CURRICULUM VITAE

PERSONAL INFORMATION

Name	**ELISABETTA PASQUINI**
Address	Professional address: Children's Hospital Meyer viale Pieraccini, 24 50139 Firenze, Italy
Telephone	+39 (0) 55 5662852
Fax	+39 (0) 55 5662569
E-mail	e.pasquini@meyer.it
Nationality	Italian
Date of birth	1st January 1960

EXPERTISE

- Principal areas of expertise

Therapeutic Area/Indication	Notes
1. Neonatal Screening	Congenital Hypotiroidism and Expanded newborn screening congenital hypothyroidism registry handled by college health ISS Metabolic Diseases registry collaboration drafting Recommendations congenital hypothyroidism
2. Metabolic disorders	Pilot Project Newborn Screening for Pompe Disease, Fabry Disease, MPS1
3. Organic acidurias	collaboration drafting Hyperphenilalaninemia Recommendations in diagnosis, therapy and care collaboration drafting guidelines expanded Newborn screening

WORK EXPERIENCE

• Dates (from – to)	2016
• Name and address of employer	CHILDREN'S HOSPITAL MEYER VIALE PIERACCINI, 24 50139 FIRENZE, ITALY
• Type of business or sector	NEUROSCIENCE ECCELLENCE CENTER
• Occupation or position held	RESPONSIBLE FOR CLINICAL NEONATAL SCREENING UNIT Didactic activity as a consultant professor

• Main activities and responsibilities	*regional screening center for metabolic diseases, screening center for congenital hyperthyroidism, cooperation with laboratory of newborn screening, day hospital and hospitalization for Metabolic and Muscular disease Unit; dealing with research in the following areas: amino acid diseases, organic aciduria, urea cycle diseases, accumulation lysosomal diseases, mitochondrial and peroxisome diseases, neuromuscular diseases and neurotransmitter metabolism alterations*
• Dates (from – to)	2012
• Name and address of employer	CHILDREN'S HOSPITAL MEYER VIALE PIERACCINI, 24 50139 FIRENZE, ITALY
• Type of business or sector	NEUROSCIENCE DEPARTMENT
• Occupation or position held	RESPONSIBLE FOR NEONATAL SCREENING CENTRE, DIAGNOSIS CONFIRMATION AND THERAPY Didactic activity as a consultant professor
• Main activities and responsibilities	*regional screening center for metabolic diseases, screening center for congenital hyperthyroidism, cooperation with laboratory of newborn screening, day hospital and hospitalization for Metabolic and Muscular disease Unit; dealing with research in the following areas: amino acid diseases, organic aciduria, urea cycle diseases, accumulation lysosomal diseases, mitochondrial and peroxisome diseases, neuromuscular diseases and neurotransmitter metabolism alterations*
• Dates (from – to)	2006
• Name and address of employer	Children's Hospital Meyer via I. Giordano, 13 Firenze, Italy University of Florence
• Type of business or sector	Metabolic and Neuromuscolar Unit Department of pediatrics
• Occupation or position held	Responsible for Newborn Screening Clinical Centre and Metabolic Diseases Laboratory (biochemical area) Didactic activity as a consultant professor
• Main activities and responsibilities	Metabolic and Muscolar disease Unit (including laboratory, regional screening centre for hyperthyroidism and metabolic diseases, day hospital and hospitalization department; dealing with research in the following areas: amino acid diseases, organic aciduria, urea cycle diseases, accumulation lysosomal diseases, mitochondrial and peroxisome diseases, neuromuscular diseases and neurotransmitter metabolism alterations)
• Dates (from – to)	1997- 2006
• Name and address of employer	Children's Hospital Meyer via I. Giordano, 13 Firenze, Italy University of Florence
• Type of business or sector	Metabolic and Neuromuscolar Unit Department of pediatrics
• Occupation or position held	assistent
• Main activities and responsibilities	Pediatric Department I (dealing with General Pediatrics and Pediatric Specialties as hereditary neuromuscular metabolic diseases, pediatric neurology and neonatology)
• Dates (from – to)	1993-1997
• Name and address of employer	Children's Hospital Meyer via I. Giordano, 13 Firenze, Italy University of Florence
• Type of business or sector	Endocrinology Unit Department of pediatrics
• Occupation or position held	assistent
• Main activities and responsibilities	

EDUCATION AND TRAINING

• Dates (from – to)	**1986**
• Name and type of organization providing education and training	Medical School, University of Florence,Florence, Italy
• Principal subjects/occupational skills covered	
• Title of qualification awarded	MD
• Level in national classification (if appropriate)	

• Dates (from – to)	**November 1986**
• 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 **1990**
• Name and type of organization providing education and training	University of Florence, Florence
• Principal subjects/occupational skills covered	specialization in pediatrics
• Title of qualification awarded	Pediatrician doctor
• Level in national classification (if appropriate)	

• Dates (from – to)	**1991 – 1993**
• Name and type of organization providing education and training	University of Pavia, Pavia PhD
• Principal subjects/occupational skills covered	Medical Science of Pediatric Age
• Title of qualification awarded	Research Doctoral Degree
• Level in national classification (if appropriate)	

• Dates (from – to)	***1991-1992***
• Name and type of organization providing education and training	University of Siena, Siena
• Principal subjects/occupational skills covered	Course in Neuropediatrics
• Title of qualification awarded	Neuropediatrician Doctor

• Dates (from – to)	**2012**
• Name and type of organization providing education and training	REGION 4 GENETICS COLLABORATIVE MAYO CLINIC, ROCHESTER, MINNESOTA
• Principal subjects/occupational skills covered	Training program for Newborn Screening by MS/MS
• Title of qualification awarded	
• Level in national classification (if appropriate)	

PERSONAL SKILLS AND COMPETENCES

Acquired in the course of life and career but not necessarily covered by formal certificates and diplomas.

Laboratory and diagnostic activity in the hereditary metabolic encephalopathy and neuromuscular diseases

Organic acidurias, biotinidase deficiencies, aminoacidopathies, urea cycle defects, accumulation lysosomial diseases, mitochondrial and peroxisome diseases, neuromuscular diseases and neurotransmitter metabolism alterations

Neonatal Screening for hypothyroidism and metabolic diseases

day hospital and hospitalization department; dealing with research

MOTHER TONGUE **Italian**

OTHER LANGUAGES

English

• Reading skills	Good
• Writing skills	Good
• Verbal skills	Good

SOCIAL SKILLS AND COMPETENCES

Living and working with other people, in multicultural environments, in positions where communication is important and situations where teamwork is essential (eg. Culture and sports), etc.. •

- *Tutorial for students* during graduation in Medicine, Biology, specialization in Pediatrics, Medical Genetics
- Didactic activity as a consultant professor in Pediatrics, Genetic Medicine physiotherapy school at University of Florence
- training courses in Neuropediatrics, Pediatric Nutrition and Neonatology
- Expanded.Newborn screening organization
- transport organization from birth centres to Meyer Hospital with delivery company
- evaluation and selection cut-off,
- interpreting analytical data,
- family contacts
- collaborations with other centres and hospitals

ORGANISATIONAL SKILLS AND COMPETENCES

Coordination and administration of people, projects and budgets at work

TECHNICAL SKILLS AND COMPETENCES *With computers, specific kinds of equipment, machinery, etc.*	Database management system Microsoft word, open office, power point Use and interpretation of GC/MS software
ADDITIONAL INFORMATION	Appointment to External Committees -- 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 (SIMMESN) Counselor and Vice President from 2009 to 2012 - Member of the Society for the Study of Inborn Errors of Metabolism (SSIEM)

SCIENTIFIC PUBLYCATIONS LIST (LAST THREE YEARS, SINCE 2006-) WHTH IMPACT FACTOR FOR EACH PUBLICATION

Huemer M, Diodato D, Schwahn B, Schiff M, Bandeira A, Benoist JF, Burlina A, Cerone R, Couce ML, Garcia-Cazorla A, la Marca G, Pasquini E, Vilarinho L, Weisfeld-Adams JD, Kožich V, Blom H, Baumgartner MR, Dionisi-Vici C.
Guidelines for diagnosis and management of the cobalamin-related remethylation disorders cblC, cblD, cblE, cblF, cblG, cblJ and MTHFR deficiency. J Inherit Metab Dis. 2016 Nov 30. [Epub ahead of print]

Cassone M, Ferradini V, Longo G, Sarchielli P, Murasecco D, Romoli M, Pasquini E, Novelli G, Prontera P, Sangiuolo F. Genotype-phenotype correlation of F484L mutation in three Italian families with Thomsen myotonia.
Muscle Nerve. 2016 Sep 17. doi: 10.1002/mus.25407. [Epub ahead of print] No abstract available.

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. doi: 10.1016/j.bbacli.2016.03.004.

Poggi C, Giusti B, Gozzini E, Sereni A, Romagnuolo I, Kura A, Pasquini E, Abbate R, Dani C. Genetic Contributions to the Development of Complications in Preterm Newborns. 2015 Jul 14;10(7):e0131741. doi: 10.1371/journal.pone.0131741. eCollection 2015.

Carducci C, Santagata S, Friedman J, Pasquini E, Carducci C, Tolve M, Angeloni A, Leuzzi V. Urine sepiapterin excretion as a new diagnostic marker for sepiapterin reductase deficiency. Mol Genet Metab. 2015 Aug;115(4):157-60. doi: 10.1016/j.ymgme.2015.06.009. Epub 2015 Jun 25.

Tonin R, Caciotti A, Funghini S, la Marca G, Pasquini E, Cayton E, Mooney SD, Guerrini R, Morrone A.Biotinidase deficiency due to a de novo mutation or gonadal mosaicism in a first child. Clin Chim Acta. 2015 May 20;445:70-2. doi: 10.1016/j.cca.2015.03.010. Epub 2015 Mar18.

Huemer M, Kožich V, Rinaldo P, Baumgartner MR, Merinero B, Pasquini E, Ribes A, Blom HJ. Newborn screening for homocystinurias and methylation disorders: systematic review and proposed guidelines. J Inherit Metab Dis. 2015 Nov;38(6):1007-19. doi: 10.1007/s10545-015-9830-z. Epub 2015 Mar 12.

Olivieri A, Fazzini C, Medda E; Italian Study Group for Congenital Hypothyroidism. Multiple factors influencing the incidence of congenital hypothyroidism detected by neonatal screening. Horm Res Paediatr. 2015;83(2):86-93. doi: 10.1159/000369394. Epub 2015 Jan 6.

Olivieri A, Radetti G, Medda E; Italian Study Group for Congenital Hypothyroidism. Incidence of congenital hypothyroidism in the Autonomous Province of Bolzano: benefit of increased iodine intake. J Endocrinol Invest. 2015 Feb;38(2):185-7. doi: 10.1007/s40618-014-0206-2. Epub 2014 Nov 18

Cavicchi C, Donati M, Parini R, Rigoldi M, Bernardi M, Orfei F, 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. Orphanet J Rare Dis. 2014 Jul 16;9(1):105. doi:10.1186/s13023-014-0105-9 [Epub ahead of print]

Diodato D, Invernizzi F, Lamantea E, Fagiolari 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 Rep. 2014 Apr 17. doil 10.1007/8904_2014_300 [Epub ahead of print]

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. ScientificWorldJournal. 2013 Oct 31;2013:625824. doi: 10.1155/2013/625824. eCollection 2013

Olivieri A, Corbetta C, Weber G, Vigone MC, Fazzini C, Medda E; Italian Study Group for Congenital Hypothyroidism. Congenital hypothyroidism due to defects of thyroid development and mild increase of TSH at screening: data from the Italian National Registry of infants with congenital hypothyroidism. J Clin Endocrinol Metab. 2013 Apr;98(4):1403-8. doi: 10.1210/jc.2012-3273.Epub2013 Feb 12.Review

Cassio A, Corbetta C, Antonozzi I, Calaciura F, Caruso U, Cesaretti G, Gastaldi R, Medda E, Mosca F, Pasquini E, Salerno MC, Stoppioni V, Tonacchera M, Weber G, Olivieri A; Italian Society for Pediatric Endocrinology and Diabetology; Italian Society for the Study of Metabolic Diseases and Neonatal Screening; Italian National Institute of Health; Italian National Coordinating Group for Congenital Hypothyroidism; Italian Thyroid Association; Italian Society of Pediatrics; Italian Society of Neonatology; Italian Society of Endocrinology; Associazione Medici Endocrinologi. The Italian screening program for primary congenital hypothyroidism: actions to improve screening, diagnosis, follow-up, and surveillance J Endocrinol Invest. 2013 Mar;36(3):195-203. doi: 10.3275/8849. Epub 2013 Feb 12. Review.

Olivieri A; Italian Study Group for Congenital Hypothyroidism. Epidemiology of congenital hypothyroidism: what can be deduced from the Italian registry of infants with congenital hypothyroidism. J Matern Fetal Neonatal Med. 2012 Oct;25(Suppl 5):7-9. doi: 10.3109/14767058.2012.714641. Review.

Poggi C, Giusti B, Vestri A, Pasquini E, Abbate R, Dani C. Genetic polymorphisms of antioxidant enzymes in preterm infants. J Matern Fetal Neonatal Med. 2012 Oct;25 Suppl 4:131-4.

Marquardt G, Currier R, McHugh DM, Gavrilov D, Magera MJ, Matern D, Oglesbee D, Raymond K, Rinaldo P, Smith EH, Tortorelli S, Turgeon CT, Lorey F, Wilcken B, Wiley V, Greed LC, Lewis B, Boemer F, Schoos R, Marie S, Vincent MF, Sica YC, Domingos MT, Al-Thihli K, Sinclair G, Al-Dirbashi OY, Chakraborty P, Dymerski M, Porter C, Manning A, Seashore MR, Quesada J, Reuben A, Chrastina P, Hornik P, Atef Mandour I, Atty Sharaf SA, Bodamer O, Dy B, Torres J, Zori R, Cheillan D, Vianey-Saban C, Ludvigson D, Stembridge A, Bonham J, Downing M, Dotsikas Y, Loukas YL, Papakonstantinou V, Zacharioudakis GS, Baráth A, Karg E, Franzson L, Jonsson JJ, Breen NN, Lesko BG, Berberich SL, Turner K, Ruoppolo M, Scolamiero E, Antonozzi I, Carducci C, Caruso U, Cassanello M, la Marca G, Pasquini E, Di Gangi IM, Giordano G, Camilot M, Teofoli F, Manos SM, Peterson CK, Mayfield Gibson SK, Sevier DW, Lee SY, Park HD, Khneisser I, Browning P, Gulamali-Majid F, Watson MS, Eaton RB, Sahai I, Ruiz C, Torres R, Seeterlin MA, Stanley EL, Hietala A, McCann M, Campbell C, Hopkins PV, de Sain-Van der Velden MG, Elvers B, Morrissey MA, Sunny S, Knoll D, Webster D, Frazier DM, McClure JD, Sesser DE, Willis SA, Rocha H, Vilarinho L, John C, Lim J, Caldwell SG, Tomashitis K, Castiñeiras Ramos DE, Cocho de Juan JA, Rueda Fernández I, Yahyaoui Macías R, Egea-Mellado JM, González-Gallego I, Delgado Pecellin C, García-Valdecasas Bermejo MS, Chien YH, Hwu WL, Childs T, McKeever CD, Tanyalcin T, Abdulrahman M, Queijo C, Lemes A, Davis T, Hoffman W, Baker M, Hoffman GL. Enhanced interpretation of newborn screening results without analyte cutoff values. Genet Med. 2012 Jul;14(7):648-55. doi: 10.1038/gim.2012.2. Epub 2012 Feb 16.

Grünert SC, Stucki M, Morscher RJ, Suormala T, Bürer C, Burda P, Christensen E, Ficicioglu C, Herwig J, Kölker S, Möslinger D, Pasquini E, Santer R, Schwab KO, Wilcken B, Fowler B, Yue WW, Baumgartner MR. 3-Methylcrotonyl-CoA Carboxylase Deficiency: Clinical, biochemical, enzymatic and molecular studies in 88 individuals. Orphanet J Rare Dis. 2012 May 29;7(1):31. [Epub ahead of print]

Giusti B, Vestrini A, Poggi C, Magi A, Pasquini E, Abbate R, Dani C. Genetic polymorphisms of antioxidant enzymes as risk factors for oxidative stress-associated complications in preterm infants. Free Radic Res. 2012 Sep;46(9):1130-9. Epub 2012 Jun 13.

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, Tifft 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. Epub 2011 Apr 7.

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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. doi: 10.1016/j.jpba.2009.03.001. Epub 2009 Mar 20

la Marca G, Malvagia S, Funghini S, Pasquini E, Moneti G, Guerrini R, Zammarchi E. The successful inclusion of succinylacetone as a marker of tyrosinemia type I in Tuscany newborn screening program. Rapid Commun Mass Spectrom. 2009 Dec;23(23):3891-3.

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. Epub 2008 Dec 13.

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Filippi L, Pezzati M, Poggi C, Pasquini E. Vein of Galen aneurysmal malformation and galactosemia in a neonate: a previously unreported association. Paediatr Anaesth. 2007 Dec;17(12):1221-3.

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. Epub 2007 Sep 26.

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.

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. Epub 2007 May 17.

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Elisabetta Pasquini