PERSONAL INFORMATION

Serena Gasperini

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Sex Female | Date of birth 05/01/1969 | Nationality Italian

JOB APPLIED FOR
POSITION

Medical Doctor, Paediatricians

WORK
EXPERIENCE:

I am a Resident physician, in charge at the Metabolic Unit-Paediatric Clinic ASST-Monza San Gerardo Hospital in Monza.
I work with a metabolic expert team including nursing, secretary, data manager, dieticians. We take care of many patients with metabolic diseases: lysosomal storage diseases (MPS, Pompe disease, Fabry, Gaucher, Niemann Pick), Urea cycle disorders and other aminoacidopathies, organic acidurias, fatty acids defects and glycogen storage diseases, glycosylation defects. I am also involved in newborn screening in Monza since 2015. I have a large experience in diagnosis, treatment and follow-up of patients diagnosed by newborn screening for my past experience in Florence at Meyer Children Hospital where I attended Paediatric specialization and I worked as physician in Metabolic and Neuromuscular disorders Unit and in Pediatric Neurology Department since 2004 till 2011.
During last 20 years I acquired expertise in many metabolic and neurometabolic disorders, in laboratory and molecular assay, in clinical trials.
When I worked in Meyer Hospital in Tuscany, I acquired a great experience in expanded newborn screening by tandem mass spectrometry: the pilot program to detect about 40 metabolic diseases started in 2002 in Tuscany, as the first region in Italy.
I participated to many clinical trials (expecially for lysosomal storage disorders) and many of them are still going.
In particular I am principal investigators for intratecal Study for MPSIIIA Phase IIb (HGT-SAN-093 and SHP-610-201) and for Orphazyme study Phase II/III on Niemann Pick type C (NPC-002). I attend to many Registries: Pompe, Fabry (FOS), Hunter (HOS), MPSVI Tyrosinemia type I. I collaborate with Fondazione Tettamanti for research on Bone alterations on mice affected by MPSI and with TIGET of San Raffaele Hospital in Milano for next gene therapy in MPSI patients.
The list of trials and ICH-GCP Experience are attached.

EDUCATION AND TRAINING

QUALIFICATIONS

	Qualification	Date	Granting Organisation
1.	MD degree	20 October 1994	*University of Florence*
2.	Licence for Medical Practice	May 1995	*Ministry of Health*
3.	Diploma of Territorial First Aid Medicine (D.E.U.)	December 1996	*U.S.L. 3 Hospital of Pistoia*
4.	Diploma of Pediatrics	24 October 2000	*University of Florence*
5.	Diploma of Neonatal Echography	2003	*Rome*
8.	Diploma of pediatric advanced life suppport (PALS)	2011	*Italian Resuscitation Council and University of Milano Bicocca*
9.	University Master course concerning Rare pediatric diseases: methodologies for assistance, learning and research	2010	*University of Florence - Department for Women and Childrens' Health*

10. Medical registration: Ordine dei Medici di Pistoia 1995 n° 1557

PRESENT POSITION

POSITION	Resident physician at the Metabolic Unit-Paediatric Clinic ASST-Monza San Gerardo Hospital Monza-Italy
ORGANISATION	ASST-Monza San Gerardo Hospital- Fondazione MBBM
FULL ADDRESS	METABOLIC RARE DISEASES UNIT – Paediatric Clinic Fondazione MBBM ASST-Monza San Gerardo Hospital Via Pergolesi, 33, 20900 Monza-Italy
START DATE	Since july 2011

PAST POSITIONS HELD

Position	Date (from-to)	Institution/Organisation
1. Resident physician at the Ereditary Metabolic Unit and Neuro- muscular disorders (including laboratory, regional screening centre for hyperthyroidism and metabolic diseases, day hospital and hospitalization department)	From 16 January 2004 to 15 July 2011	*Azienda Ospedaliero-Universitaria Meyer of Florence, Metabolic and Neurology Unit*
2. Permanent Hospital position at the Pediatric Operative Unit	From 1 November 2002 to 15 January 2004	*Hospital of Valdichiana (Siena)*

3. Temporary Hospital position at the Pediatric Operative Unit	From 16 April to 30 September 2002	*Hospital Concern of Valdichiana (Siena)*
4. Consultant at the Operative Unit of the Pediatric Department I (Metabolic and Neuromuscular rare disorders Unit)	From 26 March 2001 to 31 March 2002	*Azienda Ospedaliero-Universitaria Meyer of Firenze*
5. Fellowship at Neonatal Intensive Care Department	*From year 1997*	*Azienda Ospedaliera Careggi of Florence*
6. Fellowship at Pediatrics Department I and Neurometabolic Diseases laboratory	*From year 1996*	*Azienda Ospedaliero-Universitaria Meyer of Florence*
7. Residency at the Pediatrics Department I	*1995-1996*	*Azienda Ospedaliero-Universitaria Meyer of Florence*
8. Post-lauream apprenticeship	*November 1994- May 1995*	*Azienda Ospedaliera Careggi of Florence*

Mother tongue(s) ITALIANO

Other language(s)

	UNDERSTANDING		SPEAKING		WRITING
	Listening	Reading	Spoken interaction	Spoken production	
English	B1	B2	B1	B2	B2

Levels: A1/A2: Basic user - B1/B2: Independent user - C1/C2 Proficient user
Common European Framework of Reference for Languages

Communication skills Good communication skills gained through my experience in Hospital with parents (counselling) and children; volunteer experience in emergency unit

Organisational / managerial skills
- Good competence of team-leading acquired during my experience in emergency situations and in coordination of multidiciplinary team in Hospital

ICH-GCP Experience

ICH-GCP BASIC COURSE	**19/11/09 And 31/05/2012**	**During SIV for SHIRE Study HGT-MLD-049 And during BIOMARIN study MOR**
Therapeutic Area/Indication	**Date (from-to)**	**Notes**
1. Glycogen storage disease type II (Pompe disease)	2004	For therapeutic trial with MYOZYME (Enzymatic replacement therapy with acid maltase)
2. Glycogen storage disease type II (Pompe disease)	2006-2008	Multicenter project for Italian Guidelines
3. A multi center open label extension study of HGT-1111 (recombinant human arylsulfatase A) treatment of patients with Late Infantile Metachromatic Leukodystrophy – Shire HGT-MLD-049	2009-2010	International multicenter clinical trial for Shire HGT, Inc.

study		
4. A multi center study of ERT in MORQUIO Syndrome (MPS IV)	2010	International multicenter clinical trial for BIOMARIN
5. A randomized, controlled, open label, multicenter, Phase IIb for safety and efficacy of HGT-1410 administration via an intratecal drug delivery in Pediatric patients with MPS IIIA (HGT-SAN-093)	2015-2016	International trial Phase IIb, sponsored by Shire
6. An open label Extension study Phase IIb for safety and effeicacy of HGT-1410 administration via an intratecal drug delivery in Pediatric patients with MPS IIIA (SHP-610-201)	2016	International trial Phase IIb, extension study sponsored by Shire
7. Phase II/III study on arimocvlomol (heat shock protein) on Niemann Pick Type C patients	2015-2017	International multicenter clinical trial of ORPHAZYME

Digital competence

SELF-ASSESSMENT				
Information processing	Communication	Content creation	Safety	Problem solving
Independent user	Independent user	Independent user	Independent user	Independent user

Levels: Basic user - Independent user - Proficient user
Digital competences - Self-assessment grid

Driving licence

A and B

ADDITIONAL INFORMATION

I am a member of Italian scientific society as Società Italiana di Pediatria (SIP) and Società italiana di malattie metaboliche ereditarie e screening neonatale SIMMESN oltre che del “Society for the Study of Inborn Errors of Metabolism (SSIEM).

Medical References:

M.A. Donati, **S. Gasperini,** F. Ciani, P. Fiorini, C. Dani, C. Profeti, I. Pela, A. Morrone, E. Zammarchi:

“Distrofia Miotonica congenita: patologia non rara clinicamente eterogenea”. Atti della “Settimana Pediatrica Nazionale” tenutasi a Torino dal 20 al 24 settembre 1998 pubblicati su ***Rivista Italiana di Pediatria 1998, 24 (Suppl. 4): 163.***

M.A. Donati, C. Fonda, F. Ciani, **S. Gasperini**, E. Zammarchi:

“Le leucodistrofie”. Aggiornamento monografico.

Neurologia Pediatrica 1998; II: 2-13

M.A. Donati, **S. Gasperini**, F. Ciani, C. Fonda, G. Bieber, E. Zammarchi:

"Le nuove leucodistrofie: 2 casi di Infantile onset vacuolating leukoencephalopathy".

Atti (poster e comunicazione orale) del XXIV Congresso Nazionale della Società Italiana di Neuropediatria tenutosi a Viterbo dal 8 al 10 ottobre 1998. Neurologia Pediatrica 1998;II (Suppl. al N. 1):130.

M.A. Donati, E. Pasquini, **S. Gasperini**, F. Ciani, N. Blau, E. Zammarchi:

"Ipotonia e distonie: ipotizzare un difetto genetico del metabolismo dei neurotrasmettitori".

Atti del "XXV Congresso Nazionale della società italiana di Neuropediatria" tenutosi a Siena dal 2 al 4 Dicembre 1999.

M.A. Donati, E. Pasquini, **S. Gasperini**, A. Morrone, L. De Simone, I. Pollini, P. Fiorini, D. Toniolo, E. Zammarchi:

"Barth Syndrome an X-linked underdiagnosed cardiomyophathy".

Atti di "MilanoPediatria 2000" tenutosi a Milano dal 25 al 28 Maggio 2000, pag 246.

E. Pasquini, M.A. Donati, **S. Gasperini**, L. Palermo, N. Blau, R.A. Wevers, E. Zammarchi:

"Tyrosine hydroxylase deficiency: effectiveness of treatment with L-Dopa in a patient".

Atti di "MilanoPediatria 2000" tenutosi a Milano dal 25 al 28 Maggio 2000, pag. 227.

M.A. Donati, E. Pasquini, **S. Gasperini**, I. Pela, G.M. Poggi, B. Garavaglia, C. bruno, E. Zammarchi:

"Sintomatologia gastrointestinale e neuromuscolare in malattia mitocondriale". Atti del XXVI Congresso Nazionale della Società Italiana di Neuropediatria tenutosi ad Ancona dal 19 al 21 Ottobre 2000.

S. Gasperini, M.A. Donati, E. Pasquini, I. Pela, S. Funghini, R. Pazzaglia, D. Cianfrini, E. Zammarchi:

"Difetto di piruvato deidrogenasi: eterogeneità clinica e biochimica".

Poster e comunicazione orale presentati al XXVI Congresso

Nazionale della Società Italiana di Neuropediatria tenutosi ad Ancona dal 19 al 21 Ottobre 2000.

M.A. Donati, **S. Gasperini**, F. Ciani, S. Funghini, A. Morrone, E. Zammarchi:

"Speech delay may underlie partial biotinidase deficiency".

Atti del Congresso tenutosi a Praga dal 3 al 7 settembre 2001

J. Inherit. Metab. Dis. 2001, 24 (suppl. 1), pag. 58.

T. Bardelli, M.A. Donati, A. Benuzzi, FR. Ciani**, S. Gasperini**, E. Pasquini, A. Morrone, E. Zammarchi:

"Identificazione di mutazioni BH4-sensibili nel gene PAH di pazienti italiani con deficit di fenilalanina idrossilasi".

Atti del 4° Congresso Nazionale S.I.G.U. (Società Italiana di Genetica Umana) tenutosi a Orvieto dal 28 al 30 Novembre 2001.

S. Funghini, M.A. Donati, **S. Gasperini**, E. Pasquini, F. Ciani, A. Morrone, E. Zammarchi:

"Il deficit parziale di biotinidasi correla con la mutazione D444H nel gene BTD".
Atti del Convegno "Malattie Metaboliche ereditarie: passato e futuro tra delusioni, problemi e speranze" organizzato dalla Società Italiana per lo studio delle Malattie Metaboliche ereditarie (SISMME) tenutosi a Napoli dall'8 al 10 Novembre 2001.

M.A. Donati, **S. Gasperini**, F. Ciani, E Pasquini, S. Funghini, E. Zammarchi:

"Ritardo del linguaggio e deficit parziale di biotinidasi".

Comunicazione orale alla Riunione Società Italiana di Neuropediatria (SINP) tenutasi a Roma – Università La Sapienza in data 5 maggio 2001.

S. Funghini, M.A. Donati, E. Pasquini, **S. Gasperini**, F. Ciani, A. Morrone, E. Zammarchi:

"Two new mutations in children affected by partial biotinidase deficiency ascertained by newborn screening".

J. Inherit. Metab. Dis. 2002, 25, pag. 328-330 (short report)

S. Lori, M.A. Donati, **S. Gasperini**, M.R. Bardini, R. Nistri, G. Poggi, B. Garavaglia, E. Zammarchi:

"Combined deficiency of pyruvate dehydrogenase and mithocondrial respiratory chain complex I case report".

Journal of Peripheral Nervous System, ***Volume 7, n°1 Marzo 2002, pag. 78.***

M.A. Donati, S. Funghini, E. Pasquini, **S. Gasperini**, F. Ciani, A. Morrone, E. Zammarchi:
"Deficit parziale di biotinidasi correla con la mutazione D444H nel gene BTD".
Atti del 58° Congresso Italiano di Pediatria tenutosi a Montecatini Terme (PT) dal 28 settembre al 2 ottobre 2002.

T. Bardelli, M.A. Donati, **S. Gasperini**, F. Ciani, F. Belli, N. Blau, A. Morrone, E. Zammarchi:
" Two novel genetic lesions and a common BH4-responsive mutation of the PAH gene in Italian patients with hyperphenilalaninemia".
Mol. Genet. Metab. 2002 Nov; 77(3):260-266.

C. Cavicchi, A. Morrone, E. Pasquini, F. Ciani, **S. Gasperini**, E. Procopio, M. Sibilio, G. Parenti, E. Zammarchi, M.A. Donati. Metilmalonico aciduria con omocistinuria tipo cblC: analisi molecolare in pazienti italiani. Congresso Nazionale congiunto SISSME,SISN,GENCLI, Pesaro 11-13 ottobre 2006

G. La Marca, S. Malvagia, E. Pasquini, M.A. Donati**, S. Gasperini**, E. Procopio,F. Ciani, E. Zammarchi. Screening neonatale per le malattie metaboliche in Toscana mediante LC/MS/MS: prima esperienza italiana. Congresso Nazionale 2006 SISME-SISN-GNCLI, Pesaro 11-13 ottobre 2006

S. Gasperini, M. Cappellini, C. Fonda, S. Funghini, F. Ciani, R. Guerrini, M.A. Donati · Acute psychiatric signs in Wilson disease causing by Zinc acetate overdosage. Poster at international meeting SSIEM in Hamburg, September 2-5th, 2007.

I.Pela, **S. Gasperini**, E.Pasquini and M.A. Donati. Hyperkalemia after acute metabolic decompensation in two children with vitamin B12-unresponsive methylmalonic acidemia and normal renal function. ***Case report in Clinical Nephrology 2006, Vol.66;1:63-66***

M.A.Donati, E. Pasquini, N. Parri, **S. Gasperini**, E. Lamantea, M. Zeviani. Alpers Syndrome due to POLG1 mutation: clinical, electrophysiological, neuroradiologcal studies in two siblings. Abstract for 42th Annual Symposium, Paris 6-9 September 2005

E. Pasquini**, S. Gasperini**, C. Minetti, E. Zammarchi, MA Donati. Glycogen branching enzyme deficiency (GSD IV) presenting as a congenital myopathy.

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C. Cavicchi, S. Malvagia, G. la Marca, **S. Gasperini,** M.A. Donati, E. Zammarchi, R. Guerrini, A. Morrone, E. Pasquini. Hypocitrullinemia in expanded newborn screening by LC-MS/MS is not a reliable marker for onithine transcarbamilase deficiency. ***Journal of Pharmaceutical and Biomedical analysis, 2009 Mar 20.***

S. Malvagia, G la Marca, B Casetta, **S. Gasperini**, E. Pasquini, M.A. Donati, E. Zammarchi. 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.***

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L. Ferri, F.M. Vaz, E. Bertini, S. Malvagia, **S. Gasperini**, S. Catarzi, B.I. Albert, R. Guerrini, M.A. Donati, A. Morrone. Two

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New motor outcome function measures in evaluation of late-onset Pompe disease before and after enzyme replacement therapy.
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Il sottoscritto è a conoscenza che, ai sensi dell'art. 7**6** del DPR 445/2000**, le dichiarazioni mendaci, la falsità negli atti e l'uso di atti falsi sono puniti ai sensi del codice penale e delle leggi speciali.**

Inoltre, il sottoscritto autorizza al trattamento dei dati personali, secondo quanto previsto dalla Legge 196/2003 e dall' ***art. 13 GDPR 679/16***

Monza, 22 gennaio 2020 **Serena Gasperini**