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Curriculum Vitae

Alessandro Mussa, MD, PhD

Date and place of birth:	October 8th, 1980 in Pinerolo (TO), Italy
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Studies and Academic positions

- Degree in Medicine and Surgery in 2005 at the University of Turin
- Specialization in Pediatrics in 2011 at the Postgraduate School of Pediatrics, University of Turin
- Research Trainee at the Biochemical Genetics Laboratory (BGL) of the Mayo Clinic in Rochester, MN, U.S.A, in 2009-2010, focusing on neonatal screening by Tandem Mass Spectrometry
- Subspecialty in Pediatric Endocrinology and Diabetology at the School of Pediatric Endocrinology and Diabetology of the Italian Society of Pediatric Endocrinology and Diabetology (SIEDP), third edition, 2012
- PhD in Biomedical Sciences and Human Oncology, Experimental Pediatrics, at the University of Turin, Italy, in 2015
- National Scientific Qualification, Associate Professor (session 2016-2018, 1st quarter, question 14719, sector 06/G1 General and Specialistic Pediatrics)
- Associate Professor at the Department of Public Health and Pediatric Sciences of the University of Torino, Italy

Clinical Activity

- 2011-2014 General Pediatrics at Regina Margherita Children Hospital, Città della Salute e della Scienza di Torino, Emergency Department, Torino, Italy

- 2014- 2019 Staff Member at Neonatology and Intensive Neonatal Care Unit of the S.Anna Hospital, Città della Salute e della Scienza di Torino, Emergency Department, Torino, Italy
- 2019 – ongoing Reasearcher (up to 2/2022) and then Associate Professor and Staff Physician in charge of the Pediatric Clinical Genetics of the Regina Margherita Children Hospital, Torino, Italy

Scientific and Research Activity

Research activity has been mainly focused on pediatric endocrinology, genetic diseases of endocrinological interest and congenital inborn errors of metabolism. Since 2007 clinical research has been mainly focused on infantile endocrine-metabolic pathology including thyroid nodule in pediatric age, hyperthyotropinemia and isolated and syndromic subclinical hypothyroidism, congenital hypothyroidism, syndromic childhood thyroid disorders, autoimmune thyroid diseases, application of the ultrasonic methodology to the evaluation of the bone condition in pediatrics and pediatric endocrinology, hyperphenylalaninemia, cryptorchidism and its effects on fertility, genetic obesity, Noonan syndrome. In 2009 and 2010, as Visiting Scientist and Research Trainèe at the Biochemical Genetics Laboratory (BGL) of the Mayo Clinic in Rochester, MN, USA, specialized in molecular and biochemical diagnostics of congenital metabolic defects, the research activity aimed at the identification of rare forms of congenital adrenocortical insufficiency without elevation of 17-hydroxy-progesterone through the use of Tandem Mass Spectrometry. Since 2009, research included genomic imprinting disorders and related pediatric pathologies with particular reference to overgrowth and growth restriction syndromes (Beckwith-Wiedemann and Silver-Russell Syndromes) and focused on definition of a correlation between genotype and phenotype, definition of the epidemiology of the syndrome in pediatrics, design and implementation of a new automatic method for the determination of the alpha-fetoprotein tumor marker for the screening of hepatoblastoma in infancy, definition of cancer incidence and appropriate monitoring methods during the first decade of life, realization of Italian and European recommendations for diagnosis and follow-up, delineation of new molecular mechanisms and diagnostic methods in the syndrome by Beckwith-Wiedemann. He is a member of the European Study Group on Beckwith-Wiedemann syndrome of the EUCID.net consortium (European Network of Human Congenital Imprinting Disorders, BMB COST Action BM1208, Chair Prof. Thomas Eggerman), scientific group dedicated to the definition of European Guidelines on diagnosis and follow-up of patients with Beckwith-Wiedemann syndrome. P.I. or sub-investigator of national and international studies. PI of the CBYL719F12201 study (EPIK-P2, sponsored by Novartis). Recent research focused on lateralized overgrowth disorders of the PIK3CA/AKT/mTOR pathway.

Scientific production and bibliometric indicators (Scopus)

Dr. Mussa authored several original investigations published on peer-reviewed international journal with IF and book chapters, as well as presented original communication in several national and international meetings. Since 2006, has been author of more than 110 publications listed on Scopus, with overall more than 2100 citations and a cumulative impact factor: >350 (ISI Web of Science

InCite Journal Citation report) and an *h*-index: 35. Among these, he authored some recent research on PIK3CA-related overgrowth spectrum and overlatpping disorders (Mussa et al, J Med Genet 2022; Mussa et al, Cancers (Basel), 2021; Carli et al, Clin Genet, 2021; Carli et al, Clin Genet, 2021)

Awards and Prizes

Awarded for several presentations at National and International meetings:

- 2007: Best Oral Presentation Award Clinical Reports section at the meeting of the Italian Society of Pediatric Endocrinology and Diabetology
- 2011: Best Oral Presentation Award at the Joint Congress of the Italian Society of Inherited Metabolic Deiseases and Italian Society of Pediatric Genetics and Congenital Disabilities
- 2011: Best Oral Presentation Award at the XIV meeting of National Italian Society of Human Genetics
- 2011: Best Oral Presentation Award at the meeting of the Italian Society of Pediatric Endocrinology and Diabetology
- 2017: Best Article on Clinical Research Award at the 2017 meeting of the Italian Society of Human Reproduction
- 2018: European Journal Human G 2018 Citations Award from the European Journal of Human Genetics, 2018 European Society for Human Genetics meeting

Grant awarded and responsibilities in scientific projects

2007: Programma di Ricerca Scientifica di Rilevante Interesse Nazionale (PRIN) 2007 MIUR (prot. 007SRLBSW), "Clinical selection of patients with congenital growth disorders and molecular analysis of the 11p15.5 region using methylation-sensitive, multiplex-ligationdependent, probe amplification assay (MS-MLPA)", as member of Research Unit

2009: Programma di Ricerca Scientifica di Rilevante Interesse Nazionale (PRIN) 2009 MIUR (prot. 2009MBHZPR), "Clinical selection, 11p15 chromosomal region MS-MLPA based molecular analysis, and definition of follow-up strategies for patients affected by Beckwith -Wiedemann and Silver-Russell syndromes", as member of Research Unit

2014: Randomized Trial sponsored by AIFA "Evaluation of long-term benefit-risk profile of levothyroxine treatment in children with congenital hypothyroidism: influence of initial levothyroxine dose on neurodevelopmental, growth, cardiovascular and skeletal outcomes" - FARM8A8FHP – as member and head of Research Unit

2016: Sub-investigator in the multicentric randomized double-blinded controlled trial "A Phase 2b Randomised, Double-blind, Placebo-controlled Study to Evaluate the Safety and Efficacy of MEDI8897, a Monoclonal Antibody With an Extended Half-life Against Respiratory Syncytial Virus, in Healthy Preterm Infants" - MedImmune Protocol D5290C00003 (EU) MEDI8897 - Sponsor: MedImmune, LLC, PLC, (Gaithersburg, MD)

2017: Sub-investigator multicentric phase IV multicentric blinded controlled trial "A Phase IV, observer-blind, randomised, cross-over, placebo-controlled, multicentre study to assess the

immunogenicity and safety of a single dose of Boostrix™ in pregnant women" - DTPA (BOOSTRIX)-047 – Sponsor: GlaxoSmithKline (GSK) Biologicals

2022: PI of the CBYL719F12201 study (EPIK-P2, sponsored by Novartis), pending Ethical Committee approval.

2024: Entrusted as PI of the "Bando Ricerca Pediatrica 2024" of the Italian Society of Pediatrics (SIP) for the project entitled "National Clinical Network for the Treatment of Pediatric Genetic Syndromes with Precision and Repositioned Drugs"

Editorial Activity

• Chief Editor (since 2020) and Member of the Editorial Board of the International Journal of Pediatrics (Hindawi Publishing Corporation, ISSN: 1687-9740 (Print) ISSN: 1687-9759 (Online), DOI: 10.1155 / 7157), peer-reviewed journal, indexed on Pubmed and Web of Science (Thomson Reuters), Open Access that publishes original research, clinical studies and reviews on pediatric subjects) from June 2012 to present.
• Since 2012, Referee activities for the following peer-reviewed indexed journals: Archives of Diseases in Childhood, AIDS care, European Journal of Medical Genetics, Expert Review in Pediatric Endocrinology, International Journal of Pediatrics, Journal of Pediatric Endocrinology & Metabolism, Nutrients , Pediatric Nephrology, Pediatrics, PLoS ONE.

Society activity and consultantship

- Member of the Italian Society of Pediatric Endcrinology and Diabetes, Italian Society of Pediatrics, Italian Society of Neonatology, European Society of Human Genetics, Italian Society of Human Genetics
- Since 2014 member of the Scientific Committee of the Italian Association of Patients with Beckwith-Wiedemann Syndrome (www.AIBWS.org)
- Since 2019 member of the Scientific Committee of the Macrodactily and PIK3CA-related overgrowth spectrum (PROS) Italian Association

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