


## PERSONAL INFORMATION

## Maddalena Casale

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Date of birth 06/11/1980 | Nationality Italian

## PREFERRED JOB

## Clinical Research in Pediatrics and Pediatric Haematology

## WORK EXPERIENCE

18/11/2016–Present

## Researcher and MD

Università degli Studi "Luigi Vanvitelli", Naples (Italy)

- Teaching of Pediatrics at the School of Medicine, Università degli Studi della Campania "Luigi Vanvitelli"

- Teaching of Pediatrics at the university course in Paediatric Nursing Service, Università degli Studi della Campania "Luigi Vanvitelli"

- Teaching of Pediatrics at the university course in Nursing Service, Università degli Studi della Campania "Luigi Vanvitelli"

01/09/2011–Present

## MD at Regional Coordinating Centre for Thalassemia and Haemoglobinopathies "A. Cardarelli"

Azienda Ospedaliera di Rilievo Nazionale "A. Cardarelli", Naples (Italy)

Clinical Duties

Research

Attended Courses

15/12/2014–15/04/2016

## Clinical Research Coordinator of the the international multicentre study "DeEP- Deferiprone Evaluation in Paediatrics- Multicentre, randomised, open label, non-inferiority active-controlled trial to evaluate the efficacy and safety of deferiprone compared to deferasirox in paediatric patients aged from 1 month to less than 18 years affected by transfusion dependent haemoglobinopathies", founded from the European Union Seventh Framework Programme (FP7/2007-2013), 2011-2015

Consorzio Valutazioni Biologiche e Farmacologiche (CVBF), Pavia (Italy)

Production of scientific documentation and study procedures

Scientific support to Investigators, Clinical Research Associates and all parts involved in study conduct

Advice about all clinical and scientific issues related to study conduct

23/05/2011–03/10/2011

## MD at "G. Rummo" Hospital

"G. Rummo" Hospital, Benevento (Italy)

Business or sector Pediatric Emergency Department and Pediatric Ward

01/07/2010–31/01/2011

## Honorary Clinical Fellow in Paediatrics, Evelina Children's Hospital, London

Dr Baba Inusa

King's College London, Evelina Children's Hospital, Guy's and St Thomas' NHS

Foundation Trust, London (UK)

Clinical Duties  
Research

Attended Courses  
Teaching

18/10/2009–16/04/2010 **Clinical Fellow in Pediatric Emergency, "Bambino Gesù" Children Hospital**

Dr Antonino Reale  
"Bambino Gesù" Children Hospital, Rome (Italy)

**Business or sector** Pediatric Emergency

01/03/2009–31/07/2009 **Clinical Fellow in Pediatric Cardiology, "Monaldi" Hospital, Naples**

Dr Maria Giovanna Russo, Dipartimento di Scienze Cardio- Toraciche e Respiratorie  
"Monaldi" Hospital, Napoli (Italy)

**Business or sector** Pediatric Cardiology

EDUCATION AND TRAINING

2011–2014 **PhD - Second University of Naples, Department of Pharmacology and Clinical and Experimental Medicine**

Second University of Naples, Naples (Italy)

2005–2010 **Specialization in Pediatrics**

Second University of Naples, Napoli (Italy)

Final score 50/ 50 cum laude

2005 **Medical license**

Ordine dei Medici e degli Odontoiatri di Salerno, Salerno (Italy)

1999–2005 **Degree in Medicine**

Second University of Naples, Napoli (Italy)

Final score 110/ 110 cum laude

1994–1999 **Diploma of "Maturità Classica"**

Liceo Classico "M.T. Cicerone", Sala Consilina (SA) (Italy)

final score 100/ 100

PERSONAL SKILLS

Mother tongue(s) Italian

Foreign language(s)

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

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

ADDITIONAL INFORMATION

## Publications

- **Casale M**, Cozzolino F, Scianguetta S, Pucci P, Monaco V, Sanchez G, Santoro C, Rubino R, Cannata M, Perrotta S. Hb Vanvitelli: A new unstable  $\alpha$ -globin chain variant causes undiagnosed chronic haemolytic anaemia when co-inherited with deletion -  $\alpha(3.7)$ . *Clin Biochem*. 2019 Sep 4. pii: S0009-9120(19)30851-3. doi: 10.1016/j.clinbiochem.2019.09.002.
- Patti G, Scianguetta S, Roberti D, Di Mascio A, Balsamo A, Brugnara M, Cappa M, **Casale M**, Cavarzere P, Cipriani S, Corbetta S, Gaudino R, Iughetti L, Martini L, Napoli F, Peri A, Salerno M, Salerno R, Passeri E, Maghnie M, Perrotta S, Di Iorgi N. Familial neurohypophyseal diabetes insipidus in 13 kindreds and 2 novel mutations in the vasopressin gene. *Eur J Endocrinol*. 2019 Jun 1. pii: EJE-19-0299.R2. doi: 10.1530/EJE-19-0299.
- Russo G, De Franceschi L, Colombatti R, Rigano P, Perrotta S, Voi V, Palazzi G, Fidone C, Quota A, Graziadei G, Pietrangelo A, Pinto V, Ruffo GB, Sorrentino F, Venturelli D, **Casale M**, Ferrara F, Sainati L, Cappellini MD, Piga A, Maggio A, Forni GL. Current challenges in the management of patients with sickle cell disease - A report of the Italian experience. *Orphanet J Rare Dis*. 2019 May 30;14(1):120. doi: 10.1186/s13023-019-1099-0.
- Fermo E, Vercellati C, Marcello AP, Zaninoni A, Aytac S, Cetin M, Capolsini I, **Casale M**, Paci S, Zanella A, Barcellini W, Bianchi P. Clinical and Molecular Spectrum of Glucose-6-Phosphate Isomerase Deficiency. Report of 12 New Cases. *Front Physiol*. 2019 May 7;10:467. doi: 10.3389/fphys.2019.00467.
- Tartaglione I, Manara R, Caiazza M, Carafa PA, Caserta V, Ferrantino T, Granato I, Ippolito N, Maietta C, Oliveto T, **Casale M**, Di Concilio R, Ciancio A, De Michele E, Russo C, Elefante A, Ponticorvo S, Russo AG, Femina G, Canna A, Ermani M, Cirillo M, Esposito F, Centanni A, Gritti P, Perrotta S. Brain functional impairment in beta-thalassaemia: the cognitive profile in Italian neurologically asymptomatic adult patients in comparison to the reported literature. *Br J Haematol*. 2019 Aug;186(4):592-607.
- Russo AG, Ponticorvo S, Tartaglione I, Caiazza M, Roberti D, Elefante A, **Casale M**, Di Concilio R, Ciancio A, De Michele E, Canna A, Cirillo M, Perrotta S, Esposito F, Manara R. No increased cerebrovascular involvement in adult beta-thalassemia by advanced MRI analyses. *Blood Cells Mol Dis*. 2019 Sep;78:9-13.
- Tartaglione I, Russo C, Elefante A, Caiazza M, **Casale M**, Di Concilio R, Ciancio A, De Michele E, Amendola G, Gritti P, Carafa PA, Ferrantino T, Centanni A, Ippolito N, Caserta V, Oliveto T, Granato I, Femina G, Esposito F, Ponticorvo S, Russo AG, Canna A, Ermani M, Cirillo M, Perrotta S, Manara R. No evidence of increased cerebrovascular involvement in adult neurologically-asymptomatic  $\beta$ -Thalassaemia. A multicentre multimodal magnetic resonance study. *Br J Haematol*. 2019 May;185(4):733-742.
- Rossi F, Tortora C, Palumbo G, Punzo F, Argenziano M, **Casale M**, Di Paola A, Locatelli F, Perrotta S. CB2 Receptor Stimulation and Dexamethasone Restore the Anti-Inflammatory and Immune-Regulatory Properties of Mesenchymal Stromal Cells of Children with Immune Thrombocytopenia. *Int J Mol Sci*. 2019 Feb 28;20(5). pii: E1049. doi: 10.3390/ijms20051049.
- Marzuillo P, Guarino S, **Casale M**, Di Sessa A, Golino R, D'Angelo V, Menna G, Rossi F, Miraglia Del Giudice E, Perrotta S. Nineteen-month-old girl with persistent fever. *Arch Dis Child Educ Pract Ed*. 2019 Feb 23. pii: edpract-2018-316493. doi: 10.1136/archdischild-2018-316493.
- De Franceschi L, Lux C, Piel FB, Gianesin B, Bonetti F, **Casale M**, Graziadei G, Lisi R, Pinto V, Putti MC, Rigano P, Rosso R, Russo G, Spadola V, Pulvirenti C, Rizzi M, Mazzi F, Ruffo G, Forni GL. Access to emergency department for acute events and identification of sickle cell disease in refugees. *Blood*. 2019 Feb 11. pii: blood-2018-09-876508. doi: 10.1182/blood-2018-09-876508.
- Franchini M, Forni GL, Marano G, Cruciani M, Mengoli C, Pinto V, De Franceschi L, Venturelli D, **Casale M**, Amerini M, Capuzzo M, Grazzini G, Masiello F, Pati I, Veropalumbo E, Vaglio S, Pupella S, Liunbruno GM. Red blood cell alloimmunisation in transfusion-dependent thalassaemia: a systematic review. *Blood Transfus*. 2019 Jan;17(1):4-15.
- Roberti D, Conforti R, Giugliano T, Brogna B, Tartaglione I, **Casale M**, Piluso G, Perrotta S. A Novel 12q13.2-q13.3 Microdeletion Syndrome With Combined Features of Diamond Blackfan Anemia, Pierre Robin Sequence and Klippel Feil Deformity. *Front Genet*. 2018 Nov 19;9:549.
- Punzo F, Tortora C, Argenziano M, **Casale M**, Perrotta S, Rossi F. Iron chelating properties of Eltrombopag: Investigating its role in thalassemia-induced osteoporosis. *PLoS One*. 2018 Dec 3;13(12):e0208102. doi: 10.1371/journal.pone.0208102.
- **Casale M**, Filosa A, Ragozzino A, Amendola G, Roberti D, Tartaglione I, De Michele E, Cozzolino D, Rispoli G, Palmieri F, Pugliese U, Scianguetta S, Signoriello G, Musallam KM, Perrotta S. Long-term improvement in cardiac magnetic resonance in  $\beta$ -thalassaemia major patients treated with deferasirox extends to patients with abnormal baseline cardiac function. *Am J Hematol*. 2019

Mar;94(3):312-318.

- **Casale M**, Picariello S, Corvino F, Cerasari G, Scianguetta S, Rossi F, Persico M, Perrotta S. Life-Threatening Drug-Induced Liver Injury in a Patient with  $\beta$ -Thalassemia Major and Severe Iron Overload on Polypharmacy. *Hemoglobin*. 2018 Sep 25:1-4.
- **Casale M**, Marsella M, Ammirabile M, Spasiano A, Costantini S, Cinque P, Ricchi P, Filosa A. Predicting factors for liver iron overload at the first magnetic resonance in children with thalassaemia major. *Blood Transfus*. 2018 Jun 26:1-6.
- Ladogana S, Maruzzi M, Samperi P, Condorelli A, **Casale M**, Giordano P, Notarangelo LD, Farruggia P, Giona F, Nocerino A, Fasoli S, Casciana ML, Miano M, Tucci F, Casini T, Saracco P, Barcellini W, Zanella A, Perrotta S, Russo G; AIHA Committee of the Associazione Italiana di Ematologia ed Oncologia Pediatrica. Second-line therapy in paediatric warm autoimmune haemolytic anaemia. Guidelines from the Associazione Italiana Onco-Ematologia Pediatrica (AIEOP). *Blood Transfus*. 2018 Jul;16(4):352-357.
- **Casale M**, Borriello A, Scianguetta S, Roberti D, Caiazza M, Bencivenga D, Tartaglione I, Ladogana S, Maruzzi M, Della Ragione F, Perrotta S. Hereditary hypochromic microcytic anemia associated with loss-of-function DMT1 gene mutations and absence of liver iron overload. *Am J Hematol*. 2017 Nov 27.
- Colombatti R, Palazzi G, Maserà N, Notarangelo LD, Bonetti E, Samperi P, Barone A, Perrotta S, Facchini E, Miano M, Del Vecchio GC, Guerzoni ME, Corti P, Menzato F, Cesaro S, **Casale M**, Rigano P, Forni GL, Russo G, Sainati L; Italian Multicenter Study of Hydroxyurea in Sickle Cell Anemia Investigators. Hydroxyurea prescription, availability and use for children with sickle cell disease in Italy: Results of a National Multicenter survey. *Pediatr Blood Cancer*. 2018 Feb;65(2).
- **Casale M**, Meloni A, Filosa A, Cuccia L, Caruso V, Palazzi G, Gamberini MR, Pitrolo L, Putti MC, D'Ascola DG, Casini T, Quarta A, Maggio A, Neri MG, Positano V, Salvatori C, Toia P, Valeri G, Midiri M, Pepe A. Multiparametric Cardiac Magnetic Resonance Survey in Children With Thalassemia Major: A Multicenter Study. *Circ Cardiovasc Imaging*. 2015 Aug;8(8):e003230. doi: 10.1161/CIRCIMAGING.115.003230.
- **Casale M**, Citarella S, Filosa A, De Michele E, Palmieri F, Ragozzino A, Amendola G, Pugliese U, Tartaglione I, Della Rocca F, Cinque P, Nobili B, Perrotta S. Endocrine function and bone disease during long-term chelation therapy with deferasirox in patients with  $\beta$ -thalassemia major. *Am J Hematol*. 2014 Dec;89(12):1102-6
- Inusa B, **Casale M**, Booth C, Lucas S. Subarachnoid haemorrhage and cerebral vasculopathy in a child with sickle cell anaemia. *BMJ Case Rep*. 2014 Oct 21;2014.pii: bcr2014205464. doi: 10.1136/bcr-2014-205464. PubMed PMID: 25336550
- Colombatti R, Perrotta S, Samperi P, **Casale M**, Maserà N, Palazzi G, Sainati L, Russo G; on behalf of the Italian Association of Pediatric Hematology-Oncology (AIEOP) Sickle Cell Disease Working Group. Organizing national responses for rare blood disorders: the Italian experience with sickle cell disease in childhood. *Orphanet J Rare Dis*. 2013 Oct 20;8(1):169.
- Ricchi P, Ammirabile M, Spasiano A, Costantini S, Di Matola T, Pepe A, Cinque P, Pagano L, **Casale M**, Filosa A, Prossomariti L. Extramedullary haematopoiesis correlates with genotype and absence of cardiac iron overload in polytransfused adults with thalassaemia. *Blood Transfus*. 2013 Oct 3:1-7.
- Ricchi P, Ammirabile M, Costantini S, Spasiano A, Di Matola T, Cinque P, **Casale M**, Filosa A, Prossomariti L. Nephrolithiasis in patients exposed to deferasirox and desferioxamine: probably an age-linked event with different effects on some renal parameters. *Ann Hematol*. 2013 Jun 27.
- Perrotta S, Stiehl DP, Punzo F, Scianguetta S, Borriello A, Bencivenga D, **Casale M**, Nobili B, Fasoli S, Balduzzi A, Cro L, Nytko KJ, Wenger RH, Della Ragione F. Congenital erythrocytosis associated with gain-of-function HIF2A gene mutations and erythropoietin levels in the normal range. *Haematologica*. 2013 Oct;98(10):1624-32.
- Filosa A, Vitrano A, Rigano P, Calvaruso G, Barone R, Capra M, Cuccia L, Gagliardotto F, Pitrolo L, Prossomariti L, **Casale M**, Caruso V, Gerardi C, Campisi S, Cianciulli P, Rizzo M, D'Ascola G, Ciancio A, Maggio A. Long-term treatment with deferiprone enhances left ventricular ejection function when compared to deferoxamine in patients with thalassemia major. *Blood Cells Mol Dis*. 2013 Aug;51(2):85-8.
- **Casale M**, Cinque P, Ricchi P, Costantini S, Spasiano A, Prossomariti L, Minelli S, Frega V, Filosa A. Effect of splenectomy on iron balance in patients with  $\beta$ -thalassemia major: a long-term follow-up. *Eur J Haematol*. 2013 Jul;91(1):69-73.
- Mancusi S, La Manna A, Bellini G, Scianguetta S, Roberti D, **Casale M**, Rossi F, Della Ragione F, Perrotta S. HNF-1 $\beta$  mutation affects PKD2 and SOCS3 expression causing renal cysts and

diabetes in MODY5 kindred. J Nephrol. 2013 Jan-Feb;26(1):207-12.

- Noris P, Perrotta S, Bottega R, Pecci A, Melazzini F, Civaschi E, Russo S, Magrin S, Loffredo G, Di Salvo V, Russo G, **Casale M**, De Rocco D, Grignani C, Cattaneo M, Baronci C, Dragani A, Albano V, Jankovic M, Scianguetta S, Savoia A, Balduini CL. Clinical and laboratory features of 103 patients from 42 Italian families with inherited thrombocytopenia derived from the monoallelic Ala156Val mutation of GPIIb $\alpha$  (Bolzano mutation). Haematologica. 2012 Jan;97(1):82-8.
- Punzo F, Bertoli-Avella AM, Scianguetta S, Della Ragione F, **Casale M**, Ronzoni L, Cappellini MD, Forni G, Oostra BA, Perrotta S. Congenital dyserythropoietic anemia type II: molecular analysis and expression of the SEC23B gene. Orphanet J Rare Dis. 2011 Dec 30;6:89.
- **Casale M**, Perrotta S. Splenectomy for hereditary spherocytosis: complete, partial or not at all? Expert Rev Hematol. 2011 Dec;4(6):627-35.
- Noris P, Perrotta S, Seri M, Pecci A, Gnan C, Loffredo G, Pujol-Moix N, Zecca M, Scognamiglio F, De Rocco D, Punzo F, Melazzini F, Scianguetta S, **Casale M**, Marconi C, Pippucci T, Amendola G, Notarangelo LD, Klersy C, Civaschi E, Balduini CL, Savoia A. Mutations in ANKRD26 are responsible for a frequent form of inherited thrombocytopenia: analysis of 78 patients from 21 families. Blood. 2011 Jun 16;117(24):6673-80.

**Books:**

- Baba Inusa, **Maddalena Casale** and Nicholas Ward (2016). Introductory Chapter: Introduction to the History, Pathology and Clinical Management of Sickle Cell Disease, Sickle Cell Disease - Pain and Common Chronic Complications, Dr. Baba P.D. Inusa (Ed.), InTech, DOI: 10.5772/65648. Available from: <http://www.intechopen.com/books/sickle-cell-disease-pain-and-common-chronic-complications/introductory-chapter-introduction-to-the-history-pathology-and-clinical-management-of-sickle-cell-di>

**International Conferences**

- 27/05/2016: Apopharma Iron Overload Workshop "Thalassemia Major - Continuum of Care", Turin, Italy. Presentation Title: "Transition of patients from paediatric to adult";
- 20/05/2015: General Assembly of the International Multicentre Study "Deferiprone Evaluation in Paediatrics", Tirana, Albania. Presentation Title: "DEEP-2 Study protocol and the experience of Italian Centres";
- 21/05/2015: General Assembly of the International Multicentre Study "Deferiprone Evaluation in Paediatrics", Tirana, Albania. Presentation Title: "A critical evaluation of the deliverables submitted to the European Commission";
- 9/11/2014: 4th Pan-European Conference on Haemoglobinopathies and rare Anemias, TIF (Thalassemia International Federation), Athens, Greece. Presentation Title: "Early total body chelation to prevent iron damage in paediatric patients";
- January 2014: Curriculum in Iron Metabolism and Related Disorders European School of Haematology, Interactive Poster Session from the 2013 Annual Meeting of the American Society of Haematology (ASH). Presentation Title: "Long term efficacy of iron chelation therapy with deferasirox on endocrine function in thalassemia major";
- 17/06/2013: Medical Expert Workshop on non-transfusion- dependent thalassemia (NTDT), 18th Congress of European Hematology Association (EHA), Stockholm, Sweden. Presentation Title: "Troublesome jaundice in non-transfusion- dependent thalassemia: which is the best treatment?";
- 8-11/03/2012: First International Symposium on Allogeneic Cellular Gene Therapy in Hemoglobinopathies, Minister of Health, Rome, Italy. Presentation Title: "Immigration and SCA in Italy. The current situation in Campania"

**Poster Presentation**

- 19th Congress of European Hematology Association (EHA), Milan, Italy, 12-15 June 2014. Poster Presentation: Casale, S Citarella, G Amendola, G Iolascon, S Costantini, E De Michele, U Pugliese, I Tartaglione, F Gallicola, P Cinque, A Filosa, S Perrotta. Bone mineral density improvement in patients with thalassemia major on long-term chelation therapy with deferasirox.
- American Society of Hematology Annual Meeting, New Orleans - USA, December 6-9, 2013. Poster Presentation: M Casale, S Citarella, A Filosa, E De Michele, U Pugliese, F Palmieri, A Ragozzino, G Amendola, I Tartaglione, M Della Rocca, B Nobili, S Perrotta. Long term efficacy of iron chelation therapy with deferasirox on endocrine function in thalassemia major. Blood 2013; 122:965.
- XXXIX National Congress of Italian Association of Pediatric Haematology and Oncology, Genoa, 25-27/05/2014. Poster Presentation: "Long term therapy with deferasirox prevents and stabilizes endocrine complications in thalassemia major patients: results from a multicentre study".

- XXXIX National Congress of Italian Association of Pediatric Haematology and Oncology, Genoa, 25-27/05/2014. Poster Presentation: "Set-up of the Italian Network on Asplenia for a comprehensive management of asplenic patients: the role of scientific societies".

#### Special projects and assignments

- Selection by the American Society of Hematology (ASH) to receive the 2019 ASH Global Research Award and a research support for the project with the aim to perform a prospective multicenter nationwide analysis on mortality, morbidity, vaccine coverage and response in a large cohort of asplenic patients followed in the Italian Network of Asplenia;
  - Scientific Coordinator for the development of the Italian Guidelines on Endocrinopathies in patients affected by Emoglobinopathies endorsed by the Italian Society of Thalassemia and Emoglobinopathies;
  - Scientific Coordinator of the observational, retrospective/ prospective, multicentre study "National Census of Patients with Asplenia", approved by Coordinating Ethics Committee (Second University of Naples) 14/02/2014;
  - Scientific Coordinator of the observational, retrospective, multicentre study "Retrospective Evaluation of adult and pediatric transfusion- dependent patients treated with deferasirox therapy", ClinicalTrial.gov identifier: NCT01874405
  - Author of the Italian Recommendations for the management of infectious risk in asplenic patients (available at: [www.site.org](http://www.site.org)) Italian Society of Thalassemia and Hemoglobinopathies;
  - Author of the Italian Consensus on the management of acute complications in patients with sickle cell disease" (available at: [www.site.org](http://www.site.org)) Italian Society of Thalassemia and Hemoglobinopathies;
  - Author of the first Italian Recommendations for the management of autoimmune haemolytic anaemia in childhood (available at: [www.aieop.org](http://www.aieop.org)) Italian Association of Pediatric Hematology- Oncology (AIEOP) October 2011/ March 2013;
  - Author of the first Italian Guidelines for the management of sickle cell anemia in childhood (available at: [www.aieop.org](http://www.aieop.org)) Italian Association of Pediatric Hematology- Oncology (AIEOP) September 2008/ May 2010;
- Tutor for students from the university course in Paediatric Nursing Service of the Second University of Naples, Italy, 2011/2012 Second University of Naples, Naples (Italy);
- Collaboration, after competition based on qualifications, on the research project entitled "Citopenie costituzionali, caratterizzazione molecolare, patogenesi e approcci terapeutici innovativi", founded from PRIN 2003, Second University of Naples, (Italy) 2005/ 2006