. 2016 Jan;139(Pt 1):e3. IF: 10.103
9. Pane M, Mazzone ES, Sivo S, Sormani MP, Messina S, D'Amico A, Carlesi A, Vita G, Fanelli L, Berardinelli A, Torrente Y, Lanzillotta V, Viggiano E, D'Ambrosio P, Cavallaro F, Frosini S, Barp A, Bonfiglio S, Scalise R, De Sanctis R, Rolle E, Graziano A, Magri F, Palermo C, Rossi F, **Donati MA**, Sacchini M, Arnoldi MT, Baranello G, Mongini T, Pini A, Battini R, Pegoraro E, Previtali S, Bruno C, Politano L, Comi GP, Bertini E, Mercuri E. **Correction: Long Term Natural History Data in Ambulant Boys with Duchenne Muscular Dystrophy: 36-Month Changes.** PLoS One. 2015 Dec 4;10(12):e0144079. IF: 3.057
10. Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, **Donati MA**, Federico A, Minetti C, Moggio M, Mongini T, Santorelli FM, Servadei S, Tonin P, Toscano A, Bruno C, Bello L, Ienco EC, Cardaioli E, Catteruccia M, Da Pozzo P, Filosto M, Lamperti C, Moroni I, Musumeci O, Pegoraro E, Ronchi D, Sauchelli D, Scarpelli M, Sclacchi M, Valentino ML, Vercelli L, Zeviani M, Siciliano G. **Erratum to: Redefining phenotypes associated with mitochondrial DNA single deletion.** J Neurol. 2015 Dec;262(12):2800. IF: 3.408
11. Romani I, Borsini W, Nencini P, Morrone A, Ferri L, Frusconi S, Donadio VA, Liguori R, **Donati MA**, Falconi S, Pracucci G, Inzitari D. **De novo Diagnosis of Fabry Disease among Italian Adults with Acute Ischemic**

Stroke or Transient Ischemic Attack. J Stroke Cerebrovasc Dis. 2015 Nov;24(11):2588-95. IF: 1.599

12. Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, **Donati MA**, Federico A, Minetti C, Moggio M, Mongini T, Santorelli FM, Servidei S, Tonin P, Toscano A, Bruno C, Bello L, Caldarazzo Ienco E, Cardaioli E, Catteruccia M, Da Pozzo P, Filosto M, Lamperti C, Moroni I, Musumeci O, Pegoraro E, Ronchi D, Sauchelli D, Scarpelli M, Sciacco M, Valentino ML, Vercelli L, Zeviani M, Siciliano G. **Redefining phenotypes associated with mitochondrial DNA single deletion.** J Neurol. 2015 May;262(5):1301-9. IF: 3.408
13. Ferri L, **Donati MA**, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, Morrone A. **Intra-individual plasticity of the TAZ gene leading to different heritable mutations in siblings with Barth syndrome.** Eur J Hum Genet. 2015 Dec;23(12):1708-12. IF: 4.580
14. Nesti C, Meschini MC, Meunier B, Sacchini M, Doccini S, Romano A, Petrillo S, Pezzini I, Seddiki N, Rubegni A, Piemonte F, **Donati MA**, Brasseur G, Santorelli FM. **Additive effect of nuclear and mitochondrial mutations in a patient with mitochondrial encephalomyopathy.** Hum Mol Genet. 2015 Jun 1;24(11):3248-56. IF: 5.985
15. Ferla R, Claudiani P, Savarese M, Kozarsky K, Parini R, Scarpa M, **Donati MA**, Sorge G, Hopwood JJ, Parenti G, Fecarotta S, Nigro V, Sivri HS, Van Der Ploeg A, Andria G, Brunetti-Pierri N, Auricchio A. **Prevalence of anti-adenovirus serotype 8 neutralizing antibodies and arylsulfatase B cross-reactive immunologic material in mucopolysaccharidosis VI patient candidates for a gene therapy trial.** Hum Gene Ther. 2015 Mar;26(3):145-52 IF: 4.062
16. Caciotti A, Tonin R, Rigoldi M, Ferri L, Catarzi S, Cavicchi C, Procopio E, **Donati MA**, Ficcadenti A, Fiumara A, Barone R, Garavelli L, Rocco MD, Filocamo M, Antuzzi D, Scarpa M, Mooney SD, Li B, Skouma A, Bianca S, Concolino D, Casalone R, Monti E, Pantaleo M, Giglio S, Guerrini R, Parini R, Morrone A. **Optimizing the molecular diagnosis of GALNS: novel methods to define and characterize Morquio-A syndrome-associated mutations.** Hum Mutat. 2015 Mar;36(3):357-68. IF: 5.089
17. Pane M, Mazzone ES, Sivo S, Sormani MP, Messina S, D'Amico A, Carlesi A, Vita G, Fanelli L, Berardinelli A, Torrente Y, Lanzillotta V, Viggiano E, D'Ambrosio P, Cavallaro F, Frosini S, Barp A, Bonfiglio S, Scalise R, De Sanctis R, Rolle E, Graziano A, Magri F, Palermo C, Rossi F, **Donati MA**, Sacchini M, Arnoldi MT, Baranello G, Mongini T, Pini A, Battini R, Pegoraro E, Previtali S, Bruno C, Politano L, Comi GP, Bertini E, Mercuri E. **Long term natural history data in ambulant boys with Duchenne muscular dystrophy: 36-month changes.** PLoS One. 2014;9(10):e108205. IF: 3.234
18. 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. 2015;30(5):839-47. IF: 1.866
19. Parenti G, Fecarotta S, la Marca G, Rossi B, Ascione S, **Donati MA**, Morandi LO, Ravaglia S, Pichiecchio A, Ombrone D, Sacchini M, Pasanisi MB, De Filippi P, Danesino C, Della Casa R, Romano A, Mollica C, Rosa M, Agovino T, Nusco E, Porto C, Andria G. **A chaperone enhances blood α -glucosidase activity in Pompe disease patients treated with enzyme replacement therapy.** Mol Ther. 2014;22(11):2004-12. IF: 6.227
20. Diodato D, Invernizzi F, Lamantea E, Fagioli G, Parini R, Menni F, Parenti G, Bollani L, Pasquini E, **Donati MA**, Cassandrini D, Santorelli FM, Haack TB, Prokisch H, Ghezzi D, Lamperti C, Zeviani M. **Common and Novel TMEM70 Mutations in a Cohort of Italian Patients with Mitochondrial Encephalocardiomyopathy.** JIMD. 2015;15:71-8. IF: 3.365
21. Catteruccia M, Verrigni D, Martinelli D, Torracco A, Agovino T, Bonafé L, D'Amico A, **Donati MA**, Adorisio R, Santorelli FM, Carrozzo R, Bertini E, Dionisi-Vici C. **Persistent pulmonary arterial hypertension in the newborn (PPHN): a frequent manifestation of TMEM70 defective patients.** Mol Genet Metab. 2014;111(3):353-9. IF: 2.625
22. Cavicchi C, **Donati M**, Parini R, Rigoldi M, Bernardi M, Orfei F, Gentiloni Silveri N, Colasante A, Funghini S, Catarzi S, Pasquini E, la Marca G, Mooney S, Guerrini R, Morrone A. **Sudden unexpected fatal encephalopathy in adults with OTC gene mutations-Clues for early diagnosis and timely treatment.** Orphanet J Rare Dis. 2014;9:105. IF: 3.358
23. Pane M, Mazzone ES, Sormani MP, Messina S, Vita GL, Fanelli L, Berardinelli A, Torrente Y, D'Amico A,

- Lanzillotta V, Viggiano E, D'Ambrosio P, Cavallaro F, Frosini S, Bello L, Bonfiglio S, Scalise R, De Sanctis R, Rolle E, Bianco F, Van der Haawue M, Magri F, Palermo C, Rossi F, **Donati MA**, Alfonsi C, Sacchini M, Arnoldi MT, Baranello G, Mongini T, Pini A, Battini R, Pegoraro E, Previtali SC, Napolitano S, Bruno C, Politano L, Comi GP, Bertini E, Morandi L, Gualandi F, Ferlini A, Goemans N, Mercuri E. **6 Minute walk test in Duchenne MD patients with different mutations: 12 month changes.** PLoS One. 2014;9(1):e83400. IF: 3.234
24. Mancuso M, Orsucci D, Angelini C, Bertini E, Carelli V, Comi GP, **Donati A**, Minetti C, Moggio M, Mongini T, Servidei S, Tonin P, Toscano A, Uziel G, Bruno C, Ienco EC, Filosto M, Lamperti C, Catteruccia M, Moroni I, Musumeci O, Pegoraro E, Ronchi D, Santorelli FM, Sauchelli D, Scarpelli M, Sciacco M, Valentino ML, Vercelli L, Zeviani M, Siciliano G. **The m.3243A>G mitochondrial DNA mutation and related phenotypes. A matter of gender?** J Neurol. 2014;261(3):504-10. IF: 3.377
 25. Catarzi S, Caciotti A, Thusberg J, Tonin R, Malvagia S, la Marca G, Pasquini E, Cavicchi C, Ferri L, **Donati MA**, Baronio F, Guerrini R, Mooney SD, Morrone A. **Medium-chain acyl-CoA deficiency: outlines from newborn screening, in silico predictions, and molecular studies.** Scientific World Journal. 2013 31;2013:625824. IF: 2.43
 26. Hildick-Smith GJ, Cooney JD, Garone C, Kremer LS, Haack TB, Thon JN, Miyata N, Lieber DS, Calvo SE, Akman HO, Yien YY, Huston NC, Branco DS, Shah DI, Freedman ML, Koehler CM, Italiano JE Jr, Merckenschlager A, Beblo S, Strom TM, Meitinger T, Freisinger P, **Donati MA**, Prokisch H, Mootha VK, DiMauro S, Paw BH. **Macrocytic anemia and mitochondriopathy resulting from a defect in sideroflexin 4.** Am J Hum Genet. 2013 7;93(5):906-14. IF: 10.987
 27. Ferri L, Funghini S, Fioravanti A, Biondi EG, la Marca G, Guerrini R, **Donati MA**, Morrone A. **Aminoacylase I deficiency due to ACY1 mRNA exon skipping.** Clin Genet. 2014;86(4):367-72. IF: 3.652
 28. Caciotti A, Catarzi S, Tonin R, Lugli L, Perez CR, Michelakakis H, Mavridou I, **Donati MA**, Guerrini R, d'Azzo A, Morrone A. **Galactosialidosis: review and analysis of CTSA gene mutations.** Orphanet J Rare Dis. 2013 2;8:114. IF: 3.958
 29. Garone C, **Donati MA**, Sacchini M, Garcia-Diaz B, Bruno C, Calvo S, Mootha VK, Dimauro S **Mitochondrial encephalomyopathy due to a novel mutation in ACAD9.** JAMA Neurol. 2013 1;70(9):1177-9.
 30. Ferri L, Caciotti A, Cavicchi C, Rigoldi M, Parini R, Caserta M, Chibbaro G, Gasperini S, Procopio E, **Donati MA**, Guerrini R, Morrone A **Integration of PCR-Sequencing Analysis with Multiplex Ligation-Dependent Probe Amplification for Diagnosis of Hereditary Fructose Intolerance.** JIMD. 2012;6:31-7. IF: 4.070
 31. Ferri L, **Donati MA**, Funghini S, Malvagia S, Catarzi S, Lugli L, Ragni L, Bertini E, Vaz FM, Cooper DN, Guerrini R, Morrone A. **New clinical and molecular insights on Barth syndrome.** Orphanet J Rare Dis. 2013 14;8:27. IF: 3.958
 32. Travaglini L, Brancati F, Silhavy J, Iannicelli M, Nickerson E, Elkhartoufi N, Scott E, Spencer E, Gabriel S, Thomas S, Ben-Zeev B, Bertini E, Boltshauser E, Chaouch M, Cilio MR, de Jong MM, Kayserili H, Ogur G, Poretti A, Signorini S, Uziel G, Zaki MS; International JSRD Study Group, Johnson C, Attié-Bitach T, Gleeson JG, Valente EM. **Phenotypic spectrum and prevalence of INPP5E mutations in Joubert syndrome and related disorders.** Eur J Hum Genet. 2013;21(10):1074-8. IF: 4.225
 33. Mazzone ES, Pane M, Sormani MP, Scalise R, Berardinelli A, Messina S, Torrente Y, D'Amico A, Doglio L, Viggiano E, D'Ambrosio P, Cavallaro F, Frosini S, Bello L, Bonfiglio S, De Sanctis R, Rolle E, Bianco F, Magri F, Rossi F, Vasco G, Vita G, Motta MC, **Donati MA**, Sacchini M, Mongini T, Pini A, Battini R, Pegoraro E, Previtali S, Napolitano S, Bruno C, Politano L, Comi GP, Bertini E, Mercuri E. **24 month longitudinal data in ambulant boys with Duchenne muscular dystrophy.** PLoS One. 2013;8(1):e52512. IF: 3.534
 34. Filosto M, Scarpelli M, Tonin P, Lucchini G, Pavan F, Santus F, Parini R, **Donati MA**, Cotelli MS, Vielmi V, Todeschini A, Canonico F, Tomelleri G, Padovani A, Rovelli A. **Course and management of allogeneic stem cell transplantation in patients with mitochondrial neurogastrointestinal encephalomyopathy.** J Neurol. 2012 Dec;259(12):2699-706. IF: 3.578
 35. Manara R, Priante E, Grimaldi M, Santoro L, Polonara G, Parini R, Scarpa M; Italian MPS Neuroimaging Study Group. **Closed Meningo(encephalo)cele: a new feature in Hunter syndrome.** AJNR Am J Neuroradiol.

2012 May;33(5):873-7. IF: 3.167

36. Catarzi S, Giunti L, Papadia F, Gabrielli O, Guerrini R, **Donati MA**, Genuardi M, Morrone A. **Morquio A syndrome due to maternal uniparental isodisomy of the telomeric end of chromosome 16.** Mol Genet Metab. 2012 Mar;105(3):438-42. IF: 2.834
37. Funghini S, Thusberg J, Spada M, Gasperini S, Parini R, Ventura L, Meli C, De Cosmo L, Sibilio M, Mooney SD, Guerrini R, **Donati MA**, Morrone A. **Carbamoyl phosphate synthetase 1 deficiency in Italy: clinical and genetic findings in a heterogeneous cohort.** Gene. 2012 Feb 10;493(2):228-34. IF: 2.196
38. Angelini C, Semplicini C, Ravaglia S, Bembi B, Servidei S, Pegoraro E, Moggio M, Filosto M, Sette E, Crescimanno G, Tonin P, Parini R, Morandi L, Marrosu G, Greco G, Musumeci O, Di Iorio G, Siciliano G, **Donati MA**, Carubbi F, Ermani M, Mongini T, Toscano A; Italian GSDII Group. **Observational clinical study in juvenile-adult glycogenosis type 2 patients undergoing enzyme replacement therapy for up to 4 years.** J Neurol. 2012 May;259(5):952-8. IF: 3.578
39. Fanin M, Anichini A, Cassandrini D, Fiorillo C, Scapolan S, Minetti C, Cassanello M, **Donati MA**, Siciliano G, D'Amico A, Lilliu F, Bruno C, Angelini C. **Allelic and phenotypic heterogeneity in 49 Italian patients with the muscle form of CPT-II deficiency.** Clin Genet. 2012 Sep;82(3):232-9. IF: 4.247
40. Mazzone E, Vasco G, Sormani MP, Torrente Y, Berardinelli A, Messina S, D'Amico A, Doglio L, Politano L, Cavallaro F, Frosini S, Bello L, Bonfiglio S, Zucchini E, De Sanctis R, Scutifero M, Bianco F, Rossi F, Motta MC, Sacco A, **Donati MA**, Mongini T, Pini A, Battini R, Pegoraro E, Pane M, Gasperini S, Previtali S, Napolitano S, Martinelli D, Bruno C, Vita G, Comi G, Bertini E, Mercuri E. **Functional changes in Duchenne muscular dystrophy: a 12-month longitudinal cohort study.** Neurology. 2011 Jul 19;77(3):250-6. IF: 8.312
41. Micheletti MV, Lavoratti G, Gasperini S, **Donati MA**, Pela I. **Hemolytic uremic syndrome and rhabdomyolysis in a patient with succinate coenzyme Q reductase (complex II) deficiency.** Clin Nephrol. 2011 Jul;76(1):68-73. IF: 1.171
42. Ferri L, Guido C, la Marca G, Malvagia S, Cavicchi C, Fiumara A, Barone R, Parini R, Antuzzi D, Feliciani C, Zampetti A, Manna R, Giglio S, Della Valle CM, Wu X, Valenzano KJ, Benjamin R, **Donati MA**, Guerrini R, Genuardi M, Morrone A. **Fabry disease: polymorphic haplotypes and a novel missense mutation in the GLA gene.** Clin Genet. 2012 Mar;81(3):224-33. IF: 4.247
43. Caciotti A, Garman SC, Rivera-Colón Y, Procopio E, Catarzi S, Ferri L, Guido C, Martelli P, Parini R, Antuzzi D, Battini R, Sibilio M, Simonati A, Fontana E, Salviati A, Akinci G, Cereda C, Dionisi-Vici C, Deodato F, d'Amico A, d'Azzo A, Bertini E, Filocamo M, Scarpa M, di Rocco M, Tift CJ, Ciani F, Gasperini S, Pasquini E, Guerrini R, **Donati MA**, Morrone A. **GM1 gangliosidosis and Morquio B disease: an update on genetic alterations and clinical findings.** Biochim Biophys Acta. 2011 Jul;1812(7):782-90.
44. Manara R, Priante E, Grimaldi M, Santoro L, Astarita L, Barone R, Concolino D, Di Rocco M, **Donati MA**, Fecarotta S, Ficcadenti A, Fiumara A, Furlan F, Giovannini I, Lilliu F, Mardari R, Polonara G, Procopio E, Rampazzo A, Rossi A, Sanna G, Parini R, Scarpa M. **Brain and spine MRI features of Hunter disease: frequency, natural evolution and response to therapy.** J Inher Metab Dis. 2011 Jun;34(3):763-80. IF: 4.070
45. Mazzone E, Martinelli D, Berardinelli A, Messina S, D'Amico A, Vasco G, Main M, Doglio L, Politano L, Cavallaro F, Frosini S, Bello L, Carlesi A, Bonetti AM, Zucchini E, De Sanctis R, Scutifero M, Bianco F, Rossi F, Motta MC, Sacco A, **Donati MA**, Mongini T, Pini A, Battini R, Pegoraro E, Pane M, Pasquini E, Bruno C, Vita G, de Waure C, Bertini E, Mercuri E. **North Star Ambulatory Assessment, 6-minute walk test and timed items in ambulant boys with Duchenne muscular dystrophy.** Neuromuscul Disord. 2010 Nov;20(11):712-6. IF: 2.764
46. Gasperini S, Stagi S, Gasperini U, Guerrini R, la Marca G, **Donati MA**. **Orange-colored diapers as first sign of Lesch-Nyhan disease in an asymptomatic infant.** Pediatr Nephrol. 2010 Nov;25(11):2373-4. IF: 2.183
47. 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. IF: 9.232

48. Iannicelli M, Brancati F, Mougou-Zerelli S, Mazzotta A, Thomas S, Elkhartoufi N, Travaglini L, Gomes C, Ardissino GL, Bertini E, Boltshauser E, Castorina P, D'Arrigo S, Fischetto R, Leroy B, Loget P, Bonnière M, Starck L, Tantau J, Gentilin B, Majore S, Swistun D, Flori E, Lalatta F, Pantaleoni C, Penzien J, Grammatico P; International JSRD Study Group, Dallapiccola B, Gleeson JG, Attie-Bitach T, Valente EM. **Novel TMEM67 mutations and genotype-phenotype correlates in meckelin-related ciliopathies.** Hum Mutat. 2010 May;31(5):E1319-31. IF: 5.956
49. 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. IF: 3.206
50. Filoni C, Caciotti A, Carraresi L, Cavicchi C, Parini R, Antuzzi D, Zampetti A, Feriozzi S, Poisetti P, Garman SC, Guerrini R, Zammarchi E, **Donati MA**, Morrone A. **Functional studies of new GLA gene mutations leading to conformational Fabry disease.** Biochim Biophys Acta. 2010 Feb;1802(2):247-52. IF: 4.579
51. **Donati MA. The key-role of the pediatrician in the diagnosis of accumulation diseases: the example of Pompe's disease.** Minerva Pediatr. 2009 Dec;61(6):692-5.
52. Filippi L, Gozzini E, Fiorini P, Malvagia S, la Marca G, **Donati MA. N-carbamylglutamate in emergency management of hyperammonemia in neonatal acute onset propionic and methylmalonic aciduria.** Neonatology. 2010;97(3):286-90. IF: 1.920
53. Blau N, Bélanger-Quintana A, Demirkol M, Feillet F, Giovannini M, MacDonald A, Trefz FK, van Spronsen F; European PKU centers. **Management of phenylketonuria in Europe: survey results from 19 countries.** Mol Genet Metab. 2010 Feb;99(2):109-15. IF: 2.629
54. Travaglini L, Brancati F, Attie-Bitach T, Audollent S, Bertini E, Kaplan J, Perrault I, Iannicelli M, Mancuso B, Rigoli L, Rozet JM, Swistun D, Tolentino J, Dallapiccola B, Gleeson JG, Valente EM; International JSRD Study Group, Zankl A, Leventer R, Grattan-Smith P, Janecke A, D'Hooghe M, Sznajder Y, Van Coster R, Demerleir L, Dias K, Moco C, Moreira A, Kim CA, Maegawa G, Petkovic D, Abdel-Salam GM, Abdel-Aleem A, Zaki MS, Marti I, Quijano-Roy S, Sigaudy S, de Lonlay P, Romano S, Touraine R, Koenig M, Lagier-Tourenne C, Messer J, Collignon P, Wolf N, Philipp H, Kitsiou Tzeli S, Halldorsson S, Johannsdottir J, Ludvigsson P, Phadke SR, Udani V, Stuart B, Magee A, Lev D, Michelson M, Ben-Zeev B, Fischetto R, Benedicenti F, Stanzial F, Borgatti R, Accorsi P, Battaglia S, Fazzi E, Giordano L, Pinelli L, Boccone L, Bigoni S, Ferlini A, **Donati MA**, et al. **Expanding CEP290 mutational spectrum in ciliopathies.** Am J Med Genet A. 2009 Oct;149A(10):2173-80. IF: 2.555
55. Caciotti A, Di Rocco M, Filocamo M, Grossi S, Traverso F, d'Azzo A, Cavicchi C, Messeri A, Guerrini R, Zammarchi E, **Donati MA**, Morrone A. **Type II sialidosis: review of the clinical spectrum and identification of a new splicing defect with chitotriosidase assessment in two patients.** J Neurol. 2009 Nov;256(11):1911-5. IF: 2.359
56. Spinazzola A, Invernizzi F, Carrara F, Lamantea E, **Donati A**, Dirocco M, Giordano I, Meznaric-Petrusa M, Baruffini E, Ferrero I, Zeviani M. **Clinical and molecular features of mitochondrial DNA depletion syndromes.** J Inher Metab Dis. 2009 Apr;32(2):143-58. IF: 2.691
57. Cavicchi C, Malvagia S, la Marca G, Gasperini S, **Donati MA**, Zammarchi E, Guerrini R, Morrone A, Pasquini E. **Hypocitrullinemia in expanded newborn screening by LC-MS/MS is not a reliable marker for ornithine transcarbamylase deficiency.** J Pharm Biomed Anal. 2009 Jul 12;49(5):1292-5. IF: 2.629
58. Gasparotto N, Tomanin R, Frigo AC, Niizawa G, Pasquini E, Blanco M, **Donati MA**, Keutzer J, Zacchello F, Scarpa M. **Rapid diagnostic testing procedures for lysosomal storage disorders: alpha-glucosidase and beta-galactosidase assays on dried blood spots.** Clin Chim Acta. 2009 Apr;402(1-2):38-41. IF: 2.960
59. Bembi B, Cerini E, Danesino C, **Donati MA**, Gasperini S, Morandi L, Musumeci O, Parenti G, Ravaglia S, Seidita F, Toscano A, Vianello A. **Diagnosis of glycogenosis type II.** Neurology. 2008 Dec 2;71(23 Suppl 2):S4-11. IF: 7.043
60. Bembi B, Cerini E, Danesino C, **Donati MA**, Gasperini S, Morandi L, Musumeci O, Parenti G, Ravaglia S,

Seidita F, Toscano A, Vianello A. **Management and treatment of glycogenosis type II.** Neurology. 2008 Dec 2;71(23 Suppl 2):S12-36. IF: 7.043

61. la Marca G, Malvagia S, Casetta B, Pasquini E, **Donati MA**, Zammarchi E. **Progress in expanded newborn screening for metabolic conditions by LC-MS/MS in Tuscany: update on methods to reduce false tests.** J Inher Metab Dis. 2008 Dec;31 Suppl 2:S395-404. IF: 2.691
62. Carraresi L, Parini R, Filoni C, Caciotti A, Sersale G, Tomatsu S, Orlando C, Zammarchi E, Guerrini R, **Donati MA**, Morrone A. **GALNS gene expression profiling in Morquio A patients' fibroblasts.** Clin Chim Acta. 2008 Nov;397(1-2):72-6. IF: 2.960
63. Caciotti A, **Donati MA**, d'Azzo A, Salvioli R, Guerrini R, Zammarchi E, Morrone A. **The potential action of galactose as a "chemical chaperone": increase of beta galactosidase activity in fibroblasts from an adult GM1-gangliosidosis patient.** Eur J Paediatr Neurol. 2009 Mar;13(2):160-4. IF: 1.421
64. Brancati F, Travaglini L, Zablocka D, Boltshauser E, Accorsi P, Montagna G, Silhavy JL, Barrano G, Bertini E, Emma F, Rigoli L; International JSRD Study Group, Dallapiccola B, Gleeson JG, Valente EM. **RPGRIP1L mutations are mainly associated with the cerebello-renal phenotype of Joubert syndrome-related disorders.** Clin Genet. 2008 Aug;74(2):164-70. IF: 3.206
65. Filoni C, Caciotti A, Carraresi L, **Donati MA**, Mignani R, Parini R, Filocamo M, Soliani F, Simi L, Guerrini R, Zammarchi E, Morrone A. **Unbalanced GLA mRNAs ratio quantified by real-time PCR in Fabry patients' fibroblasts results in Fabry disease.** Eur J Hum Genet. 2008 Nov;16(11):1311-7. IF: 3.925
66. Traverso M, Bruno C, Broccolini A, Sotgia F, **Donati MA**, Assereto S, Gazzero E, Lo Monaco M, Modoni A, D'Amico A, Gasperini S, Ricci E, Zara F, Lisanti M, Minetti C. **Truncation of Caveolin-3 causes autosomal-recessive Rippling Muscle Disease.** J Neurol Neurosurg Psychiatry. 2008 Jun;79(6):735-7. IF: 4.622
67. Pittis MG, Donnarumma M, Montalvo AL, Dominissini S, Kroos M, Rosano C, Stroppiano M, Bianco MG, **Donati MA**, Parenti G, D'Amico A, Ciana G, Di Rocco M, Reuser A, Bembi B, Filocamo M. **Molecular and functional characterization of eight novel GAA mutations in Italian infants with Pompe disease.** Hum Mutat. 2008 Jun;29(6):E27-36. IF: 7.033
68. la Marca G, Malvagia S, Pasquini E, Innocenti M, Fernandez MR, **Donati MA**, Zammarchi E. **The inclusion of succinylacetone as marker for tyrosinemia type I in expanded newborn screening programs.** Rapid Commun Mass Spectrom. 2008;22(6):812-8. IF: 2.772
69. Traverso M, Gazzero E, Assereto S, Sotgia F, Biancheri R, Stringara S, Giberti L, Pedemonte M, Wang X, Scapolan S, Pasquini E, **Donati MA**, Zara F, Lisanti MP, Bruno C, Minetti C. **Caveolin-3 T78M and T78K missense mutations lead to different phenotypes in vivo and in vitro.** Lab Invest. 2008 Mar;88(3):275-83. IF: 4.580
70. Caciotti A, **Donati MA**, Adami A, Guerrini R, Zammarchi E, Morrone A. **Different genotypes in a large Italian family with recurrent hereditary fructose intolerance.** Eur J Gastroenterol Hepatol. 2008 Feb;20(2):118-21. IF: 2.080
71. Pela I, Seracini D, **Donati MA**, Lavoratti G, Pasquini E, Materassi M. **Peritoneal dialysis in neonates with inborn errors of metabolism: is it really out of date?** Pediatr Nephrol. 2008 Jan;23(1):163-8. IF: 2.321
72. Pela I, **Donati MA**, Procopio E, Fiorini P. **Lesch-Nyhan syndrome presenting with acute renal failure in a 3-day-old newborn.** Pediatr Nephrol. 2008 Jan;23(1):155-8. IF: 2.321
73. Verbeek MM, Steenbergen-Spanjers GC, Willemsen MA, Hol FA, Smeitink J, Seeger J, Grattan-Smith P, Ryan MM, Hoffmann GF, **Donati MA**, Blau N, Wevers RA. **Mutations in the cyclic adenosine monophosphate response element of the tyrosine hydroxylase gene.** Ann Neurol. 2007 Oct;62(4):422-6. IF: 9.935
74. Cerini E, Bini M, **Donati A**, Andaloro L, Compagnoni G. **Pompe's disease: the role for early diagnosis and treatment.** Pediatr Med Chir. 2007 Sep-Oct;29(5):270-2.
75. Malvagia S, Papi L, Morrone A, **Donati MA**, Ciani F, Pasquini E, la Marca G, Scholte HR, Genuardi M, Zammarchi E. **Fatal malonyl CoA decarboxylase deficiency due to maternal uniparental isodisomy of the telomeric end of chromosome 16.** Ann Hum Genet. 2007 Nov;71(Pt 6):705-12. IF: 2.195
76. la Marca G, Malvagia S, Pasquini E, Innocenti M, **Donati MA**, Zammarchi E. **Rapid 2nd-tier test for**

measurement of 3-OH-propionic and methylmalonic acids on dried blood spots: reducing the false-positive rate for propionylcarnitine during expanded newborn screening by liquid chromatography-tandem mass spectrometry. Clin Chem. 2007 Jul;53(7):1364-9. IF: 5.579

77. Fernandez-Vizarra E1, Bugiani M, Goffrini P, Carrara F, Farina L, Procopio E, **Donati A**, Uziel G, Ferrero I, Zeviani M. **Impaired complex III assembly associated with BCS1L gene mutations in isolated mitochondrial encephalopathy.** Hum Mol Genet. 2007 May 15;16(10):1241-52. IF: 7.249
78. Caciotti A, **Donati MA**, Procopio E, Filocamo M, Kleijer W, Wuyts W, Blaumeiser B, d'Azzo A, Simi L, Orlando C, McKenzie F, Fiumara A, Zammarchi E, Morrone A. **GM1 gangliosidosis: molecular analysis of nine patients and development of an RT-PCR assay for GLB1 gene expression profiling.** Hum Mutat. 2007 Feb;28(2):204. IF: 7.033
79. Parenti G, Zuppaldi A, Gabriela Pittis M, Rosaria Tuzzi M, Annunziata I, Meroni G, Porto C, Donaudy F, Rossi B, Rossi M, Filocamo M, **Donati A**, Bembi B, Ballabio A, Andria G. **Pharmacological enhancement of mutated alpha-glucosidase activity in fibroblasts from patients with Pompe disease.** Mol Ther. 2007 Mar;15(3):508-14. IF: 5.970
80. la Marca G, Casetta B, Malvagia S, Pasquini E, Innocenti M, **Donati MA**, Zammarchi E. **Implementing tandem mass spectrometry as a routine tool for characterizing the complete purine and pyrimidine metabolic profile in urine samples.** J Mass Spectrom. 2006 Nov;41(11):1442-52. IF: 2.940
81. **Donati MA**, Malvagia S, Pasquini E, Morrone A, La Marca G, Garavaglia B, Toniolo D, Zammarchi E. **Barth syndrome presenting with acute metabolic decompensation in the neonatal period.** J Inherit Metab Dis. 2006 Oct;29(5):684. IF: 2.691
82. Pela I, Gasperini S, Pasquini E, **Donati MA**. **Hyperkalemia after acute metabolic decompensation in two children with vitamin B12-unresponsive methylmalonic acidemia and normal renal function.** Clin Nephrol. 2006 Jul;66(1):63-6. IF: 1.4013
83. la Marca G, Malvagia S, Casetta B, Pasquini E, Pela I, Hirano M, **Donati MA**, Zammarchi E. **Pre- and post-dialysis quantitative dosage of thymidine in urine and plasma of a MNGIE patient by using HPLC-ESI-MS/MS.** J Mass Spectrom. 2006 May;41(5):586-92. IF: 2.940
84. Cavicchi C, **Donati MA**, Funghini S, la Marca G, Malvagia S, Ciani F, Poggi GM, Pasquini E, Zammarchi E, Morrone A. **Genetic and biochemical approach to early prenatal diagnosis in a family with mutant methylmalonic aciduria.** Clin Genet. 2006 Jan;69(1):72-6. IF: 3.206
85. Cavicchi C, **Donati MA**, Pasquini E, Poggi GM, Dionisi-Vici C, Parini R, Zammarchi E, Morrone A. **Mutational spectrum in ten Italian patients affected by methylmalonyl-CoA mutase deficiency.** J Inherit Metab Dis. 2005;28(6):1175-8. IF: 1.722
86. Malvagia S, La Marca G, Casetta B, Gasperini S, Pasquini E, **Donati MA**, Zammarchi E. **Falsely elevated C4-carnitine as expression of glutamate formiminotransferase deficiency in tandem mass spectrometry newborn screening.** J Mass Spectrom. 2006 Feb;41(2):263-5. IF: 2.940
87. Caciotti A, **Donati MA**, Bardelli T, d'Azzo A, Massai G, Luciani L, Zammarchi E, Morrone A. **Primary and secondary elastin-binding protein defect leads to impaired elastogenesis in fibroblasts from GM1-gangliosidosis patients.** Am J Pathol. 2005 Dec;167(6):1689-98. IF: 5.796
88. Melis D, Parenti G, Gatti R, Casa RD, Parini R, Riva E, Burlina AB, Dionisi Vici C, 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. IF: 3.412
89. Malvagia S, Morrone A, Pasquini E, Funghini S, la Marca G, Zammarchi E, **Donati MA**. **First prenatal molecular diagnosis in a family with holocarboxylase synthetase deficiency.** Prenat Diagn. 2005 Dec;25(12):1117-9. IF: 1.640
90. Bertini E, **Donati MA**, Broda P, Cassandrini D, Petrini S, Dionisi-Vici C, Ballerini L, Boldrini R, D'Amico A, Pasquini E, Minetti C, Santorelli FM, Bruno C. **Phenotypic heterogeneity in two unrelated Danon patients associated with the same LAMP-2 gene mutation.** Neuropediatrics. 2005 Oct;36(5):309-13. IF: 1.377

91. Funghini S, Morrone A, Pasquini E, Zammarchi E, **Donati MA. Successful prenatal molecular diagnosis of carbamyl-phosphate synthetase I deficiency in two at-risk pregnancies.** J Inherit Metab Dis. 2005;28(5):801-2. IF: 1.722
92. la Marca G, Malvagìa S, Pasquini E, **Donati MA**, Gasperini S, Procopio E, Zammarchi E. **Hyperhydroxyprolinaemia: a new case diagnosed during neonatal screening with tandem mass spectrometry.** Rapid Commun Mass Spectrom. 2005;19(6):863-4. IF: 3.087
93. Caciotti A, **Donati MA**, Boneh A, d'Azzo A, Federico A, Parini R, Antuzzi D, Bardelli T, Nosi D, Kimonis V, Zammarchi E, Morrone A. **Role of beta-galactosidase and elastin binding protein in lysosomal and nonlysosomal complexes of patients with GM1-gangliosidosis.** Hum Mutat. 2005 Mar;25(3):285-92. IF: 7.923
94. Castori M, Valente EM, **Donati MA**, Salvi S, Fazzi E, Procopio E, Galluccio T, Emma F, Dallapiccola B, Bertini E; Italian MTS Study Group. **NPHP1 gene deletion is a rare cause of Joubert syndrome related disorders.** J Med Genet. 2005 Feb;42(2):e9.
95. Bugiani M, Invernizzi F, Alberio S, Briem E, Lamantea E, Carrara F, Moroni I, Farina L, Spada M, **Donati MA**, Uziel G, Zeviani M. **Clinical and molecular findings in children with complex I deficiency.** Biochim Biophys Acta. 2004 Dec 6;1659(2-3):136-47.
96. Bruno C, van Diggelen OP, Cassandrini D, Gimpelev M, Giuffrè B, **Donati MA**, Introvini P, Alegria A, Assereto S, Morandi L, Mora M, Tonoli E, Mascelli S, Traverso M, Pasquini E, Bado M, Vilarinho L, van Noort G, Mosca F, DiMauro S, Zara F, Minetti C. **Clinical and genetic heterogeneity of branching enzyme deficiency (glycogenosis type IV).** Neurology. 2004 Sep 28;63(6):1053-8. IF: 5.973
97. Cosma MP, Pepe S, Parenti G, Settembre C, Annunziata I, Wade-Martins R, Di Domenico C, Di Natale P, Mankad A, Cox B, Uziel G, Mancini GM, Zammarchi E, **Donati MA**, Kleijer WJ, Filocamo M, Carrozzo R, Carella M, Ballabio A. **Molecular and functional analysis of SUMF1 mutations in multiple sulfatase deficiency.** Hum Mutat. 2004 Jun;23(6):576-81. IF: 6.845
98. Malvagìa S, Morrone A, Caciotti A, Bardelli T, d'Azzo A, Ancora G, Zammarchi E, **Donati MA. New mutations in the PPBG gene lead to loss of PPCA protein which affects the level of the beta-galactosidase/neuraminidase complex and the EBP-receptor.** Mol Genet Metab. 2004 May;82(1):48-55. IF: 2.502
99. la Marca G, Malvagìa S, **Donati MA**, Morrone A, Pasquini E, Zammarchi E. **Rapid diagnosis of medium chain Acyl Co-A dehydrogenase (MCAD) deficiency in a newborn by liquid chromatography/tandem mass spectrometry.** Rapid Commun Mass Spectrom. 2003;17(23):2688-92. IF: 2.789
100. Funghini S, **Donati MA**, Pasquini E, Zammarchi E, Morrone A. **Structural organization of the human carbamyl phosphate synthetase I gene (CPS1) and identification of two novel genetic lesions.** Hum Mutat. 2003 Oct;22(4):340-1. IF: 6.328
101. Lucchiari S, **Donati MA**, Melis D, Filocamo M, Parini R, Bresolin N, Comi GP. **Mutational analysis of the AGL gene: five novel mutations in GSD III patients.** Hum Mutat. 2003 Oct;22(4):337. IF: 6.328
102. Malvagìa S, Poggi GM, Pasquini E, **Donati MA**, Pela I, Morrone A, Zammarchi E. **The de novo Q167K mutation in the POU1F1 gene leads to combined pituitary hormone deficiency in an Italian patient.** Pediatr Res. 2003 Nov;54(5):635-40. IF: 3.064
103. Caciotti A, Morrone A, Domenici R, **Donati MA**, Zammarchi E. **Severe prognosis in a large family with hypokalemic periodic paralysis.** Muscle Nerve. 2003 Feb;27(2):165-9. IF: 2.282
104. Lucchiari S, **Donati MA**, Parini R, Melis D, Gatti R, Bresolin N, Scarlato G, Comi GP. **Molecular characterisation of GSD III subjects and identification of six novel mutations in AGL.** Hum Mutat. 2002 Dec;20(6):480. IF: 6.894
105. Bardelli T, **Donati MA**, Gasperini S, Ciani F, Belli F, Blau N, Morrone A, Zammarchi E. **Two novel genetic lesions and a common BH4-responsive mutation of the PAH gene in Italian patients with hyperphenylalaninemia.** Mol Genet Metab. 2002 Nov;77(3):260-6. IF: 2.476
106. Funghini S, **Donati MA**, Pasquini E, Gasperini S, Ciani F, Morrone A, Zammarchi E. **Two new mutations in**

children affected by partial biotinidase deficiency ascertained by newborn screening. J Inherit Metab Dis. 2002 Aug;25(4):328-30. IF: 1.623

107. Venturi N, Rovelli A, Parini R, Menni F, Brambillasca F, Bertagnolio F, Uziel G, Gatti R, Filocamo M, **Donati MA**, Biondi A, Goldwurm S. **Molecular analysis of 30 mucopolysaccharidosis type I patients: evaluation of the mutational spectrum in Italian population and identification of 13 novel mutations.** Hum Mutat. 2002 Sep;20(3):231. IF: 6.894
108. Morrone A, Malvagia S, **Donati MA**, Funghini S, Ciani F, Pela I, Boneh A, Peters H, Pasquini E, Zammarchi E. **Clinical findings and biochemical and molecular analysis of four patients with holocarboxylase synthetase deficiency.** Am J Med Genet. 2002 Jul 22;111(1):10-8. IF: 10.649
109. Bisanzi S, Morrone A, **Donati MA**, Pasquini E, Spada M, Strisciuglio P, Parenti G, Parini R, Papadia F, Zammarchi E. **Genetic analysis in nine unrelated Italian patients affected by OTC deficiency: detection of novel mutations in the OTC gene.** Mol Genet Metab. 2002 Jun;76(2):137-44. IF: 2.476
110. Pela I, **Donati MA**, Zammarchi E. **Effect of ramipril in a patient with glycogen storage disease type I and nephrotic-range proteinuria** J Inherit Metab Dis. 2001 Nov;24(6):681-2. IF: 1.790
111. Spinazzola A, Marti R, Nishino I, Andreu AL, Naini A, Tadesse S, Pela I, Zammarchi E, **Donati MA**, Oliver JA, Hirano M **Altered thymidine metabolism due to defects of thymidine phosphorylase.** J Biol Chem. 2002 Feb 8;277(6):4128-33. IF: 6.696
112. Funghini S, Pasquini E, Cappellini M, **Donati MA**, Morrone A, Fonda C, Zammarchi E. **3-Hydroxy-3-methylglutaric aciduria in an Italian patient is caused by a new nonsense mutation in the HMGCL gene.** Mol Genet Metab. 2001 Jul;73(3):268-75. IF: 2.345
113. Salvi S, Santorelli FM, Bertini E, Boldrini R, Meli C, **Donati A**, Burlina AB, Rizzo C, Di Capua M, Fariello G, Dionisi-Vici C. **Clinical and molecular findings in hyperornithinemia-hyperammonemia-homocitrullinuria syndrome.** Neurology. 2001 Sep 11;57(5):911-4. IF: 5.212
114. Poggi GM, Lamantea E, Ciani F, **Donati MA**, Carrara F, Bartalena L, Garavaglia B, Zammarchi E. **Fatal neonatal outcome in a case of muscular mitochondrial DNA depletion.** J Inherit Metab Dis. 2000 Nov;23(7):755-7. IF: 1.307
115. Bonten EJ, Arts WF, Beck M, Covanis A, **Donati MA**, Parini R, Zammarchi E, d'Azzo A. **Novel mutations in lysosomal neuraminidase identify functional domains and determine clinical severity in sialidosis.** Hum Mol Genet. 2000 Nov 1;9(18):2715-25. IF: 9.048
116. Ciani F, **Donati MA**, Tulli G, Poggi GM, Pasquini E, Rosenblatt DS, Zammarchi E. **Lethal late onset cblB methylmalonic aciduria.** Crit Care Med. 2000 Jun;28(6):2119-21. IF: 3.824
117. Ciani F, Poggi GM, Pasquini E, **Donati MA**, Zammarchi E. **Prolonged exclusive breast-feeding from vegan mother causing an acute onset of isolated methylmalonic aciduria due to a mild mutase deficiency.** Clin Nutr. 2000 Apr;19(2):137-9.
118. Nishino I, Spinazzola A, Papadimitriou A, Hammans S, Steiner I, Hahn CD, Connolly AM, Verloes A, Guimarães J, Maillard I, Hamano H, **Donati MA**, Semrad CE, Russell JA, Andreu AL, Hadjigeorgiou GM, Vu TH, Tadesse S, Nygaard TG, Nonaka I, Hirano I, Bonilla E, Rowland LP, DiMauro S, Hirano M. **Mitochondrial neurogastrointestinal encephalomyopathy: an autosomal recessive disorder due to thymidine phosphorylase mutations.** Ann Neurol. 2000 Jun;47(6):792-800. IF: 8.480
119. Morrone A, Bardelli T, **Donati MA**, Giorgi M, Di Rocco M, Gatti R, Parini R, Ricci R, Taddeucci G, D'Azzo A, Zammarchi E. **Beta-galactosidase gene mutations affecting the lysosomal enzyme and the elastin-binding protein in GM1-gangliosidosis patients with cardiac involvement.** Hum Mutat. 2000;15(4):354-66. IF: 3.666
120. Giorgi M, Morrone A, **Donati MA**, Ciani F, Bardelli T, Biasucci G, Zammarchi E. **Lymphocyte mRNA analysis of the ornithine transcarbamylase gene in Italian OTCD male patients and manifesting carriers: identification of novel mutations.** Hum Mutat. 2000 Apr;15(4):380-1. IF: 3.666
121. Bettinelli A, Rusconi R, Ciarmatori S, Righini V, Zammarchi E, **Donati MA**, Isimbaldi C, Bevilacqua M, Cesareo L, Tedeschi S, Garavaglia R, Casari G. **Gitelman disease associated with growth hormone deficiency,**

disturbances in vasopressin secretion and empty sella: a new hereditary renal tubular-pituitary syndrome? *Pediatr Res.* 1999 Aug;46(2):232-8.

122. Patrizi AL, Tiziano F, Zappata S, **Donati MA**, Neri G, Brahe C. **SMN protein analysis in fibroblast, amniocyte and CVS cultures from spinal muscular atrophy patients and its relevance for diagnosis.** *Eur J Hum Genet.* 1999 Apr;7(3):301-9.
123. Minetti C, Sotgia F, Bruno C, Scartezzini P, Broda P, Bado M, Masetti E, Mazzocco M, Egeo A, **Donati MA**, Volonte D, Galbiati F, Cordone G, Bricarelli FD, Lisanti MP, Zara F. **Mutations in the caveolin-3 gene cause autosomal dominant limb-girdle muscular dystrophy.** *Nat Genet.* 1998 Apr;18(4):365-8.
124. Zammarchi E, Ciani F, Pasquini E, Buonocore G, Shih VE, **Donati MA**. **Neonatal onset of hyperornithinemia-hyperammonemia-homocitrullinuria syndrome with favorable outcome.** *J Pediatr.* 1997 Sep;131(3):440-3. Erratum in: *J Pediatr* 1998 Apr;132(4):747.
125. D'Adamo P, Fassone L, Gedeon A, Janssen EA, Bione S, Bolhuis PA, Barth PG, Wilson M, Haan E, Orstavik KH, Patton MA, Green AJ, Zammarchi E, **Donati MA**, Toniolo D. **The X-linked gene G4.5 is responsible for different infantile dilated cardiomyopathies.** *Am J Hum Genet.* 1997 Oct;61(4):862-7.
126. Mora M, Cartegni L, Di Blasi C, Barresi R, Bione S, Raffaele di Barletta M, Morandi L, Merlini L, Nigro V, Politano L, **Donati MA**, Cornelio F, Cobiauchi F, Toniolo D. **X-linked Emery-Dreifuss muscular dystrophy can be diagnosed from skin biopsy or blood sample.** *Ann Neurol.* 1997 Aug;42(2):249-53.
127. Morrone A, Zammarchi E, Scacheri PC, **Donati MA**, Hoop RC, Servidei S, Galluzzi G, Hoffman EP. **Asymptomatic dystrophinopathy.** *Am J Med Genet.* 1997 Mar 31;69(3):261-7.
128. Ciani F, Pasquini E, Ciardetti A, **Donati MA**, Zammarchi E. **Hyperglycinemia in clinical-laboratory practice.** *Pediatr Med Chir.* 1997 Mar-Apr;19(2):109-12.
129. Zammarchi E, **Donati MA**, Pasquini E, Ciani F, Lori S, Fonda C. **Electromyographic alterations in hyperphenylalaninemia due to dihydropteridine reductase deficiency.** *J Child Neurol.* 1997 Feb;12(2):137-9.
130. Baccetti T, Defraia E, **Donati MA**. **Craniofacial abnormalities associated with congenital fiber type disproportion myopathy.** *J Clin Pediatr Dent.* 1997 Winter;21(2):167-71.
131. Zammarchi E, **Donati MA**, Morrone A, Donzelli GP, Zhou XY, d'Azzo A. **Early-infantile galactosialidosis: clinical, biochemical, and molecular observations in a new patient.** *Am J Med Genet.* 1996 Aug 23;64(3):453-8.
132. Zammarchi E, Filippi L, Novembre E, **Donati MA**. **Biochemical evaluation of a patient with a familial form of leucine-sensitive hypoglycemia and concomitant hyperammonemia.** *Metabolism.* 1996 Aug;45(8):957-60.
133. Zammarchi E, Filippi L, Fonda C, Benedetti PA, Pistone D, **Donati MA**. **Different neurologic outcomes in two patients with neonatal hyperinsulinemic hypoglycemia.** *Childs Nerv Syst.* 1996 Jul;12(7):413-6.
134. Zammarchi E, **Donati MA**, Filippi L, Resti M. **Cryptogenic hepatitis masking the diagnosis of ornithine transcarbamylase deficiency.** *J Pediatr Gastroenterol Nutr.* 1996 May;22(4):380-3.
135. Zammarchi E, **Donati MA**, Ciani F. **Transient neonatal nonketotic hyperglycinemia: a 13-year follow-up.** *Neuropediatrics.* 1995 Dec;26(6):328-30.
136. de Martino M, Zammarchi E, Filippi L, **Donati MA**, Mannelli F, Galli L, Vierucci A. **Redox potential status in children with perinatal HIV-1 infection treated with zidovudine.** *AIDS.* 1995 Dec;9(12):1381-3.
137. Zammarchi E, **Donati MA**, Ciani F, Rubetti P, Pasquini E. **Fructose-1,6-diphosphatase deficiency misdiagnosed as Reye syndrome.** *Clin Pediatr (Phila).* 1995 Oct;34(10):561-4.
138. Zammarchi E, **Donati MA**, Ciani F, Pasquini E, Pela I, Fiorini P. **Failure of early dextromethorphan and sodium benzoate therapy in an infant with nonketotic hyperglycinemia.** *Neuropediatrics.* 1994 Oct;25(5):274-6.
139. Zammarchi E, Savelli A, **Donati MA**, Pasquini E. **Self-mutilation in a patient with mucopolipidosis III.** *Pediatr*

- Neurol. 1994 Jul;11(1):68-70.
140. Zammarchi E, **Donati MA**, Masi S, Sarti A, Castelli S. **Familial infantile myasthenia: a neuromuscular cause of respiratory failure.** Childs Nerv Syst. 1994 Jul;10(5):347-9.
 141. Zammarchi E, **Donati MA**, Tucci F, Fonda C, Fanelli F, Pazzaglia R. **Acute onset of X-linked adrenoleukodystrophy mimicking encephalitis.** Brain Dev. 1994 May-Jun;16(3):238-40.
 142. Baccetti T, Pierleoni L, Filippi L, **Donati MA**, Tollaro I, Zammarchi E. **Dental and craniofacial findings in a child affected by glycogen storage disease type III.** J Clin Pediatr Dent. 1994 Fall;19(1):55-60.
 143. Picco P, Leveratto L, Cama A, Vigliarolo MA, Levato GL, Gattorno M, Zammarchi E, **Donati MA.** **Immotile cilia syndrome associated with hydrocephalus and precocious puberty: a case report.** Eur J Pediatr Surg. 1993 Dec;3 Suppl 1:20-1.
 144. Ciafaloni E, Ricci E, Shanske S, Moraes CT, Silvestri G, Hirano M, Simonetti S, Angelini C, **Donati MA**, Garcia C, et al. **MELAS: clinical features, biochemistry, and molecular genetics.** Ann Neurol. 1992 Apr;31(4):391-8.
 145. Zammarchi E, Vichi GF, Falorni S, Bartolini P, **Donati MA**, Lenzi G. **Osteomyelitis caused by atypical mycobacteria in multiple colonies.** Pediatr Med Chir. 1987 Jan-Feb;9(1):57-61.
 146. Matteini M, Cotozzoli G, Relli P, Zammarchi E, **Donati MA**, Lazzari T. **A new variant of Sutcliffe's metaphysial dysostosis associated with Madelung's disease.** Arch Putti Chir Organi Mov. 1984;34:381-90.
 147. Bartolozzi G, Bernini C, Ciampi C, Perri I, Zammarchi E, **Donati A**, Vichi GF, Giovannucci ML, Allegra M. **A cluster of mucopolipidosis III in a village of Calabria.** Pediatr Med Chir. 1984 Mar-Apr;6(2):285-94.
 148. Milanesi A, Zammarchi E, Bernini G, **Donati MA**, Bardini MR. **Case report of a stage IV S neuroblastoma regressing without any aggressive treatment.** Pediatr Med Chir. 1983 Jul-Aug;5(4):227-30.
 149. Zammarchi E, Bardini MR, **Donati MA**, Savelli A. **Results of screening the urine of 26,014 newborn infants for amino acid disorders.** Riv Neurobiol. 1981 Jul-Dec;27(34):681-6.

diverse 05-12-2016

Maria Alice Donati

