Europass Curriculum Vitae

Personal information

First name(s) / Surname(s)	**MAIORANA ARIANNA**
Address(es)	
Telephone(s)	+39
E-mail	
Nationality	Italian
Date of birth	
Gender	Female

Desired employment / Occupational field

Clinical practice and research in Metabolic Diseases

Work experience

Aug 2023 →	Head of High Specialization in Metabolic Hypoglycemia, Metabolic Unit of Ospedale Pediatrico Bambino Gesù, piazza S. Onofrio 4, 00165 Rome, Italy
May 2009 →	MD, PhD, Pediatric Assistant at Metabolic Unit of Ospedale Pediatrico Bambino Gesù, piazza S. Onofrio 4, 00165 Rome, Italy
October 2002-October 2004	Trainings: Research fellow at Kimmel Cancer Center, Thomas Jefferson University, Microbiology and Immunology Laboratory of Prof. Baserga R, Philadelphia, PA, USA
November 2004-April 2008	Research fellow at Laboratory of Metabolic Diseases of Rina Balducci Center of Paediatric Endocrinology, Rome, Italy
April 2006-November 2007	Research fellow in Laboratory of Autoimmunity and Organ Regeneration Ospedale Pediatrico Bambino Gesù, Rome, Italy
January 2006-December 2007	Member of the SIEDP (Italian Society of Pediatric Endocrinology and Diabetology) Young Committee

Education

14.7.1999	Medical School Degree at University of Messina, Italy (110 with honours/110)
May 2000	Italian Board
26.11.2004	Residency degree in Pediatrics at Tor Vergata University of Rome, Italy (50 with honours/50)
04.04.2008	PhD research degree in Molecular Pediatrics, Tor Vergata University of Rome, Italy
01.07.2002-01.09.2015	Inclusion on the Association of Physicians and Surgeons of Messina n° 8418
02.09.2015 →	Inclusion on the Association of Physicians and Surgeons of Rome n° 61891/M

Training

01.12.2009	Theoretical and practical course about continuous glucose monitoring in pediatrics, Ospedale Pediatrico Bambino Gesù, Rome, Italy
30.11.2009	Theoretical and practical course about continuous glucose monitoring in pediatrics, Ospedale Pediatrico Bambino Gesù, Rome, Italy
03-07.11.2009	8th European Metabolic Course, Nijmegen, Netherlands
02-06.03.2008	Specialization school of Paediatric Endocrinology and Diabetology SIEDP/ISPED 2008, 1st step, Riccione, Italy
05-09.10.2008	Specialization school of Paediatric Endocrinology and Diabetology SIEDP/ISPED 2008, 2nd, Riccione, Italy
01.03.2003-30.06.2003	English Conversation Class at the Nationalities Service Center of Philadelphia, Philadelphia, Pennsylvania, USA
13-15.09.2001	IV workshop about the quality in the evaluation of the bone age, September 13-15th 2001, Genova, Italy

Personal skills and competences

Mother tongue(s) **Italian**

Other language(s) **English**

Self-assessment	Understanding		Speaking		Writing
European level (*)	Listening	Reading	Spoken interaction	Spoken production	
English	good	good	good	good	good

(*) Common European Framework of Reference for Languages

Social skills and competences: ABILITY TO WORK IN MEDICAL TEAM WHERE IS INDISPENSABLE COLLABORATION BETWEEN DIFFERENT ROLES. ABILITY TO ORGANIZE IN MEDICAL ROUNDS (AFTERNOON, NIGHT, HOLIDAYS AND WEEK-END ON CALL)

Organisational skills and competences: COORDINATION OF PEOPLE, ADMINISTRATION OF RESEARCH PROJECTS, ABILITY TO WORK IN SITUATION OF STRESS

Computer skills and competences	COMPUTER SOFTWARE: MAC, WINDOWS XP, POWER POINT, ADOBE PHOTOSHOP, ADOBE ILLUSTRATOR, INTERNET EXPLORER, GOOGLE CHROME. USE, POSITIONING, INTERPRETATION OF CONTINUOUS GLUCOSE MONITORING SYSTEM
Artistic skills and competences	DANCE, CARIBBEAN DANCE, INTERIOR DESIGN
Other skills and competences	SPORT: TOTAL BODY, GAG, AEROBICS, STEP STYLE, AEROBICS RACE, VOLLEYBALL ,SWIMMING
Driving licence	European Drive licence B

Annexes
Clinical trials

2002-2003	The Genetics and Neuroendocrinology of Short Stature International Study, protocol approved by Eli Lilly Company, Pediatric Department, S. Eugenio Hospital, Tor Vergata University of Rome, Italy
2013-2017	Principal Investigator of An Open-Label, Multicenter, Multinational Study of the Safety, Efficacy and Pharmacokinetics of Asfotase Alfa (human recombinant tissue non-specific alkaline phosphatase fusion protein) in Infants and Children ≤ 5 Years of Age with hypophosphatasia (HPP), ENB-010-10, approved by Alexion, 2013-2017, Metabolic Unit, Pediatric Department, Bambino Gesù Children's Hospital, Rome, Italy
2014-2019	SOBI-NTBC-005. Trial to evaluate long-term safety of Orfadin® in the treatment of Tyrosinemia type 1 in standard clinical care
2017-2021	ALX-HPP-501(Alexion). Hypophasphatasia (HPP) International Registry

Awards

2018-2020	Winner of the Nutricia Metabolics Research Fund for the project entitled "Nutritional ketosis to improve neuroprotection in poorly-responsive Hyperinsulinemic hypoglycemia",
2013	Winner of the prize "Best oral presentation" entitled "Reversible myopathy due to E3 deficiency: chaperone effect of riboflavin", at the National Metabolic and Genetic Meeting SIMMESN-SIMGePed 26th-28th November 2013, Naples, Italy
2009	Winner of the National Prize "Rina Balducci" for the best young researcher's paper in Pediatric Endocrinology 2009: “Impact of growth hormone therapy on adult height of children born small for gestational age: a systematic review of randomized controlled trials”, November 7th 2009, Napoli, Italy
2008	Winner of the financing in Pediatrics, Endocrinology and Auxology of Cesare Serono Foundation 2008, for the project entitled “Isolation and characterization of omental adipose progenitor cells in children: a potential tool to unravel the pathogenesis of metabolic syndrome”.

Most important Congresses and Courses

XXV National Congress of AIGlico (Italian Association of Glycogen Storage Diseases), with personal presentation entitled “Therapy with Empagliflozin in Glycogen storage disease type 1b: review od the literature and update of the Italian restrospective multicentric study”, October 14 th-15th 2023, Hotel Galilei, Pisa, Italy

International Congress “Hyperinsulinism - Novel genes, drugs and guidelines”, with personal presentation entitled "Congenital hyperinsulinism: descriptive analysis of 154 patients from an Italian cohort ", April 13th-14th 2023, The Children's Hospital of Philadelphia, Philadelphia, PA, USA, Accreditation Council for Continuing Medical Education (ACCME)

ICH Good Clinical Practice E6 (R2), The Global Healt Network, e-learning course, Jan 4th 2023

Specialization school of Pediatric Endocrinology and Diabetology SIEDP/ISPED 2022, VIII Edition, 2nd step, with personal presentation entitled "Neonatal hypoglycemia: the diagmostic algorithm", October 18th-21st, Hotel Nautico Riccione, Italy

CHI Family Conference, with personal presentation entitled "Congenital hyperinsulinism (HI) in Italy: Diagnosis and treatment", September16th-18th 2022, Hotel NH Villa Carpegna, Rome, Italy

52nd EMG Conference "Food for thought: lipids for cellular functions", chairman of Workshop Management of Tyrosinemia type I, May 19th-21st 2022, Innsbruck, Austria

Congress "Highlights ICIEM 2021: Hereditary metabolic diseases", with personal presentation entitled "Glycogen storage diseases and galactosemia", April 7th-8th 2022, Hotel Baglioni, Florence, Italy

Basic Life Support (BLS), for health care provider course, November 11th 2021, Ospedale Pediatrico Bambino Gesù, Rome, Italy

Teacher in Univerisity Master's degree in Inborn Errors of Metabolism and Newborn Screening on "Nutritional approach in Glycogen storage diseases and Congenital Hyperinsulinism. Ketogenic diet in inborn errors of metabolism", October 16th 2021, online

Congress "Glycogen storage diseases days", with personal presentation entitled "Growth and overtreatment risk" and "Ketogenic diet in Glycogen storage disease type III: experiences compared - panel discussion", October 15th 2021, NH Hotel RomeCenter, Rome, Italy

Long Chain Fatty Acids Oxidation defects Advisory Board, with personal presentation entitled "Experience con triheptanoin", October 6th 2021, Hotel Minerva, Florence, Italy

Updating meeting "Ketogenic diet 2020: 100 years later" with personal presentation entitled "The ketogenic diet in neurological diseases beyond epilepsy", September 17th 2021, Congress Center FAST, Milan, Italy

XXII National Congress AIG (Italian Association of Glycogen Storage Diseases), with personal presentation entitled "Precision medicine in glycogen storage diseases: the importance of continuous glucose monitoring", October 18th 2020, live streaming

International Congress "Updates in diagnosis and management of Hyperinsulinism and neonatal hypoglycemia", with personal presentation entitled "Efficacy of ketogenic diet in patients with drug-unresponsive congenital hyperinsulinism caused by glucokinase mutations", September 5th-6th 2019, The Children's Hospital of Philadelphia, Philadelphia, PA, USA, Accreditation Council for Continuing Medical Education (ACCME)

Advisory Board Hypophosphatasia, June 19th 2017, Hilton Milan, Milan, Italy

1st European Conference on GLUT1 Deficiency, October 7th-8th 2016, Atahotel Expo Fiera, Milan, Italy

SSIEM Annual Symposium Rome 2016, "Metabolic Pathways, cellular networks and beyond", as speaker with personal communication entitled "Congenital Hyperinsulinism" in plenary session about "Clinical Novelties", and 6 poster presentations, September 6th-9th 2016, Palazzo dei Congressi, Rome, Italy, European Accreditation Council for Continuing Medical Education (EACCME)

47th EMG Conference, as speaker with personal communication entitled "Ketogenic diet in Congenital Hyperinsulinism: a novel approach to prevent brain damage", 11th-13rd June 2015, NH Laguna Palace, Venice Mestre, Italy

EU Partner Meeting about Tyrosinemia type 1 at Hannover Medical School, 12th March 2015, Hannover, Germany

Satellite Symposium: Tyrosinemia type 1: from guidelines to reality, with personal communication of "Imaging and early detection of hepatic malignancy", ICIEM, September 3rd 2013, Barcelona, Spain

Workshop on Hepatorenal Tyrosinemia (HT1), with personal communication of "Renal disease in HT1", November 22nd-23rd 2012, Hannover, Germany

1st advanced meeting on metabolic and genetic disorders affecting the liver, March 28th-30th 2012, Crown Plaza Hotel, Rome, Italy, European Accreditation Council for Continuing Medical Education (EACCME)

Theoretical and practical course about continuous glucose monitoring in pediatrics, November 30th and December 1st 2009, Ospedale Pediatrico Bambino Gesù, Rome, Italy

ENDO 2003, June 19-22th , Philadelphia, PA, USA

XIII National Meeting SIEDP (Italian Society of Paediatric Endocrinology and Diabetology), October 10-13th 2001, Trieste, Italy

Original articles

Maines E, Cardellini MC, Stringari G, Leonardi L, Piccoli G, Urru SAM, Maiorana A, Soffiati M, Franceschi R.
Drug-Induced Hypoglycemia in Neonates Born to Nondiabetic Women Treated with Medications during the Pregnancy or the Labor: A Systematic Review of the Literature.
Am J Perinatol. 2023 Oct 17. doi: 10.1055/s-0043-1776061.

Siri B, D'Alessandro A, Maiorana A, Porzio O, Ravà L, Dionisi-Vici C, Cappa M, Martinelli D.
Adrenocortical function in patients with Single Large Scale Mitochondrial DNA Deletions: a retrospective single centre cohort study.
Eur J Endocrinol. 2023 Nov 8;189(5):485-494. doi: 10.1093/ejendo/lvad137.

Maines E, Urru SAM, Leonardi L, Fancellu E, Campomori A, Piccoli G, Maiorana A, Soffiati M, Franceschi R.
Drug-induced hyperinsulinemic hypoglycemia: An update on pathophysiology and treatment.
Rev Endocr Metab Disord. 2023 Aug 8. doi: 10.1007/s11154-023-09828-y.

Damiani V, Lamolinara A, Cicalini I, Cufaro MC, Del Pizzo F, Di Marco F, Del Boccio P, Dufrusine B, Hahne M, Lattanzio R, Pieragostino D, Iezzi M, Federici M, Turco MC, Maiorana A, Dionisi-Vici C, De Laurenzi V.
Pancreatic beta-cell specific BAG3 knockout results in chronic hyperinsulinemia inducing insulin resistance.
Mol Metab. 2023 Jun 10:101752. doi: 10.1016/j.molmet.2023.101752.

Maines E, Maiorana A, Leonardi L, Piccoli G, Soffiati M, Franceschi R
A narrative review on pathogenetic mechanisms of hyperinsulinemic hypoglycemia in Kabuki syndrome
Endocrine Regulations 2023 Jun 7;57(1):128-137. doi: 10.2478/enr-2023-0016

Maiorana A, Tagliaferri F, Dionisi-Vici C
Current understanding on pathogenesis and effective treatment of glycogen storage disease type Ib with empagliflozin: new insights coming from diabetes for its potential implications in other metabolic disorders.
Front Endocrinol (Lausanne). 2023 Apr 21;14:1145111. doi:10.3389/fendo.2023.1145111

Maines E, Moretti M, Vitturi N, Gugelmo G, Fasan I, Lenzini L, Piccoli G, Gragnaniello V, Maiorana A, Soffiati M, Burlina A, Franceschi R
Understanding the pathogenesis of cardiac complications in patients with propionic acidemia and exploring therapeutic alternatives for those who are not eligible or are waiting for liver transplantation.
Metabolites. 2023 Apr 16;13(4):563. doi: 10.3390/metabo13040563.

Spagnoletti G, Larghi Z, Saffioti MC, Maritato S, Maiorana A, Garganese MC, Dionisi-Vici C, Grimaldi C, Spada M
Pylorus-preserving pancreatoduodenectomy for focal congenital hyperinsulinism in a 5-month-old baby: video report.
Front Surg 2023 Jan 30;9:1085238. doi: 10.3389/fsurg.2022.1085238

Tagliaferri F, Massese M, Russo L, Commone A, Gasperini S, Pretese R, Dionisi-Vici C, Maiorana A
Hepatic glycogen storage diseases type 0, VI and IX: description of an Italian cohort
Orphanet J Rare Dis 2022 Jul 19;17(1):285. doi: 10.1186/s13023-022-02431-5

Massese M, Tagliaferri F, Dionisi-Vici C, Maiorana A
Glycogen storage diseases with liver involvement: a literature review of GSD type 0, IV, VI, IX and XI
Orphanet J Rare Dis 2022 Jun 20;17(1):241. doi: 10.1186/s13023-022-02387-6

Pizzoferro M , Masselli G, Maiorana A, Casciani E, Sollaku S, Dionisi-Vici C, Spada M, Altini C, Villani MF, Rufini V, Gualdi G, Garganese MC
PET/CT in congenital hyperinsulinism: transforming patient's lives by molecular hybrid imaging
Am J Nucl Med Mol Imaging 2022; Apr 15;12(2):44-53. eCollection 2022. PMID: 35535120

Grünert S, Derks TGJ, Adrian K, Al-Thihli K, Ballhausen D, Bidiuk J, Bordugo A, Boyer M, Bratkovic D, Brunner-Krainz M, Burlina A, Chakrapani A, Corpeleijn W, Cozens A, Dawson C, Dhamko H, Djordjevic Milosevic M, Eiroa H, Finezilber Y, Fischinger Moura de Souza C, Garcia-Jiménez MC, Gasperini S, Haas D, Häberle J, Halligan R, Hiu Fung L, Hörbe-Blindt A, Horka LM, Huemer M, Kalkan Uçar S, Kecman B, Kilavuz S, Kriván G, Lindner M, Lüsebrink N, Makrilkakis K, Mei-Kwun Kwok A, Maier EM, Maiorana A, McCandless SE, Mitchell JJ, Mizumoto H, Mundy H, Ochoa C, Pierce K, Quijada Fraile P, Regier D, Rossi A, Santer S, Schuman HC, Piotr Sobieraj,Spenger J, Spiegel R, Stepien KM, Tal G, Zerjav Tanšek M, Drole Torkar A, Tchan M, Thyagu S, Schrier Vergano SA, Vucko E, Weinhold N,Zsidegh P, Wortmann SB
Efficacy and safety of Empagliflozin in Glycogen storage disease type IB: data from an international questionnaire
Genet Med 2022 Aug;24(8):1781-1788. doi: 10.1016/j.gim.2022.04.001

Maines E, Urru SA, Leonardi L, Fancellu E, Campomori A, Piccoli G, Maiorana A, Soffiati M, Franceschi R

Drug-induced hyperinsulinemic hypoglycemia: a review of the literature
Pediatr Endocrinol Rev 2022
In press

Maiorana A, Lepri FR, Novelli A, Dionisi-Vici C
Hypoglycaemia Metabolic Gene Panel Testing
Frontiers in Endocrinology, section Pediatric Endocrinology, Special Edition "The Problem of Childhood Hypoglycaemia"
Front Endocrinol (Lausanne). 2022 Mar 29;13:826167. doi:10.3389/fendo.2022.826167

Santoro L, Pjetraj D, Velmishi V, Campana C, Catassi C, Dionisi-Vici C, Maiorana A
A new phenotype of aldolase a deficiency in a 14 year-old boy with epilepsy and rhabdomyolysis - case report
Ital J Pediatr. 2022 Mar 4;48(1):39. doi: 10.1186/s13052-022-01228-3

Radenkovic S, Martinelli D, Zhang Y, Preston GJ, Maiorana A, Terracciano A, Dentici ML, Pisaneschi E, Novelli A, Ranatunga W, Ligezka AN, Ghesquière B, Deyle DR, Kozicz T, Pinto E Vairo F, Witters P, Morava E
TRAPPC9-CDG: A novel congenital disorder of glycosylation with dysmorphic features and intellectual disability.
Genet Med. 2022 Jan 15:S1098-3600(21)05471-X. doi:10.1016/j.gim.2021.12.012

Maiorana A, Gasperini S
Impatto della terapia con Empagliflozine su due pazienti con Glicogenosi 1b e Malattia infiammatoria cronica intestinale
La rivista italiana delle Malattie Rare 2021, year V, Oct 2021;3:21-23

Maiorana A, Caviglia S, Greco B, Alfieri P, Cumbo F, Campana C, Bernabei SM, Cusmai R, Mosca A, Dionisi-Vici C
Ketogenic diet as elective treatment in patients with drug-unresponsive hyperinsulinemic hypoglycemia caused by glucokinase mutations.
Orphanet J Rare Dis. 2021 Oct 11;16(1):424. doi: 10.1186/s13023-021-02045-3

Rossi A, Hoogeveen IJ, Lubout CMA, de Boer F, Fokkert-Wilts MJ, Rodenburg IL, van Dam E, Grünert SC, Martinelli D, Scarpa M; CONNECT MetabERN collaboration group, Dekker H, Te Boekhorst ST, van Spronsen FJ, Derks TGJ
A generic emergency protocol for patients with inborn errors of metabolism causing fasting intolerance: a retrospective, single-center study and the generation of www.emergencyprotocol.net.
J Inherit Metab Dis. 2021 Sep;44(5):1124-1135. doi: 10.1002/jimd.12386

Napolitano A, Longo D, Lucignani M, Pasquini L, Rossi-Espagnet MC, Lucignani G, Maiorana A, Elia D, De Liso P, Dionisi-Vici C, Cusmai R.
The Ketogenic Diet Increases In Vivo Glutathione Levels in Patients with Epilepsy.
Metabolites. 2020 Dec 10;10(12):504. doi: 10.3390/metabo10120504

Maiorana A, Sabia A, Corsetti T, Dionisi-Vici C
Safety of vaccines administration in hereditary fructose intolerance.
Orphanet J Rare Dis. 2020 Oct 1;15(1):274. doi: 10.1186/s13023-020-01552-z

Maines E, Catesini G, Boenzi S, Mosca A, Candusso M, Dello Strologo L, Martinelli D, Maiorana A, Liguori A, Olivieri G, Taurisano G, Piemonte F, Rizzo C, Spada M, Dionisi-Vici C
Plasma methylcitric acid and its correlations with other disease biomarkers: The impact in the follow up of patients with propionic and methylmalonic acidemia.
J Inherit Metab Dis. 2020 Nov;43(6):1173-1185. doi: 10.1002/jimd.12287

Rossi A, Hoogeveen IJ, Bastek VB, de Boer F, Montanari C, Meyer U, Maiorana A, Bordugo A, Dianin A, Campana C, Rigoldi M, Kishnani PS, Pendyal S, Strisciuglio P, Gasperini S, Parenti G, Parini R, Paci S, Melis D, Derks TGJ
Dietary lipids in glycogen storage disease type III: A systematic literature study, case studies, and future recommendations.
J Inherit Metab Dis. 2020 Jul;43(4):770-777. doi: 10.1002/jimd.12224

Ponzi E, Alesi V, Lepri FR, Genovese S, Loddo S, Mucciolo M, Novelli A, Dionisi-Vici C, Maiorana A
Uniparental isodisomy of chromosome 1 results in Glycogen Storage Disease type III with profound growth retardation
Mol Genet Genom Med 2019; 7(5):e634

Ponzi E & Maiorana A*, Lepri FR, Mucciolo M, Semeraro M, Taurisano R, Olivieri G, Novelli A, Dionisi-Vici C*
Persistent hypoglycemia in children: targeted gene panel improves the diagnosis of hypoglycemia due to Inborn Errors of Metabolism
J Pediatr 2018; 202:272-278.e4

Maiorana A, Dionisi-Vici C
Hyperinsulinemic hypoglycemia: clinical, molecular and therapeutical novelties.
J Inherit Metab Dis. 2017 40:531-542

Maiorana A, Cotugno G, Manganozzi L, Dionisi-Vici C
Ketogenic diets: pathophysiology and therapeutic implications
Giorn Gastr Epatol Nutr Ped 2015;VII:108-112, Pediatric Nutrition & Health and Food Science. File 3

Maiorana A, Manganozzi L, Barbetti F, Bernabei S, Gallo G, Cusmai R, Caviglia S, Dionisi-Vici C
Ketogenic diet in a patient with congenital hyperinsulinism: a novel approach to prevent brain damage.
Orphanet J Rare Dis. 2015 Sep 24;10(1):120. doi: 10.1186/s13023

Carrozzo R, Torraco A, Fiermonte G, Martinelli D, Di Nottia M, Rizza T, Vozza A, Verrigni D, Diodato D, Parisi G, Maiorana A, Rizzo C, Pierri CL, Zucano S, Piemonte F, Bertini E, Dionisi-Vici C.
Riboflavin responsive mitochondrial myopathy is a new phenotype of dihydrolipoamide dehydrogenase deficiency. The chaperon-like effect of vitamin B2.
Mitochondrion. 2014 Sep;18:49-57

Maiorana A, Malamisura M, Emma F, Boenzi S, Di Ciommo VM, Dionisi-Vici C.
Early effect of NTBC on renal tubular dysfunction in hereditary tyrosinemia type 1.
Mol Genet Metab. 2014 Nov;113(3):188-193

Mayorandan S, Meyer U, Gokcay G, Segarra N, de Baulny H, van Spronsen F, Zeman J, de Laet C, Spiekerkoetter U, Thimm E, Maiorana A, Dionisi-Vici C, Moeslinger D, Brunner-Krainz M, Lotz-Havla A, Cocho de Juan J, Couce Pico M, Santer R, Scholl-Bürgi S, Mandel H, Bliksrud Y, Freisinger P, Aldamiz-Echevarria L, Hochuli M, Gautschi M, Endig J, Jordan J, McKiernan P, Ernst S, Morlot S, Vogel A, Sander J, Das A.
Cross-sectional study of 168 patients with hepatorenal tyrosinaemia and implications for clinical practice.
Orphanet J Rare Dis. 2014 Aug 1;9(1):107

Taurisano R, Maiorana A, De Benedetti F, Dionisi-Vici C, Boldrini R, Deodato F
Wolman Disease associated with hemophagocytic lymphohistiocytosis: attempts for an explanation.
Eur J Pediatrics* 2014 *Oct;173(10):1391-1394

Maiorana A, Vergine G, Coletti V, Luciani M, Rizzo C, Emma F, Dionisi-Vici C
Acute thiamine deficiency and refeeding syndrome: similar findings but different pathogenesis.
Nutrition 2014, Jul-Aug;30(7-8):948-52

Maiorana A, Barbetti F, Boiani A, Rufini V, Pizzoferro M, Francalanci P, Faletra F, Nichols CG, Grimaldi C, de Ville de Goyet J, Rahier J, Henquin JC, Dionisi-Vici C
Focal congenital hyperinsulinism managed by medical treatment: a diagnostic algorithm based on molecular genetic screening.
Clin Endocrinol (Oxf). 2014 Nov;81(5):679-688

Faletra F, Athanasakis E, Morgan A, Biarnés X, Fornasier F, Parini R, Furlan F, Boiani A, Maiorana A, Dionisi-Vici C, Giordano L, Burlina A, Ventura A, Gasparini P.
Congenital hyperinsulinism: clinical and molecular analysis of a large Italian cohort.
Gene. 2013 May 25;521(1):160-165

Loechner KJ, Akrouh A, Kurata HT, Dionisi-Vici C, Maiorana A, Pizzoferro M, Rufini V, de Ville de Goyet J, Colombo C, Barbetti F, Koster JC, Nichols CG.
Congenital hyperinsulinism and glucose hypersensitivity in homozygous and heterozygous carriers of Kir6.2 (KCNJ11) mutation V290M mutation: KATP channel inactivation mechanism and clinical management.
Diabetes 2011, 60(1): 209-217

Maiorana A, Nobili V, Calandra S, Francalanci P, Bernabei S, El Hachem M,Monti L, Gennari F, Torre G, De Ville de Goyet J, Bartuli A.
Preemptive liver transplantation in a child with familial hypercholesterolemia.
Pediatr Transplant 2011,15(2): E25-29

Maiorana A, Fierabracci A and Cianfarani S
Isolation and characterization of omental adipose progenitor cells children: a potential tool to unravel the pathogenesis of metabolic syndrome.
Horm Res 2009; 72(6): 348-358

Maiorana A and Cianfarani S.
Impact of growth hormone therapy on adult height of children born small for gestational age: a systematic review of randomized controlled trials.
Pediatrics 2009, 124(3): e519-531

Maiorana A, Del Bianco C and Cianfarani S.
Adipose tissue: a metabolic regulator. Potential implications for the metabolic outcome of subjects born small for gestational age (SGA).
Rev Diabet Stud 2007; 4(3): 134-146

Ghirri P, Ladaki C, Bartali A, Scirè G, Vuerich M, Spadoni GL, Maiorana A, Geremia C, Boldrini A, Cianfarani S
Low birth weight for gestational age associates with reduced glucose concentrations at birth, infancy and childhood.
Horm Res, 2007, 67(3): 123-131

Cianfarani S, Martinez C, Maiorana A, Scirè G, Spadoni GL, Boemi S
Adiponectin levels are reduced in children born small for gestational age and are inversely related to postnatal catch-up growth.
J Clin Endocrinol Metab, 2004 Mar, 89(3): 1346-51

A. Maiorana, G. Cheng and R. Baserga
Role of Pescadillo in the Trasformation and Immortalization of Mammalian Cells
Oncogene, Sep 2004, 23(42): 7116-7124

M. Prisco, A. Maiorana, C. Guerzoni, G. Calin, B. Calabretta, R. Voit, I. Grummt and R. Baserga The role of Pescadillo and Upstream Binding Factor in the Proliferation and Differentation of Murine Myeloid Cells
Molecular and Cellular Biology, June 2004, 24(12): 5421-5433

X. Tu, A. Wu, A. Maiorana and R. Baserga
Subcellular localization of IRS-1 in cell proliferation and differentiation
Horm Metab Res, 2003 Nov-Dec,35(11-12): 734-9

Cianfarani S, Maiorana A, Geremia C, Scirè G, Spadoni GL, Germani D
Blood glucose concentrations are reduced in children born small for gestational age (SGA), and thyroid-stimulating hormone levels are increased in SGA with blunted postnatal catch-up growth.
J Clin Endocrinol Metab, 2003 Jun, 88(6): 2699-705

Cianfarani S, Geremia C, Germani D, Scirè G, Maiorana A, Boemi S
Insulin resistance and insulin-like growth factors in children with intrauterine growth retardation. Is catch-up growth a risk factor?
Horm Res, 2001, 55 Suppl 1: 7-10.

Book chapters

Cianfarani S, Geremia C, Puglianiello A, Maiorana A, Germani D. **Late effects of disturbed IGF signaling in congenital diseases. Intrauterine and post-natal growth retardation**. Lorini R, Maghnie M, D'Annunzio G, Loche S, Savage MO (eds): Congenital Endocrinopathies. New Insights into Endocrine Diseases and Diabetes. Endocr Dev. Basel, Karger, 2007, vol 11, pp 16-27

Maiorana A, Dionisi-Vici C. NTBC and correction of renal dysfunction. In**: Hereditary tyrosinemia: pathogenesis, screening and management**. Tanguay R, editor. Advances in Experimental Medicine and Biology Book 959, 1st ed. 2017 Edition, Kindle Edition. P. 93-100

Arnoux JB & Maiorana A*, Rio M, de Lonlay P*. Congenital hyperinsulinism. In: **Physician's guide to the diagnosis, treatment and follow-up of inherited metabolic diseases**. Blau N, Dionisi-Vici C, Ferreira CR, Vianey-Saban C, van Karnebeek CDM, editors. 2nd ed. 2022 Edition, Springer Nature Edition. P. 713-736.

E-Book

Maiorana A, Lepri FR, Novelli A, Dionisi-Vici C
Hypoglycaemia Metabolic Gene Panel Testing. In: **The problem of childhood hypoglycemia.** Banerjee, I., Mohnike, K., eds. (2023). Lausanne: Frontiers Media SA. doi: 10.3389/978-2-8325-2623-1, ISSN 1664-8714, ISBN 978-2-8325-2623-1.