CURRICULUM VITAE ET STUDIORUM

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Educational background:

1993-1998: Diploma Maturità Scientifica, Liceo Scientifico G. Vasco, Mondovì (**100/100**)

1999-2005: MD University of Torino (**110/110 Cum Laude**)

2006-2011: Residency in Pediatrics, training at the Regional Center for Metabolic Diseases, Department of Pediatrics, University of Torino (**70/70 Cum Laude**)

2009-2010: Visiting Scientist at the Biochemical Genetics Laboratory of the Division of Laboratory Genetics, Department of Laboratory Medicine and Pathology, Mayo Clinic College of Medicine, Rochester, Minnesota, U.S.A

2012-2016: Doctoral School in Life and Health Sciences, PhD in Biomedical Sciences and Oncology, Experimental Pediatrics, University of Torino

2017: Licensed Associate Professor in Pediatrics (Abilitazione Scientifica Nazionale)

Current position: Pediatrician and consultant for inborn errors of metabolism at the SC Pediatria, Azienda Ospedaliero-Universitaria Città della Salute e della Scienza di Torino, Ospedale Infantile Regina Margherita

International Publications

1. Kalantari S, Brezzi B, Bracciamà V, Barreca A, Nozza P, Vaisitti T, Amoroso A, Deaglio S, Manganaro M, **Porta F**, Spada M. Adult-onset CblC deficiency: a challenging diagnosis involving different adult clinical specialists. Orphanet J Rare Dis. 2022 Feb 2;17(1):33.
2. Masnada S, Sarret C, Antonello CE, Fadilah A, Krude H, Mura E, Mordekar S, Nicita F, Olivotto S, Orcesi S, **Porta F**, Remerand G, Siri B, Wilpert N, Amir-Yazdani P, Bertini E, Schuelke M, Bernard G, Boespflug-Tanguy O, Tonduti D. Movement disorders in MCT8 deficiency/Allan-Herndon-Dudley syndrome. Mol Genet Metab. 2022 Jan;135(1):109-113.
3. van Geest FS, Groeneweg S, van den Akker ELT, Bacos I, Barca D, van den Berg SAA, Bertini E, Brunner D, Brunetti-Pierri N, Cappa M, Cappuccio G, Chatterjee K, Chesover AD, Christian P, Coutant R, Craiu D, Crock P, Dewey C, Dica A, Dimitri P, Dubey R, Enderli A, Fairchild J, Gallichan J, Garibaldi LR, George B, Hackenberg A, Heinrich B, Huynh T, Kłosowska A, Lawson-Yuen A, Linder-Lucht M, Lyons G, Lora FM, Moran C, Müller KE, Paone L, Paul PG, Polak M, **Porta F**, Reinauer C, de Rijke YB, Seckold R, Menevşe TS, Simm P, Simon A, Spada M, Stoupa A, Szeifert L, Tonduti D, van Toor H, Turan S, Vanderniet J, de Waart M, van der Wal R, van der Walt A, van Wermeskerken AM, Wierzba J, Zibordi F, Zung A, Peeters RP, Visser WE. Long-term efficacy of T3 analogue Triac in children and adults with MCT8 deficiency: a real-life retrospective cohort study. J Clin Endocrinol Metab. 2021 Oct 22:dgab750.
4. Kuseyri Hübschmann O, Horvath G, Cortès-Saladelafont E, Yıldız Y, Mastrangelo M, Pons R, Friedman J, Mercimek-Andrews S, Wong S, Pearson T, Zafeiriou D, Kulhánek J, Kurian M, López-Laso E, Oppebøen M, Kılavuz S, Wassenberg T, Goez H, Scholl-Bürgi S, **Porta F**, Honzik T, Santer R, Burlina A, Sivri HS, Leuzzi V, Hoffmann G, Jeltsch K, Hübschmann D, Garbade S, iNTD Registry Study Group, Garcia-Cazorla A, and Opladen T. Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines. Nat Commun. 2021 Sep 20;12(1):5529.
5. van Wegberg AMJ, Trefz F, Gizewska M, Ahmed S, Chabraoui L, Zaki MS, Maillot F, van Spronsen FJ; Study Group on Missed PKU and Missed to Follow-Up. Undiagnosed Phenylketonuria Can Exist Everywhere: Results From an International Survey. J Pediatr. 2021 Aug 30:S0022-3476(21)00855-6.
6. Siri B, Varesio C, Freri E, Darra F, Gana S, Mei D, **Porta F**, Fontana E, Galati G, Solazzi R, Niceta M, Veggiotti P, Alfei E. CDKL5 deficiency disorder in males: Five new variants and review of the literature. Eur J Paediatr Neurol. 2021 Apr 30;33:9-20.

7. Tucci S, Wagner C, Grünert SC, Matysiak U, Weinhold N, Klein J, **Porta F**, Spada M, Bordugo A, Rodella G, Furlan F, Sajeva A, Menni F, Spiekerkoetter U. Genotype and residual enzyme activity in medium-chain acyl-CoA dehydrogenase (MCAD) deficiency: Are predictions possible? J Inherit Metab Dis. 2021 Jul;44(4):916-925.
8. **Porta F**, Siri B, Chiesa N, Ricci F, Nika L, Sciortino P, Spada M. SLC25A19 deficiency and bilateral striatal necrosis with polyneuropathy: a new case and review of the literature. J Pediatr Endocrinol Metab. 2020 Nov 19;34(2):261-266.
9. Kalantari S, **Porta F**, Vaisitti T, Deaglio S, Spada M. Unusual Presentation of Remethylation Disorders: A Case of Later Onset CblE Deficiency. Acta Scientific Paediatrics 2021; 4.1: 11-14.
10. **Porta F**, Giorda S, Ponzone A, Spada M. Tyrosine metabolism in health and disease: slow-release amino acids therapy improves tyrosine homeostasis in phenylketonuria. J J Pediatr Endocrinol Metab. 2020 Nov 19;33(12):1519-1523.
11. **Porta F**, Ponzone A, Spada M. Phenylalanine and tyrosine metabolism in DNAJC12 deficiency: A comparison between inherited hyperphenylalaninemias and healthy subjects. Eur J Paediatr Neurol. 2020 Sep;28:77-80.
12. Hillert A, Anikster Y, Belanger-Quintana A, Burlina A, Burton BK, Carducci C, Chiesa AE, Christodoulou J, Đorđević M, Desviat LR, Eliyahu A, Evers RAF, Fajkusova L, Feillet F, Bonfim-Freitas PE, Giżewska M, Gundorova P, Karall D, Kneller K, Kutsev SI, Leuzzi V, Levy HL, Lichter-Konecki U, Muntau AC, Namour F, Oltarzewski M, Paras A, Perez B, Polak E, Polyakov AV, **Porta F**, Rohrbach M, Scholl-Bürgi S, Spécola N, Stojiljković M, Shen N, Santana-da Silva LC, Skouma A, van Spronsen F, Stoppioni V, Thöny B, Trefz FK, Vockley J, Yu Y, Zschocke J, Hoffmann GF, Garbade SF, Blau N. The Genetic Landscape and Epidemiology of Phenylketonuria. Am J Hum Genet. 2020 Jul 1:S0002-9297(20)30194-4.
13. **Porta F**, Ponzone A, Spada M. Neonatal phenylalanine wash-out in phenylketonuria. Metab Brain Dis. 2020 Jul 13. doi: 10.1007/s11011-020-00602-6.
14. Manti F, Nardecchia F, Banderali G, Burlina A, Carducci C, Carducci C, Donati MA, Gueraldi D, Paci S, Pochiero F, **Porta F**, Ortolano R, Rovelli V, Schiaffino MC, Spada M, Blau N, Leuzzi V. Long-term clinical outcome of 6-pyruvoyl-tetrahydropterin synthase-deficient patients. Mol Genet Metab. Sep-Oct 2020;131(1-2):155-162.
15. Groeneweg S, van Geest FS, Abacı A, Alcantud A, Ambegaonkar GP, Armour CM, Bakhtiani P, Barca D, Bertini ES, van Beynum IM, Brunetti-Pierri N, Bugiani M, Cappa M, Cappuccio G, Castellotti B, Castiglioni C, Chatterjee K, de Coo IFM, Coutant R,

Craiu D, Crock P, DeGoede C, Demir K, Dica A, Dimitri P, Dolcetta-Capuzzo A, Dremmen MHG, Dubey R, Enderli A, Fairchild J, Gallichan J, George B, Gevers EF, Hackenberg A, Halász Z, Heinrich B, Huynh T, Kłosowska A, van der Knaap MS, van der Knoop MM, Konrad D, Koolen DA, Krude H, Lawson-Yuen A, Lebl J, Linder-Lucht M, Lorea CF, Lourenço CM, Lunsing RJ, Lyons G, Malikova J, Mancilla EE, McGowan A, Mericq V, Lora FM, Moran C, Müller KE, Oliver-Petit I, Paone L, Paul PG, Polak M, **Porta F**, Poswar FO, Reinauer C, Rozenkova K, Menevse TS, Simm P, Simon A, Singh Y, Spada M, van der Spek J, Stals MAM, Stoupa A, Subramanian GM, Tonduti D, Turan S, den Uil CA, Vanderniet J, van der Walt A, Wémeau JL, Wierzba J, de Wit MY, Wolf NI, Wurm M, Zibordi F, Zung A, Zwaveling-Soonawala N, Visser WE. Disease characteristics of MCT8 deficiency: an international, retrospective, multicentre cohort study. Lancet Diabetes Endocrinol. 2020 Jul;8(7):594-605.

16. Opladen T, López-Laso E, Cortès-Saladelafont E, Pearson TS, Sivri HS, Yildiz Y, Assmann B, Kurian MA, Leuzzi V, Heales S, Pope S, **Porta F**, García-Cazorla A, Honzík T, Pons R, Regal L, Goez H, Artuch R, Hoffmann GF, Horvath G, Thöny B, Scholl-Bürgi S, Burlina A, Verbeek MM, Mastrangelo M, Friedman J, Wassenberg T, Jeltsch K, Kulhánek J, Kuseyri Hübschmann O; International Working Group on Neurotransmitter related Disorders (iNTD). Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH(4)) deficiencies. Orphanet J Rare Dis. 2020 May 26;15(1):126.

17. Spada M, **Porta F**, Righi D, Tandoi F, Ferrero I, Fagioli F, Herrera MB, Calvo PL, Biamino E, Gunetti M, Contursi C, Lauritano C, Conio A, Amoroso A, Salizzoni M, Silengo L, Camussi G, Romagnoli R. Intrahepatic administration of human liver stem cells in infants with inherited neonatal-onset hyperammonemia: a Phase I study. Stem Cell Rev Rep. 2020 Feb;16(1):186-197.

18. **Porta F**, Romagnoli R, Busso M, Tandoi F, Spada M. Differential intraoperative effect of liver transplant in different inborn errors of metabolism. J Pediatr Gastroenterol Nutr. 2019 Aug;69(2):160-162.

19. **Porta F**, Chiesa N, Martinelli D, Spada M. Clinical, biochemical, and molecular spectrum of short/branched-chain acyl-CoA dehydrogenase deficiency: two new cases and review of literature. J Pediatr Endocrinol Metab. 2019 Feb 25;32(2):101-108.

20. **Porta F**, Busso M, Giorda S, Spada M. Columbus' egg: a practical approach to nutritional management of maple syrup urine disease. J Pediatr Endocrinol Metab. 2019 Jan 28;32(1):83-84.

21. Porta F. The dream of bedside complex biochemical diagnoses comes true. Science 2018 Nov; sciencemag.org/content/361/6407/1122/tab-e-letters.

22. Spada M, Pagliardini V, Ricci F, Biamino E, Mongini T, **Porta F**. Early higher dosage of alglucosidase alpha in classic Pompe disease. J Pediatr Endocrinol Metab. 2018 Dec 19;31(12):1343-1347.

23. Porta F, Peruzzi L, Bonaudo R, Pieretti S, Busso M, Cocchi E, Conio A, Pagliardini V, Spada M. Differential response to renal replacement therapy in neonatal-onset inborn errors of metabolism. Nephrology (Carlton). 2018 Oct;23(10):957-961.

24. Ricci F, Brusa C, Rossi F, Rolle E, Placentino V, Berardinelli A, Pagliardini V, **Porta F**, Spada M, Mongini T. Functional assessment tools in children with Pompe disease: a pilot comparative study to identify suitable outcome measures for the standard of care. Eur J Paediatr Neurol. 2018 In press.

25. Menduti G, Biamino E, Vittorini R, Vesco S, Puccinelli MP, **Porta F**, Capo C, Leo S, Ciminelli BM, Iacovelli F, Spada M, Falconi M, Malaspina P, Rossi L. Succinic semialdehyde dehydrogenase deficiency: the combination of a novel ALDH5A1 gene mutation and a missense SNP strongly affects SSADH enzyme activity and stability. Mol Genet Metab. 2018 Jul;124(3):210-215

26. Quarello P, Spada M, **Porta F**, Vassallo E, Timeus F, Fagioli F. Hematopoietic stem cell transplantation in Niemann-Pick disease type B monitored by chitotriosidase activity. Pediatr Blood Cancer. 2018 Feb;65(2).

27. Porta F, Pagliardini V, Celestino I, Pavanello E, Pagliardini S, Guardamagna O, Ponzone A, Spada M. Neonatal screening for biotinidase deficiency: a 30-year single center experience. Mol Genet Metab Rep. 2017 Sep 20;13:80-82.

28. Porta F, Spada M, Ponzone A. Early screening for tetrahydrobiopterin responsiveness in phenylketonuria. Pediatrics. 2017 Aug;140(2). pii: e20161591. doi: 10.1542/peds.2016-1591.

29. Calvo PL, Spada M, Rabbone I, Pinon M, **Porta F**, Cisarò F, Reggiani S, Cefalù AB, Sturiale L, Garozzo D, Lefeber DJ, Jaeken J. An Unexplained Congenital Disorder of Glycosylation-II in a Child with Neurohepatic Involvement, Hypercholesterolemia and Hypoceruloplasminemia. JIMD Rep. 2017 Jun 23. doi: 10.1007/8904_2017_35.

30. Spada M, Kasper D, Pagliardini V, Biamino E, Giachero S, **Porta F**. Metabolic progression to clinical phenotype in classic Fabry disease. Ital J Pediatr. 2017 Jan 3;43(1):1.

31. **Porta F**, Ponzone A, Spada M. Long-term safety and effectiveness of pramipexole in tetrahydrobiopterin deficiency. Eur J Paediatr Neurol. 2016 Nov;20(6):839-842.

32. Romagnolo A, Merola A, **Porta F**, Spada M, Lopiano L, Rizzone MG. Transdermal rotigotine in dihydropteridine reductase deficiency. J Neurol Sci. 2016 Aug 15;367:237-8.

33. Jansen JC, Timal S, van Scherpenzeel M, Michelakakis H, Vicogne D, Ashikov A, Moraitou M, Hoischen A, Huijben K, Steenbergen G, van den Boogert M, **Porta F**, Calvo PL, Mavrikou M, Cenacchi G, van den Bogaart G, Salomon J, Holleboom A, Rodenburg R, Drenth J, Huynen M, Wevers R, Morava E, Foulquier F, Veltman J, Lefeber DJ. TMEM199 deficiency, a glycosylation disorder with elevated aminotransferases, alkaline phosphatase and hypercholesterolemia. Am J Hum Genet. 2016 Feb 4;98(2):322-30.

34. **Porta F**, Ponzone A, Spada M. Target prolactin range in treatment of tetrahydrobiopterin deficiency. J Pediatr. 2016 Jan;168:236-239.

35. **Porta F**, Mussa A, Baldassarre G, Perduca V, Farina D, Spada M, Ponzone A. Genealogy of breastfeeding. Eur J Pediatr. 2016 Jan;175(1):105-12.

36. Spada M, Calvo PL, Brunati A, Peruzzi L, dell'Olio D, Romagnoli R, **Porta F**. Liver transplantation in severe methylmalonic acidemia: the sooner, the better. J Pediatr. 2015 Nov;167(5):1173.

37. Spada M, Calvo PL, Brunati A, Peruzzi L, dell'Olio D, Romagnoli R, **Porta F**. Early liver transplantation for neonatal-onset methylmalonic acidemia. Pediatrics. 2015 Jul;136(1):e252-6.

38. **Porta F**, Ponzone A, Spada M. Short prolactin profile for monitoring treatment in BH4 deficiency. Eur J Paediatr Neurol. 2015 May;19(3):360-3.

39. **Porta F**, Pagliardini V, Roasio L, Biamino E, Spada M. Playing competitive basketball in face of later-onset Pompe disease. Muscle Nerve. 2015 Feb;51(2):302-3.

40. **Porta F**, Pagliardini S, Pagliardini V, Ponzone A, Spada M. Newborn screening for galactosemia: a 30 years single Centre experience. World J Pediatr. 2015 May;11(2):160-4.

41. Mussa A, Baldassarre G, **Porta F**. Fracture odds and body mass index in children. J Pediatr. 2014. pii: S0022-3476(14)00747-1.

42. **Porta F**, Volpe V. Weaning the normal infant. Acta Paediatr. 2014 Apr;103(4):353-5.

43. **Porta F**, Volpe V. Feeding the normal newborn: whose art is it? Acta Paediatr. 2013 Jun 3. doi: 10.1111/apa.12309.

44. Santarelli F, Cassanello M, Enea A, Poma F, D'Onofrio V, Guala G, Garrone G, Puccinelli P, Caruso U, **Porta F**, Spada M. A neonatal case of 3-hydroxy-3-methylglutaric-coenzyme A lyase deficiency. Ital J Pediatr. 2013 May 24;39(1):33

45. Spada M, **Porta F**, Vercelli L, Pagliardini V, Chiadò-Piat L, Boffi P, Pagliardini S, Remiche G, Ronchi D, Comi G, Mongini T. Screening for later-onset Pompe's disease in patients with paucisymptomatic hyperCKemia. Mol Genet Metab. 2013 Mar 14. doi:pii: S1096-7192(13)00091-7. 10.1016/j.ymgme.2013.03.002.

46. Spada M, Enea A, Morrone A, Fea A, **Porta F**. Cornea verticillata and Fabry disease. J Pediatr. 2013. pii: S0022-3476(13)00289-8. doi: 10.1016/j.jpeds.2013.03.013

47. Porta F, Mussa A, Spada M. Later effects of metabolic control in phenylketonuria. Mol Genet Metab. 2013 May;109(1):118

48. Spada M, Garelli D, Riggi C, Pagliardini V, Lualdi S, Filocamo M, **Porta F**. Spontaneous regression of hypertrophic cardiomyopathy in an infant with Pompe's disease. Mol Genet Metab. 2012 Dec;107(4):763.

49. Porta F, Pagliardini V, Barbera C, Calvo P, Pagliardini S, Lualdi S, Filocamo M, Spada M. Neonatal chitotriosidase activity is not predictive for Niemann-Pick disease type A/B: implications for newborn screening for lysosomal storage disorders. Mol Genet Metab. 2013 Jan;108(1):106.

50. Lovera C, **Porta F**, Caciotti A, Catarzi S, Cassanello M, Caruso U, Gallina MR, Morrone A, Spada M. Sudden unexpected infant death (SUDI) in a newborn due to medium chain acyl CoA dehydrogenase (MCAD) deficiency with an unusual severe genotype. Ital J Pediatr. 2012 Oct 24;38(1):59.

51. Porta F, Mussa A. Bed-sharing is bad-sharing. Pediatrics. 2012; http://pediatrics.aappublications.org/content/129/4/630/reply#pediatrics_el_53637.

52. Porta F, Mussa A, Concolino D, Spada M, Ponzone A. Dopamine agonists in dihydropteridine reductase deficiency. Mol Genet Metab. 2012;105(4):582-4.

53. Mussa A, Camilla R, Monticone S, **Porta F**, Tessaris D, Verna F, Mulatero P, Einaudi S. Polyuric-polydipsic syndrome in a pediatric case of non-glucocordicoid remediable Familial Hyperaldosteronism. Endocr J. 2012 Jun 30;59(6):497-502.

54. Mussa A, **Porta F**, Baldassarre G, Tuli G, de Terlizzi F, Matarazzo P, Einaudi S, Lala R, Corrias A. Phalangeal quantitative ultrasound in 1,719 children and adolescents with bone disorders. Osteoporos Int. 2012 Jul;23(7):1987-98.

55. Mussa A, **Porta F**, Baldassarre G, Corrias A. Determinants of thyrotropin rise in congenital hypothyroidism. J Pediatr. 2011; 159(6):1050.

56. Porta F, Mussa A, Pagliardini S, Dotta A, Pagliardini V, Spada M. Lysosomal enzyme activities in phenylketonuria. Mol Genet Metab. 2011;102(4):508.

57. Porta F, Mussa A, Garelli D, Spada M. Phenotyping and treatment of phenylketonuria. Lancet 2011;5;377(9764):465.

58. Porta F, Spada M, Garelli D, Mussa A, Ponzone A. Tetrahydrobiopterin and phenylketonuria. J Pediatr. 2011;158(5):864.

59. Porta F, Roato I, Mussa A, D'amico L, Fiore L, Spada M, Garelli D, Ferracini R. Bone impairment in phenylketonuria is characterized by circulating osteoclast precursors and activated T cell increase. PLoS One. 2010;30;5(11):e14167.

60. Porta F, Mussa A, Zanin A, Greggio NA, Burlina A, Spada M. Impact of metabolic control on bone quality in phenylketonuria and mild hyperphenylalaninemia. J Pediatr Gastroenterol Nutr. 2011;52(3):345-50.

61. Mussa A, Bertorello N, **Porta F**, Galletto C, Nicolosi MG, Manicone R, Corrias A, Fagioli F. Fractures and skeletal complications should be the gold standard for validation of methods for bone appraisal in pediatrics. Bone. 2010; 47(4):837-8. http://dx.doi.org/10.1016/j.bone.2010.07.003

62. Mussa A, Repici M, Fiore L, Tuli G, **Porta F**, Matarazzo P. Bone quantitative ultrasound in congenital and acquired childhood multiple pituitary failure. Ultrasound Med Biol. 2010;36(5):726-32.

63. Mussa A, Chiesa N, **Porta F**, Baldassarre G, Silengo M, Ferrero GB. The overlap between Sotos and Beckwitt-Wiedemann syndromes. J Pediatr. 2010;156(6):1035-6.

64. Porta F, Mussa A, Ponzone A. Breastfeeding effects on newborn screening. J Pediatr. 2010;156(6):1033.

65. Mussa A, Bertorello N, **Porta F**, Galletto C, Nicolosi MG, Manicone R, Corrias A, Fagioli F. Prospective bone ultrasound patterns during childhood acute lymphoblastic leukemia treatment. Bone. 2010;46:1016-20.

66. Leuzzi V, Carducci C, Carducci C, Pozzessere S, Burlina A, Cerone R, Concolino D, Donati A, Fiori L, Meli C, Ponzone A, **Porta F**, Strisciuglio P, Antoniozzi I, Blau N. Phenotypic variability, neurological outcome and genetics background of 6-pyruvoyl-tetrahydropterin synthase deficiency. Clin Genet. 2010;77(3):249-57.

67. Ponzone A, **Porta F**, Mussa A, Alluto A, Ferraris S, Spada M. Unresponsiveness to tetrahydrobiopterin of phenylalanine hydroxylase deficiency. Metabolism. 2010;59:645-52.

68. Ferrero GB, **Porta F**, Biamino E, Mussa A, Garelli E, Chiappe F, Veltri A, Cirillo Silengo M, Gennari F. Remittent hyperammonemia in congenital portosystemic shunt. Eur J Pediatr. 2009:169:369-72.

69. Ponzone A, Mussa A, **Porta F**. In response to van Spronsen et al (2009) Phenylalanine tolerance can already reliably be assessed at the age of 2 years in patients with PKU. J Inherit Metab Dis 32: 27-31. Inherit Metab Dis. 2009;32:584.

70. Porta F, Mussa A, Concolino D, Spada M, Ponzone A. Dopamine agonists in 6-pyruvoyl tetrahydropterin synthase deficiency. Neurology. 2009;73:633-7.

71. Porta F, Roato I, Mussa A, Repici M, Gorassini E, Spada M, Ferracini R. Increased spontaneous osteoclastogenesis from peripheral blood mononuclear cells in phenylketonuria. J Inherit Metab Dis. 2008; DOI: 10.1007/s10545-008-0907-9.

72. Porta F, Spada M, Lala R, Mussa A. Phalangeal quantitative ultrasound in children with phenylketonuria: a pilot study. Ultrasound Med Biol. 2008;34:1049-52.

73. Ponzone A, Spada M, Roasio L, **Porta F**, Mussa A, Ferraris S. Impact of neonatal protein metabolism and nutrition on screening for phenylketonuria. J Pediatr Gastroenterol Nutr. 2008;46:561-9.

74. Ferrero GB, Pagliardini S, Veljkovic A, **Porta F**, Bena C, Tardivo I, Restagno G, Silengo MC, Bignamini E. In vivo specific reduction of arylsulfatase B enzymatic activity in children with cystic fibrosis. Mol Genet Metab. 2008;94:139.

75. Jäggi L, Zurflüh MR, Schuler A, Ponzone A, **Porta F**, Fiori L, Giovannini M, Santer R, Hoffmann GF, Ibel H, Wendel U, Ballhausen D, Baumgartner MR, Blau N. Outcome and long-term follow-up of 36 patients with tetrahydrobiopterin deficiency. Mol Genet Metab. 2008;93:295-305.

76. Mussa A, **Porta F**, Gianoglio B, Gaido M, Nicolosi MG, De Terlizzi F, de Sanctis C, Coppo R. Bone alterations in children and young adults with renal transplant assessed by phalangeal quantitative ultrasound. Am J Kidney Dis. 2007;50:441-9.

Selected abstracts

1. Raymond K, **Porta F**, Turgeon C, Magera MJ, Liedtke K, Gavrilov D, Oglesbee D, Tortorelli S, Rinaldo P, Matern D. Simultaneous transferrin and apolipoprotein cIII glycoforms analysis by online immuno-affinity chromatography electrospray ionization mass spectrometry. J Inherit Metab Dis 2013 (Supplement 2): 238.

2. Spada M, Vercelli L, Mongini T, **Porta F.** Symptomatic carriers of Pompe disease revealed by selective screening in hyperCKemia. J Inherit Metab Dis 2013 (Supplement 1).
3. **Porta F**, Turgeon C, Tortorelli S, Gavrilov D, Oglesbee D, Rinaldo P, Matern D, Raymond K. Advances in the diagnosis of congenital disorders of glycosylation. J Inherit Metab Dis 2010 (Supplement 1).
4. **Porta F**, Tortorelli S, Gavrilov D, Oglesbee D, Raymond K, Matern D, Rinaldo P. Expanded newborn screening in the NICU. J Inherit Metab Dis 2010 (Supplement 1).
5. Garelli D, Pagliardini V, Ignaccolo MG, Mussa A, Porta F, Meli C, Ponzone A, Spada M. Screening for BH4 responsiveness in PKU: results from a quantitative method. J Inherit Metab Dis 2010 (Supplement 1).
6. Mussa A, Roato I, Spada M, Ferracini R, **Porta F.** Increased spontaneous osteoclastogenesis in PKU. Mol Genet Metab 2010 99: 227.
7. **Porta F**, Mussa A, Concolino D, Spada M, Ponzone A. Pramipexole in dihydropteridine reductase deficiency. Mol Genet Metab 2010 99: 230
8. Leuzzi V, Burlina A, Cerone R, Concolino D, Donati MA, Fiori L, Ponzone A, **Porta F**, Strisciuglio P, Carducci C, Carducci C, Vagnoni C, Pozzessere S, Antoniozzi I. The phenotypic variability in 6-pyruvoiltetrahydrobiopterin synthase (PTPS) deficiency (PTPSD). Clinical presentation and outcome of the italian patients. J Inherit Metab Dis 2008 31 (Supplement 1): 76
9. Jaggi L, Zulfluh MR, Schuler A, Ponzone A, **Porta F**, Salvatici E, Giovannini M, Santer R, Hoffmann GF, Ibel H, Wendel U, Ballhausen D, Baumgartner MR, Blau N. Long-term follow-up and out come of patients with tetrahydrobiopterin deficiency. J Inherit Metab Dis 2008 31 (Supplement 1): 83
10. **Porta F**, Roato I, Spada M, Ferracini R, Mussa A. Increased spontaneous osteoclastogenesis in phenylketonuria. J Inherit Metab Dis 2008 31 (Supplement 1): 74
11. **Porta F**, Mussa A, Concolino D, Spada M, Ponzone A. Pramipexole in tetrahydrobiopterin deficiency. J Inherit Metab Dis 2008 31 (Supplement 1): 137
12. **Porta F**, Mussa A, Spada M, Ponzone A. Ineffectiveness of tetrahydrobiopterin in phenylalanine hydroxylase deficiency. J Inherit Metab Dis 2008 31 (Supplement 1): 80
13. **Porta F**, Mussa A, Spada M, Ponzone A. Dopamine agonists in tetrahydrobiopterin deficiency. J Inherit Metab Dis 2007 30 (Supplement 1): 138
14. **Porta F**, Mussa A, Ferraris S, Spada M, Ponzone A. "Responsiveness" and unresponsiveness to BH4 of PAH deficiency. J Inherit Metab Dis 2007 30 (Supplement 1): 16

15. **Porta F,** Spada M, Baldassarre G, De Sanctis C, Mussa A. Bone condition at phalangeal quantitative ultrasound in phenylketonuria. J Inherit Metab Dis 2006: 98
16. **Porta F**, Alluto A, Mussa A, Spada M, Ponzone A. A comparison between simple and combined loading test in phenylketonuria. J Inherit Metab Dis 2006: 70
17. Mussa A, **Porta F**, Gianoglio B, Gaido M, Camilla R, De Terlizzi F, Amore A. Bone condition in pediatric renal transplant recipient (PRTR): evaluation with quantitative ultrasound. Pediatr Nephrol 2006 21: 1505.

Book Chapters

Spada M, **Porta F**. Urgenze metaboliche. In: Pediatria d'urgenza, Urbino AF ed. Biomedia 2020

Porta F, Dionisi Vici C. Malattie metaboliche. Novità in tema di malattie metaboliche: nuove malattie, ricerche nel campo della fisiopatologia, nuove terapie. Prospettive in Pediatria 2020; 50: 197.

Mussa A, **Porta F**, Hoffmann GF, Sarafoglou K. Emergency Assessment and Management of Suspected Inborn Errors of Metabolism and Endocrine Disorders. In: Pediatric Endocrinology and Inborn Errors of Metabolism, Second Edition. Sarafoglou K, Hoffman G, Roth K, eds. Mcgraw Hill, 2017.

Porta F. Medicina metabolomica. Lessico del XXI secolo, Treccani, 2013.

Porta F. Galattosemia. Malattie Metaboliche Ereditarie. Collana monografica SIP, SIMMESN, SIMGePed. In press.

National and international oral communications

1. Porta F. Homocysteine as a marker of metabolic diseases. **National Intersocietary SIMMESN/SIBIOC Webinar Congress, 2021, Italy.**
2. Porta F. **Chairman, Italian Society for the Study of Inborn Errors of Metabolism (SIMMESN) Webinar Congress, 2020, Italy.**

3. Porta F. **Inauguration Doctorate School, University of Torino. Cavallerizza Reale, November 4, 2019, Torino.**
4. Porta F. Liver transplantation in inborn errors of metabolism. **Round table, Italian society for the Study of Inborn Errors of Metabolism Congress, 2019, Torino, Italy.**
5. Porta F. Neonatal phenylalanine wash-out in phenylketonuria. Oral communication, **Italian society for the Study of Inborn Errors of Metabolism Congress, 2019, Torino, Italy.**
6. Porta F. Pregnancy and phenylketonuria. **Italian society for the Study of Inborn Errors of Metabolism Congress, 2019, Torino, Italy.**
7. Porta F, Martinelli D. Neurotransmitters and disorders of neurotransmission. **Plenary session, Italian society for the Study of Inborn Errors of Metabolism Congress, 2019, Torino, Italy.**
8. Porta F. Tyrosine metabolism in health and disease. **Italian society for the Study of Inborn Errors of Metabolism Congress, 2019, Torino, Italy.**
9. Porta F. Management of PKU from birth to childhood. **Lecture at the meeting "Management of PKU", 2019, Firenze, Italy.**
10. Porta F. Intraoperative metabolic profile after liver transplantation in methylmalonic acidemia, argininosuccinic aciduria, and maple syrup urine disease. **European Metabolic Group (EMG) meeting, 2019, Lyon, France.**
11. Porta F. Early metabolic effect of liver transplantation in methylmalonic acidemia, argininosuccinic aciduria, and maple syrup urine disease. **Italian society for the Study of Inborn Errors of Metabolism Congress, 2018, Catania, Italy.**
12. Porta F. DNAJC12: quantitative and comparative metabolic evaluation of inherited hyperphenylalaninemias. **Italian society for the Study of Inborn Errors of Metabolism Congress, 2018, Catania, Italy.**
13. Porta F. Long-term safety and effectiveness of pramipexole in tetrahydrobiopterin deficiency. **Italian society of Pediatrics Congress, 2018, Torino, Italy.**
14. Porta F. Management of Phenylketonuria for Optimal Outcome: From Theory to Real Practice. **Seminar, 2018, Genova, Italy.**
15. Porta F. Liver transplantation in urea cycle defects: the Italian experience. **Round table at the meeting "Management of urea cycle defects", 2018, Frascati, Italy.**
16. Porta F. Hematological manifestations of inborn errors of metabolism. **Lecture at the national Congress "Rete ematologica piemontese", 2018, Orbassano, Italy.**
17. Porta F. Expanded newborn screening. **Lecture at the national Congress "Prevenzione in pediatria", 2018, Torino, Italy.**

18. Porta F. **Chairman at Italian society for the Study of Inborn Errors of Metabolism Congress, 2017, Roma, Italy.**
19. Porta F. Urea cycle disorders: barriers to drug adherence in the current therapies. **Lecture at the Italian Society for the Study of Inborn Errors of Metabolism Congress, 2017, Roma, Italy.**
20. Porta F. Malattie metaboliche ereditarie. **Communication at the Congress "Malattie rare in età pediatrica: ricerca, farmaci orfani e reti europee di riferimento", 2017, Napoli, Italy.**
21. Porta F. Il ruolo del pediatra nella prevenzione dei disturbi visivi. **Lecture at the Congress "La rete oculistica metropolitana", 2017, Torino, Italy.**
22. Porta F. Organic acidurias. **Chairman at Society for the Study of Inborn Errors of Metabolism Symposium, 2016, Roma, Italy.**
23. Porta F. Phenylketonuria in adulthood. **Lecture at Phenylketonuria (PKU) Advisory Board, 2016, Barcelona, Spain.**
24. Porta F. Prolactin for monitoring inherited dopamine deficiency. **Lecture at International Conference on Aromatic Amino Acid Decarboxylase Deficiency, 2016, London, UK.**
25. Porta F. Advances in inherited hyperphenylalaninemias. **Seminar at University Federico II, 2016, Napoli, Italy.**
26. Porta F. Galactosemia. **Lecture at the Italian Society for the Study of Inborn Errors of Metabolism Congress, 2015, Firenze, Italy.**
27. Porta F. Monitoring and treatment of BH4 deficiency. **Communication at the Italian Society for the Study of Inborn Errors of Metabolism Congress, 2015, Firenze, Italy.**
28. Porta F. Fabry disease. **Lecture at the Congress "Clinical practice in Pediatrics", 2015, Milano, Italy.**
29. Porta F. Galactosemia. **Lecture at the Italian Society for the Study of Inborn Errors of Metabolism Congress, 2014, Milano, Italy.**
30. Porta F. Dopamine agonists in BH4 deficiency. **Communication at the Italian Society for the Study of Inborn Errors of Metabolism Congress, 2013, Napoli, Italy.**
31. Porta F. Congenital disorders of glycosylation and cutis laxa. **Case conference at Mayo Clinic College of Medicine, 2011, Rochester, MN, USA**.
32. Porta F**.** Mucopolysaccharidosis: Hurler, Hunter, and Maroteaux-Lamy. **Case conference at Mayo Clinic College of Medicine, 2010, Rochester, MN, USA.**

33. Porta F. Ineffectiveness of tetrahydrobiopterin in phenylalanine hydroxylase deficiency. **Communication at the International Congress of Inborn Errors of Metabolism**, **2008, Lisboa, Portugal.**

34. Porta F**.** Pramipexole in tetrahydrobiopterin deficiency. **Communication at the International Congress of Inborn Errors of Metabolism, 2008, Lisboa, Portugal**.

35. Porta F. In vivo specific reduction of ARSB in patients with cystic fibrosis. **Communication at the Italian Society for the Study of Inborn Errors of Metabolism Congress, 2007, Roma, Italy.**

36. Porta F. A comparison between simple and combined loading test in phenylketonuria. **Communication at the International Congress of Inborn Errors of Metabolism, 2006, Chiba, Japan.**

37. Porta F. Hematological and neurological manifestations of vitamin B12 deficiency. **Communication at the National Congress of the Italian Society of Pediatrics, 2006, Savigliano, Italy.**

38. Porta F. Rhabdomyolisis and CPTII deficiency. **Communication at the Italian Society for the Study of Inborn Errors of Metabolism Congress, 2005, Pollenzo, Italy.**

International scientific group participation

Participation to the **International Working Group on Neurotransmitter Related Disorders (iNTD)**

International editorial committee

2018: Associate Editorial Board Member of **Current Gene Therapy (IF: 2.78)**

2012: Editor of the **International Journal of Pediatrics**

Awards and honours

"Cultore della materia" in Pediatrics (MED38) at the University of Torino

Elected advisory member of the **Società Italiana Per lo Studio delle Malattie Metaboliche Ereditarie e lo Screening Neonatale (SIMMESN) (2018-2021)**

Member of the **Società Italiana Per lo Studio delle Malattie Metaboliche Ereditarie e lo Screening Neonatale (SIMMESN)**

Finalist at **Premio Giovedì Scienza 2014** for young investigators with the research "Expanded newborn screening".

Finalist at **Premio Giovedì Scienza 2013** for young investigators with the research "Genealogy of breastfeeding".

Specific scientific experiences

Visiting Scientist at the **Biochemical Genetics Laboratory of the Division of Laboratory Genetics, Department of Laboratory Medicine and Pathology, Mayo Clinic College of Medicine, Rochester, Minnesota, U.S.A**

International rewards

- Co-starring in the Netflix medical docuseries "Diagnosis", 2019
- Invitation at the European Metabolic Group (EMG) meeting, 2018, Hamburg, Germany
- Invitation at the International Young Metabolicians Days, 2018, Vienna, Austria
- Invitation at the European Metabolic Group (EMG) meeting, 2017, Zagreb, Croatia
- Cochrane Membership
- Chairman at Society for the Study of Inborn Errors of Metabolism Symposium (SSIEM), 2016, Roma, Italy.

Teaching experience

- Teacher at the Master (II level) "Malattie metaboliche ereditarie e screening neonatale", Bologna, 2020-2021.

- Teacher at the Ph.D. Program in Biomedical Sciences and Oncology, University of Torino