Curriculum Vitae

Marcella Devoto

October 2020

Education

1983	Laurea in Statistics and Demography (110/110 cum laude) University of Bologna, Italy
1986	M.Sc. in Applied Statistics, Linacre College, University of Oxford, UK

Postgraduate Training and Fellowship Appointment

1984-85	Post-graduate fellow, Genetics Laboratory, Clinical Neurology, University of Bologna, Italy
1985-89	Post-graduate fellow, Laboratory of Molecular Genetics, Istituto G. Gaslini, Genoa, Italy

Faculty Appointments

1993-04	Assistant Professor of Human Genetics, University of Genoa, School of Medicine, Genoa, Italy
1995-97	Assistant Professor of Clinical Public Health in Psychiatry, Department of Psychiatry, Columbia University, New York, NY
2000-05	Head, Genetic Epidemiology Research Laboratory Nemours Children's Clinic - Wilmington, Wilmington, DE
2000-05	Research Associate Professor of Pediatrics, Thomas Jefferson University, Philadelphia, PA
2005-	Associate Professor of Medical Genetics, Università Sapienza, Rome, Italy
2006-12	Associate Professor of Pediatrics at Children's Hospital of Philadelphia (primary), Division of Human Genetics, Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2006-12	Associate Professor of Epidemiology (secondary), Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2012-21	Professor of Pediatrics at Children's Hospital of Philadelphia (primary), Division of Human Genetics, Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2012-21	Professor of Epidemiology (secondary), Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA

2015-21	Professor of Genetics (secondary), Department of Genetics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA

Other Appointments

1989-92	Research Scientist, Istituto G.Gaslini, Genoa, Italy
1992-93	Associate Research Scientist, New York State Psychiatric Institute, New York, NY
1993-95	Associate Research Scientist, Department of Psychiatry, Columbia University, New York, NY
1997-98	Associate Research Scientist, Laboratory of Statistical Genetics, Rockefeller University, New York, NY
2002-06	Adjunct Member, College of Graduate Studies, Thomas Jefferson University, Philadelphia, PA
2006-21	Research Scientist, Children's Hospital of Philadelphia Research Institute, Philadelphia, PA
2006-21	Senior Scholar, Center for Clinical Epidemiology and Biostatistics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2006	Founding Member, Center for Biomedical Informatics, Children's Hospital of Philadelphia, Philadelphia, PA
2006-21	Member, Graduate Group in Epidemiology and Biostatistics, Biomedical Graduate Studies, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2007-15	Member, Penn Center for Musculoskeletal Disorders, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2011-21	Member, Graduate Group in Genomics and Computational Biology, Biomedical Graduate Studies, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2013-18	Co-Director, Center for Genetics and Complex Traits, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2015	Associate Director, Doctoral Program in Epidemiology, Graduate Group in Epidemiology and Biostatistics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2016-18	Chair, Doctoral Program in Epidemiology, Graduate Group in Epidemiology and Biostatistics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA

Specialty Certification

2007-	American College of Epidemiology (Fellow)

Awards, Honors and Membership in Honorary Societies

1981	Premio di Studio Cavalieri del Lavoro dell'Emilia Romagna
1982	Visiting Student Award, University of Hull, UK

1983	Premio di Laurea A. Raunich, University of Bologna, Italy
1987	Premio FISME, Best Postdoctoral Presentation, Fisme, Siena, Italy
1989	Premio A.I.R.H. 1989, Junior Investigator Award in Human Genetics, Italy
1993	NIH-NIMH Stipend, 1993 World Congress on Psychiatric Genetics, New Orleans, LO
2013	Scientific Merit Award from the Italian General Consulate in Philadelphia

Memberships in Professional and Scientific Societies

1992-	European Society of Human Genetics (Member of Director Board, 1994-1999) (Member of the Scientific Program Committee, 1998-2002)
1994-	American Society of Human Genetics
2000-	International Genetic Epidemiology Society
2016-19	International Epidemiological Association
2012-15	Association for Women in Science

Other Professional Activities

2007	Frontiers in Population Genomics: Research Directions for NHGRI, presenter and discussion panelist
2017	Edward S. Cooper Society Leadership Development Program, Wharton School, University of Pennsylvania
2017	European Union Horizon 2020 Program, Industry Special Monitoring Group member, WITDOM

Grant reviewer for:

2000	Deutsches Zentrum fur Luft- und Raumfahrt, Germany
2001	NWO, The Netherlands
2001-03	Boston Area Diabetes Endocrinology Research Center (BADERC), MA
2002	Fondazione CARIPLO, Italy
2004	Science Foundation Ireland
2004	Israel Science Foundation
2005, 2016-17	Research Grant Council, Hong Kong
2005, 2015, 2017	Medical Research Council, UK
2005-07	NRSA Postdoctoral Fellowships in Genes, Genetics, & Genomics
2006-13	NHLBI Program Project Review Committee
2007	NIMH Whole Genome Association Analysis Review Committee
2009	NIH Challenge Grants in Health and Science Research
2009	Biomedical Research Council – National Medical Research Council, Singapore
2010	NIH Director's Opportunity for Research in Five Thematic Areas Review Panel
2011	Wellcome Trust, UK

2011	ARSEP (Fondation pour l'Aide à la Recherche sur la Sclérose En Plaques), France
2011	NIH Director's Early Independence Awards
2011, 2014	NIDDK Special Emphasis Panel
2014	Fund for Scientific Research-FNRS, Belgium
2014	NIH Center for Scientific Review, Infectious disease, Reproductive health, Asthma and Pulmonary conditions (IRAP) Study Section, *ad-hoc* member
2014	Foundation for NIH
2015	NIMH Psychiatric Gene Network Review Committee
2016	FWF - Austrian Science Fund, Austria
2016-	NIH Center for Scientific Review, IRAP Study Section, permanent member
2018	Crohn's Disease Program, The Leona M. and Harry B. Helmsley Charitable Trust
2020	European Research Council, Starting Grants 2020

Editorial Positions

1994-05	Section Editor, *European Journal of Human Genetics*
1999-19	Member of Editorial Board, *Human Heredity*
2004-12	Editor in Chief, *Human Heredity*
2019-	Member of Editorial Board, *Genetics and Genomics Next*

Journal reviewer for (with year of first review):

2000	*American Journal of Human Genetics*
2001	*Cancer Research*, *Human Genetics*, *European Journal of Human Genetics*
2002	*Arthritis and Rheumatism*, *Molecular Medicine*
2003	*Human Heredity*, *Human Mutation*
2005	*Biological Psychiatry*, *BMC Medical Genetics*
2006	*Annals of Human Genetics*, *Genomics*, *Molecular Vision*, *Bone*
2007	*Osteoporosis International*, *Clinical Genetics*, *Psychiatry Research*, *American Journal of Medical Genetics*, *Genetic Analysis Workshops*
2008	*Calcified Tissue International*, *PLoS Genetics*
2009	*Human Molecular Genetics*, *Neuroscience Letters*
2010	*Schizophrenia Research*, *Journal of Adolescent Health*, *BMC Genomics*
2011	*International Journal of Neuropsychopharmacology*
2013	*Therapeutic Advances in Cardiovascular Disease*
2014	*PLoS ONE*
2015	*Journal of Crohn's and Colitis*, *Cancer Discovery*, *WIREs Systems Biology and Medicine*, *Oncotarget*, *ME Journal of Medical Genetics*
2016	*Genome Medicine*

2017	*Oncotarget*
2018	*BMC Medical Genomics*

Academic and Institutional Committees

2007-09	Committee for Appointment and Promotions, Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2007-08	Biostatistics and Epidemiology Strategic Planning Committee, Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2007-21	Research Seminar Committee, Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2008-21	Chair, Scientific Review Committee, Division of Human Genetics, Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2010-21	Special Programs in Education Committee, Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2012-13	Search Committee, Director of Research, University of Pennsylvania School of Dental Medicine, Philadelphia, PA
2012-20	Committee for Appointment and Promotions, Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2012-21	Diversity Search Advisor, Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2013-21	Epidemiology Faculty Recruitment Committee, Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2015-16	Co-Chair, Candidacy Exam Committee, Genomics and Computational Biology Graduate Group, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2015	Search Committee, Division of Neonatology Chair, Department of Pediatrics, The Children's Hospital of Philadelphia, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2015-21	Search Committee, Genetic Epidemiology, Department of Biostatistics and Epidemiology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2015-16	Chair, Admissions Committee, Doctoral Program in Epidemiology, Graduate Group in Epidemiology and Biostatistics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2015-18	Secretary-Elect/Secretary/Past Secretary, Faculty Senate Executive Committee, University of Pennsylvania, Philadelphia, PA
2015-20	Advisor, Faculty Advising Program, Department of Pediatrics, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA

2016	Committee on Committees, Faculty Senate Executive Committee, University of Pennsylvania, Philadelphia, PA
2017	Search Committee, Division of Endocrinology Chair, Department of Pediatrics, The Children's Hospital of Philadelphia, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2019-21	Admissions Committee, Doctoral Program in Genomics and Computational Biology, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA

Major Academic and Clinical Teaching Responsibilities

1989-90	Human Genetics, University of Genoa School of Medicine, Genoa, Italy
1990-92	Population Genetics, Course Director University of Genoa School of Medicine, Genoa, Italy
1994-97	Human Genetics, University of Genoa School of Medicine, Genoa, Italy
1994-96	Human Genetics for Basic Science, Columbia University College of Physicians and Surgeons, New York, NY
1995-96	Probability and Statistics, Course Director Columbia University School of Public Health, New York, NY
2001-20	Human Genetics GE637, Thomas Jefferson University, Philadelphia, PA
2006-	Genetics, Sapienza University International Medical School, Rome, Italy
2006-07	Director of Human Genetics Track, MSCE Program, CCEB, Perelman School of Medicine, University of Pennsylvania
2006-20	Introduction to Genetic Epidemiology EPID575, Course Co-Director, Perelman School of Medicine, University of Pennsylvania
2007-09	Human Genetics N561, University of Pennsylvania School of Nursing
2007-12	Topics in Human Genetics and Disease CAMB630, Course Co-Director, Perelman School of Medicine, University of Pennsylvania
2010-18	Advanced Topics in Behavioral Genetics NGG578/CAMB578/BIOL488, Perelman School of Medicine, University of Pennsylvania
2010	Advanced Methods for Analysis of Complex Genetic Traits EP675, Course Co-Director, Perelman School of Medicine, University of Pennsylvania
2012	Introduction to Bioinformatics GCB535, Perelman School of Medicine, University of Pennsylvania
2013-20	Genetic Principles CAMB550, Perelman School of Medicine, University of Pennsylvania
2013	Genetic Analysis BIOL540/CAMB541, Perelman School of Medicine, University of Pennsylvania
2015-20	Genetic Foundations of Disease, MD Program, Perelman School of Medicine, University of Pennsylvania
2015-18	Advanced Methods for Analysis of Complex Genetic Traits EPID675, Course Co-Director, Perelman School of Medicine, University of Pennsylvania

2015-18	Co-Director, Human Genetics Concentration, Master of Science in Clinical Epidemiology, CCEB, Perelman School of Medicine, University of Pennsylvania, Philadelphia, PA
2016	Reproductive Epidemiology, EPID646, Perelman School of Medicine, University of Pennsylvania
2016-20	Advanced Topics in Epidemiology, EPID640, Perelman School of Medicine, University of Pennsylvania
2017	Human Genetics, HG637, Jefferson College of Biomedical Sciences, Thomas Jefferson University

Students and mentees:

1990-92	Alessandra Bolino, undergraduate student, University of Genoa, Italy
1993-96	Alessandra Bolino, PhD student, University of Genoa, Italy
1994-98	Valeria Brancolini, post-doctoral fellow, Columbia University
1997	Paola Forabosco, post-doctoral fellow, Rockefeller University
1999-03	Claudia Specchia, PhD student, University of Genoa, Italy
2003-07	Francesca Lantieri, PhD student, University of Genoa, Italy
2004	Patrizia Zavattari, post-doctoral fellow, Nemours Children's Clinic
2004	Maria Rosa Valvano, post-doctoral fellow, Nemours Children's Clinic
2006-10	Kathryn Blake, PharmD, Nemours Children's Clinic (K23 co-sponsor)
2006	Pedro Sanchez, Master of Science in Clinical Epidemiology student, University of Pennsylvania
2006-08	Haitao Zhang, post-doctoral fellow, Children's Hospital of Philadelphia
2007-08	Mario Capasso, post-doctoral fellow, Children's Hospital of Philadelphia
2007-09	Paula Goldenberg, Master of Science in Clinical Epidemiology student, University of Pennsylvania
2007	Luca Longo, MSc student, University of Genoa, Italy
2008	Zafar Zaheer, PhD student, Statistics, University of Peshawar, Pakistan
2008-09, 2011-12	Francesca Lantieri, post-doctoral fellow, Children's Hospital of Philadelphia
2008-09	Fabrice Danjou, PhD student, Genetics, University of Cagliari, Italy
2008-09	Marco Simonini, Nephrology fellow, University San Raffaele, Milan, Italy
2008-09	Stephanie Ciosek, MPH student, University of Pennsylvania
2009-10	Silvia Francisci, post-doctoral fellow, Children's Hospital of Philadelphia
2009-11	Valeria Latorre, PhD student, University of Calabria, Italy
2010-13	Ellen Tsai, PhD student, Genomics and Computational Biology, University of Pennsylvania

2010-15	Stephanie Ciosek, PhD student, Epidemiology, University of Pennsylvania
2013-17	Ying Chen, PhD student, Genomics and Computational Biology, University of Pennsylvania
2013-	Judith Kelsen, MD, Children's Hospital of Philadelphia (K23 mentor)
2015	Rajashree Mishra, PhD student, Genomics and Computational Biology, University of Pennsylvania (rotation mentor)
2015-17	Edward Zhao, undergraduate student, Vagelos Scholars Program in the Molecular Life Sciences, University of Pennsylvania
2015-17	Rebecka Hess, DVM, Master of Science in Clinical Epidemiology student, University of Pennsylvania
2015	Sabine Schneider, MD/PhD student, Genetics, University of Pennsylvania (rotation mentor)
2016-19	Maire Conrad, MD, Master in Translational Research student, University of Pennsylvania
2016	Sanjana Sundaresan, undergraduate student, SASTRA University, India (research project mentor)
2016-17	Lauren Hochman, undergraduate student, School of Arts and Science, University of Pennsylvania
2017	Ramyiadarsini Elangovan, MD student, University of Oxford, Oxford, UK (research project mentor)
2018	Shobana Sankar, undergraduate student, SASTRA University, India (research project mentor)
2018	Sowndharya Subramanian, undergraduate student, SASTRA University, India (research project mentor)
2018	Alexa Woodward, PhD student, Epidemiology, University of Pennsylvania (rotation mentor)
2018-19	Alessandro Testori, MD, PhD, post-doctoral fellow, The Children's Hospital of Philadelphia
2018	Tara Klingner, PhD student, Epidemiology, University of Pennsylvania (rotation mentor)
2018	Vaishnave Subbramanian, undergraduate student, SASTRA University, India (research project mentor)
2019-	Alejandro Q. Nato, Jr., PhD, Assistant Professor, Marshall University, WV-CTSI OPEN Grant mentor
2019-20	Tancredi Pentimalli, MD student, University of Rome Sapienza (dissertation advisor)

Participation in PhD dissertation and other academic oversight committees:

1995	Fatimah Haghighi, PhD student, Genetics, Columbia University (PhD Committee member)
2006	Mark Levenstien, PhD student, Statistical Genetics, Rockefeller University (Faculty Advisory Committee member, External Examiner)

2013-14	Yun (Rose) Li, MD/PhD student, Genomics and Computational Biology, University of Pennsylvania (PhD Committee member)
2014	Joseph Glessner, PhD student, Genomics and Computational Biology, University of Pennsylvania (PhD Committee member)
2015-17	Maire Conrad, MD, GI fellow, Children's Hospital of Philadelphia (Scholarship Oversight Committee member)
2015-17	Michelle Kaplinski, MD, Cardiology fellow, Children's Hospital of Philadelphia (Scholarship Oversight Committee member)
2016-	Ramakrishnan Rajagopalan, PhD student, Drexel University, Philadelphia (PhD Committee member)
2016-19	Katie Siewert, PhD student, Genomics and Computational Biology, University of Pennsylvania (PhD Committee chair)
2016-18	Elizabeth Bhoj, MD PhD, post-doctoral fellow, The Children's Hospital of Philadelphia (Scientific Advisory Committee member)
2016-19	Rajashree Mishra, PhD student, Genomics and Computational Biology, University of Pennsylvania (PhD Committee chair)
2017	Laura Bryant, PhD student, Neuroscience, University of Pennsylvania (PhD Committee member)
2018-23	Elizabeth Bhoj, MD PhD, Children's Hospital of Philadelphia (K08 Scientific Advisory Committee member)
2019-20	Laura Egolf, PhD student, Cell and Molecular Biology, University of Pennsylvania (PhD Committee member)
2019-20	Dan Ju, PhD student, Cell and Molecular Biology, University of Pennsylvania (PhD Committee member)
2019-20	Jing Zhang, PhD student, Epidemiology, University of Pennsylvania (PhD Committee chair)

Organizing Roles in Scientific Meetings

1990	International Workshop "The Identification of the CF Gene: Recent Progress and New Research Strategies". Sestri Levante, Italy. Member of Scientific Committee
1997	XXIX European Human Genetics Conference. Genova, Italy. Member of Local Organizing Committee
1998-02	European Human Genetics Conference. Member of Scientific Program Committee
1999-05	Introductory Course in Genetic/Linkage Analysis, Rockefeller University, New York, NY. Course Director
2001	10th International Congress of Human Genetics. Vienna, Austria. Member of Scientific Program Committee
2005-10	Statistical Genetic Analysis of Complex Phenotypes European School of Genetic Medicine, Bologna, Italy. Course Director
2007	15th Annual International Conference on Intelligent Systems for Molecular Biology (ISMB) & 6th European Conference on Computational Biology (ECCB), Vienna, Austria. Member of Program Committee for Track Bioinformatics of Disease

2011-	International Sardinian Summer School in Genomics, Polaris Technology Park, Pula (CA), Italy. Course Director
2013-14	Mid-Atlantic Genetic Epidemiology and Statistics Conference, Philadelphia, PA. Chair of Organizing Committee
2015-	Symposium on Advances in Genomics, Epidemiology, and Statistics, Philadelphia, PA. Chair of Organizing Committee
2018	European Mathematical Genetics Meeting, Cagliari, Italy. Member of Scientific Organizing Committee

Bibliography

Research Publications, peer reviewed (* indicates Dr. Devoto's mentee):

1. Romeo G, Bianco M, **Devoto M**, Menozzi P, Mastella G, Giunta AM, Micalizzi C, Antonelli M, Battistini A, Santamaria F, Castello D, Marianelli A, Marchi AG, Manca A, Miano A: Incidence in Italy, genetic heterogeneity and segregation analysis of cystic fibrosis. *Am J Hum Genet* 37:338-349, 1985.

2. **Devoto M**, Prosperi L, Dagna Bricarelli F, Coviello DA, Croci G, Zelante L, Ferranti G, Tanconi R, Stomeo C, Romeo G: Frequency of consanguineous marriages among parents and grandparents of Down patients. *Hum Genet* 70:256-258, 1985.

3. Branzi A, Romeo G, Specchia S, Lolli C, Binetti G, **Devoto M**, Bacchi M, Magnani B: Genetic heterogeneity of hypertrophic cardiomiopathy. *Int J Cardiol* 7:129-133, 1985.

4. Brignola C, Lanfranchi GA, Campieri M, Bazzocchi G, **Devoto M**, Boni P, Farruggia P, Veggetti S, Tragnone A: Importance of laboratory parameters in the evaluation of Crohn's disease activity. *J Clin Gastroenterol* 8:245-248, 1986.

5. **Devoto M**, Lozito A, Staffa G, D'Alessandro R, Saquegna T, Romeo G: Segregation analysis of migraine in 128 families. *Cephalalgia* 6:101-105, 1986.

6. Romeo G, **Devoto M**, Bianco M: Homogeneity vs. heterogeneity of cystic fibrosis in Italy. *Am J Hum Genet* 39:283-284, 1986.

7. Vitale E, **Devoto M**, Mastella G, Romeo G: Homogeneity of cystic fibrosis in Italy. *Am J Hum Genet* 39:832-836, 1986. PMCID: PMC1684116

8. Tonini GP, Verdona G, **Devoto M**, Sansone R, Cornaglia-Ferraris P: N-myc oncogene amplification and catecholamine metabolism in patients with neuroblastoma. *Lancet* ii:795, 1987.

9. Romeo G, **Devoto M**, Archidiacono N, Ferlini A, Roncuzzi L, Melis MA, Paderi E, Ferrari M, Tedeschi S, Galluzzi G, Felicetti L: Italian experience regarding the prevention of Duchenne and Becker muscular dystrophies. *Eur J Pediatr* 147:412-415, 1988.

10. Schwartz M, Super M, Schmidtke J, Buys C, Farrall M, Halley D, Krawczak M, Poncin JE, Loukopoulos D, **Devoto M**: Prenatal diagnosis of cystic fibrosis using linked DNA probes. *Prenat Diagn* 8:619-624, 1988.

11. Estivill X, Farrall M, Williamson R, Ferrari M, Seia M, Giunta AM, Novelli G, Potenza L, Dallapiccola B, Borgo G, Gasparini P, Pignatti PF, De Benedetti L, Vitale E, **Devoto M**, Romeo G: Linkage disequilibrium between cystic fibrosis and linked DNA polymorphisms in Italian families: a collaborative study. *Am J Hum Genet* 43:23-28, 1988.

12. Farrall M, Wainwright BJ, Feldman GL, Beaudet A, Sretenovic D, Halley D, Simon M, Dickerman L, **Devoto M**, Romeo G, Kaplan J-C, Kitzis A, Williamson R: Recombinations between IRP and cystic fibrosis. *Am J Hum Genet* 43:471-475, 1988.

13. Romeo G, **Devoto M**, Costa G, Roncuzzi L, Catizone L, Zucchelli P, Germino GG, Keith T, Weatherall DJ, Reeders ST: A second genetic locus for autosomal dominant polycystic kidney disease. *Lancet* 2:8-11, 1988.

14. Barabino A, Haupt R, Rosati U, Scarsi P, Boni L, **Devoto M**, Poggi O, Durand P: La ricerca pediatrica in Italia: indagine sulla produzione nel triennio 1983-1985. *Riv Ital Ped (IJP)* 14:408-411, 1988.

15. Kitzis A, et al. (among others **Devoto M**): Unusual segregation of cystic fibrosis alleles. *Nature* 336:316, 1988.

16. Figus A, Lampis R, **Devoto M**, Ristaldi MS, Ideo A, De Virgilis S, Nurchi AM, Corrias A, Corda R, Lai ME, Tocco A, Deplano A, Solinas A, Zancan L, Lee W-H, Cao A, Pirastu M, Balestrieri A: Carrier detection and early diagnosis of Wilson's disease by restriction fragment lenght polymorphism analysis. *J Med Genet* 26:78-82, 1989. PMCID: PMC1015554

17. **Devoto M**, De Benedetti L, Seia M, Piceni Sereni L, Ferrari M, Bonduelle ML, Malfroot A, Lissens W, Balassopoulou A, Adam G, Loukopoulos D, Cochaux P, Vassart G, Szibor R, Hein J, Grade K, Berger W, Wainwright B, Romeo G: Haplotypes in cystic fibrosis patients with or without pancreatic insufficiency from four European populations. *Genomics* 5:894-898, 1989.

18. Ferrari M, Antonelli M, Bellini F, Borgo G, Castiglione O, Curcio L, Dallapiccola B, **Devoto M**, Estivill X, Gasparini P, Giunta A, Marianelli L, Mastella G, Novelli G, Pignatti P, Romano C, Romeo G, Seia M, Williamson R: Genetic differences in cystic fibrosis patients with and without pancreatic insufficiency. An Italian collaborative study. *Hum Genet* 84:435-438, 1990.

19. **Devoto M**, Ronchetto P, Romano L, Romeo G: Analysis of deltaF508 does not confirm a previously reported recombination in a cystic fibrosis family. *Am J Hum Genet* 46:1004-1005, 1990. PMCID: PMC1683601

20. Gasparini P, Cappello N, Dallapiccola B, **Devoto M**, Estivill X, Ferrari M, Leoni G, Novelli G, Piazza A, Pignatti PF, Romeo G, Rosatelli C, Savoia A, Seia M, Williamson R: Regional distribution of cystic fibrosis linked DNA haplotypes in Italy: a collaborative study. *Gene Geogr* 4:53-64, 1990.

21. Dean M, Amos JA, Lynch J, Romeo G, **Devoto M**, Ward K, Halley D, Oostra B, Ferrari M, Russo S, Weir BS, Finn PB, Collins FS, Iannuzzi MC: Prenatal diagnosis and linkage disequilibrium with cystic fibrosis for markers surrounding D7S8. *Hum Genet* 85: 275- 278, 1990.

22. Balassopoulou A, Loukopoulos D, Kollia P, **Devoto M**, Adam G, Arvanitakis S, Hadjisevastou H: Cystic fibrosis in Greece: typing with DNA probes and identification of the common molecular defect. *Hum Genet* 85:393-394, 1990.

23. Cremonesi L, Ruocco L, Seia M, Russo S, Giunta A, Ronchetto P, Fenu L, Romano L, **Devoto M**, Romeo G, Ferrari M: Frequency of the deltaF508 mutation in a sample of 175 Italian cystic fibrosis patients. *Hum Genet* 85:400-402, 1990.

24. Ronchetto P, **Devoto M**, Puliti A, Romeo G, Sokolov B, Kalinin VN, Vorsanova SG, Krainiaia GV, Reznik BY: Preliminary results on the frequency of the deltaF508 mutation in cystic fibrosis patients from the USSR. *Hum Genet* 85:423-424, 1990.

25. European Working Group on Cystic Fibrosis Genetics (EWGCFG): Gradient of distribution in Europe of the major CF mutation and of its associated haplotype. *Hum Genet* 85:436-442, 1990.

26. Dianzani I, Camaschella C, Saglio G, Ferrero GB, Romeo G, **Devoto M**, Romano C, Cerone R, Giovannini M, Riva E, Trefz FK, Lichter-Konecki U, Woo SLC: Haplotype distribution and molecular defects of PKU in Italy. *J Inher Metab Dis* 13:292-294, 1990.

27. Dianzani I, **Devoto M**, Camaschella C, Saglio G, Ferrero GB, Cerone R, Romano C, Romeo G, Giovannini M, Riva E, Angeneydt F, Trefz FK, Okano Y, Woo SLC: Haplotype distribution and molecular defects at the phenylalanine hydroxylase locus in Italy. *Hum Genet* 86:69-72, 1990.

28. Ferrari M, Colombo C, Sebastio G, Castiglione O, Quattrucci S, Dallapiccola B, Leoni G, Zanda M, Romano L, **Devoto M**: Cystic fibrosis patients with liver disease are not genetically distinct. *Am J Hum Genet* 48:815-816, 1991.

29. Kalaydjieva L, Dworniczak B, Aulehla Scholz C, **Devoto M**, Romeo G, Stuhrmann M, Horst J: Phenylketonuria mutations in Southern Europeans. *Lancet* 337:865, 1991.

30. **Devoto M**, Ronchetto P, Fanen P, Telleria Orriols JJ, Romeo G, Goossens M, Ferrari M, Magnani C, Seia M, Cremonesi L: Screening for non deltaF508 mutations in 5 exons of the cystic fibrosis transmembrane conductance regulator (CFTR) gene in Italy. *Am J Hum Genet* 48:1127-1132, 1991.

31. **Devoto M**, Castagnola S, Saha N, Chetsanga C, Allen M, Gyllensten U, Romeo G: Screening for the major cystic fibrosis mutation in non-Caucasian populations. *Am J Hum Genet* 49:903-904, 1991. PMCID: PMC1683156

32. Peral B, Hernandez-Chico C, San Millan JL, Granell R, Molano J, Carrasco S, Telleria JJ, **Devoto M**, Moreno F: The deltaF508 mutation and RFLP-linked loci in Spanish cystic fibrosis families. *Hum Genet* 87:516-517, 1991.

33. Kalaydjieva L, Dworniczak B, Aulehla Scholz C, **Devoto M**, Romeo G, Stuhrmann M, Kucinskas V, Yurgelyavicius V, Horst J: Silent mutations in the phenylalanine hydroxylase gene as an aid to the diagnosis of phenylketonuria. *J Med Genet* 28:686-690, 1991. PMCID: PMC1017055

34. Ronchetto P, Telleria JJ, Fanen P, Cremonesi L, Ferrari M, Magnani C, Seia M, Goossens M, Romeo G, **Devoto M**: A nonsense mutation (R1158X) and a splicing mutation (3849+4A->G) in exon 19 of CFTR. *Genomics* 12:417-418, 1992.

35. Cossu P, Pirastu M, Nucaro A, Figus A, Balestrieri A, Borrone C, Giacchino R, **Devoto M**, Monni G, Cao A: Prenatal diagnosis of Wilson's disease by analysis of DNA polymorphism. *N Engl J Med* 327:57, 1992.

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