Diana Carli

MEDICAL DOCTOR SPECIALIST IN MEDICAL GENETICS

After graduating in Medicine and Surgery at the University of Modena and Reggio Emilia in 2012, I specialized in Medical Genetics at the University of Genoa, Turin aggregate headquarters, in 2017 and I completed the PhD in Biomedical Sciences and Oncology in 2022 at the University of Turin. My research activity has been mainly focused on pediatric genetic diseases, congenital malformations, syndromes predisposing to the development of childhood cancers and monogenic disease susceptible to solid organ and hematopoietic stem cells transplantation. I currently work as Assistant Professor in Medical Genetics at the Department of Medical Sciences of the University of Turin.

PERSONAL DATA AND SCIENTIFIC METRICS

Name	Diana
Surname	Carli
Scopus ID	55644192700
Web of Science Researcher ID	K-4169-2018
H-Index	14 (March 27, 2023)
Indexed publications	55 (March 27, 2023)
Total citations	722 (March 27, 2023)

CLINICAL ACTIVITY

Oct 2012 – Jul 2013 **Attending physician**
University Hospital of Modena, Department of Mother & Child, Medical Genetics Unit, Modena, Italy
Clinical genetic evaluation of patients with congenital malformations of the upper limb.

Aug 2013 – Aug 2015 **Post graduate trainee in Medical Genetics**
San Luigi Gonzaga University Hospital, Department of Clinical & Biological Sciences, Medical Genetics Unit, Orbassano, Italy
Clinical genetic evaluation of patients with arrhythmogenic cardiomyopathies, cancer predisposition syndromes and neurodevelopmental disorders. Sanger sequencing.

Sept 2015 – Sept 2017 **Post graduate trainee in Medical Genetics**
Città della Salute e della Scienza University Hospital, Medical Genetics Unit, Torino, Italy
Clinical genetic evaluation of patients with reproductive disorders, neurodevelopmental disorders and congenital malformations, prenatal counseling.

Oct 2017 – Dec 2020 **PhD student**
Department of Public Health and Pediatrics, School of Medicine, University of Torino
Clinical genetic evaluation of pediatric patients with complex phenotypes, congenital malformations and neurodevelopmental disorders. Interpretation of data obtained through next generation sequencing technologies.

Jan 2019 Feb 2019 **PhD student**
Genetics and Rare Diseases Research Division, Ospedale Pediatrico Bambino Gesù, Roma
Interpretation of genomic data obtained through exome and genome sequencing in patients with rare diseases.

Apr 2021 Oct 2022 **Staff Member**
Pediatric Onco-Hematology, Stem Cell Transplantation and Cell Therapy Division, Regina Margherita Children's Hospital, Città della Salute e della Scienza di Torino
Diagnosis, treatment, and genetic evaluation of pediatric patients with oncological diseases.

Oct 2023 Today **Assistant Professor (RTDB) in Medical Genetics**
Department of Medical Sciences, University of Torino, Italy
Genetic evaluation of patients with monogenic disease susceptible to solid organ and hematopoietic stem cells and transplantation.

STUDIES AND ACADEMIC POSITIONS

Oct 2006 Oct 2012 **Degree in Medicine and Surgery**
University of Modena e Reggio Emilia, Italy

Aug 2013 Sept 2017 **Specialization in Medical Genetics**
Postgraduate School of Medical Genetics at the University of Genova, Italy

Oct 2017 Dec 2020 **PhD in Biomedical Sciences and Human Oncology**
University of Torino, Italy

AWARDS AND PRIZES

Dic 2017 **Best clinical article written by an Italian group in 2017 ("Studi TOP 2017")**
1° Conference of the Italian Society of Human Reproduction (SIRU), Roma
Article: Mussa A, Molinatto C, Cerrato F, Palumbo O, Carella M, Baldassarre G, Carli D, Peris C, Riccio A, Ferrero GB. Assisted Reproductive Techniques and Risk of Beckwith-Wiedemann Syndrome. Pediatrics. 2017 Jul;140(1):e20164311. doi: 10.1542/peds.2016-4311. Epub 2017 Jun 20. PMID: 28634246.

Oct 2018 **Best Poster**
XXI Conference of the Italian Society of Human Genetics (SIGU), Catania, 2018
Poster: NBAS associated disease: defining facial features and genotype-phenotype correlation.

Oct 2021 **Reviewers' Choice poster award**
American Society of Human Genetics (ASHG), Virtual Meeting, USA
Poster: Genotype-phenotype correlations in PIK3CA-related overgrowth spectrum (PROS) and overlapping phenotypes: a systematic review of 1007 patients with PIK3CA pathogenic variants.

Dic 2022 **Mijno Family Foundation award 2022**
Department of Medical Sciences, University of Turin, Italy
Project: Functional evaluation of a novel variant of the CASR gene identified in a patient with chronic hypocalcemia.

INVITED TALKS

Oct 2018 **Mutazioni dell'istone H3F3 sono responsabili di disturbi multipli dello sviluppo somatico e neurologico**
Simposio satellite, Riunione Gruppo di Lavoro Epigenetica, XXI Congresso Nazionale Società Italiana di Genetica Umana (SIGU), Catania, Italia

Apr 2020 **Sindrome di Stickler**
Genetica multidisciplinare per specialisti in oftalmologia, FAD online, Genet, Italia

Nov 2020 **Diagnosi Prenatale della Sindrome di Noonan**
Noonan Update - Attualità in tema di Percorsi Diagnostici Terapeutici nella Sindrome di Noonan, FAD online, Dynamicon Education, Italia

Lug 2021 **Prenatal Features in BWSp**
Deciphering Beckwith-Wiedemann Spectrum, Virtual Course, Children's Hospital of Philadelphia, USA

Gen 2022 **Lateralized and Segmental Overgrowth in Children**
3rd International Webinar on Cancer Research and Oncology, Scientific Meditech, Virtual Conference, UK

May 2022 **NGS: potenzialità e limiti in ematologia e immunologia**
Incontro della rete ematologica pediatrica del Piemonte e della Valle d'Aosta (REP), Torino, AccMed, Italia

Jun 2022 **Beckwith Wiedemann Syndrome in adults**
1st International Congress on Beckwith-Wiedemann Syndrome, Cervia, Italy

Sep 2022 **Sindromi con predisposizione oncologica ad esordio pediatrico**
Riunione del Gruppo di Lavoro Genetica Clinica, Società Italiana di Genetica Umana (SIGU), modalità telematica

Oct 2022 **Sindromi con predisposizione oncologica infantile: diagnosi e sorveglianza**
Incontro di Genetica Clinica "Casi Clinici Diagnosticati con Clinical Exome", Arcispedale Santa Maria Nuova, Reggio Emilia

EDITORIAL ACTIVITY

Abstracts reviewer European Society of Human Genetics (ESHG) Conference 2021

Journal reviewer Clinical Genetics; Pediatrics; Hepatology; BMC Medical Genetics; Advances in Therapy; Journal of the American Heart Association

Member of the Editorial board	Journal of Pediatric Genetics – Thieme - ISSN: 2146-4596. 2019-today International Journal of Pediatrics – Hindawi - ISSN: 1687-9759. 2021-today

TEACHING ACTIVITY

Lecturer	University of Turin, School of Specialization in Hospital Pharmacy, Course of Elements of Genetics and Genetic Bases of Diseases Academic years 2021-2022 University of Turin, School of Specialization in Pediatrics, Course of English and Medical Genetics 2 Academic year 2021-2022 University of Turin, School of Specialization in Child Neuropsychiatry, Course of Medical Genetics 2 Academic year 2021-2022
Teaching assistant	University of Turin, Degree Course in Neuro and Psychomotor Therapy of Developmental Age, Course of Pediatric and Medical-Surgical Sciences Academic years 2019-2020, 2020-2021 University of Turin, Degree Course in Pediatric Nursing, Course of Health Problems in Pediatrics 1 Academic years 2019-2020, 2020-2021 University of Turin, Degree Course in Nursing - Local Health Authority "Città di Torino", Course of Pediatrics Academic years 2019-2020, 2020-2021

PARTICIPATION IN SCIENTIFIC PROJECTS

2022-oggi	Sub-Investigator of the multicenter trial “CBYL719F12201 EPIK-P2: A Phase II double-blind study with an upfront, 12-week randomized, placebo-controlled period, to assess the efficacy, safety and pharmacokinetics of alpelisib (BYL719) in pediatric and adult patients with PIK3CA-related overgrowth spectrum (PROS)”. Sponsor: Novartis, PHARMA AG.
2021-2022	Team member of the Phase I Clinical Trials Unit of the Pediatric Onco-hematology, Department of Child Pathology and Care “Regina Margherita”, Città della Salute e della Scienza of Turin, Italy.
2017-2020	Participant in the Scientific Research Program of Relevant National Interest - Programma di Ricerca Scientifica di Rilevante Interesse Nazionale (PRIN) 2015 MIUR (prot. 2015JHLY35), “Molecular genetics and new directions for clinical management of growth disorders associated with genomic imprinting”, University of Turin, Italy.

PROFESSIONAL MEMBERSHIPS

Member	**Italian Society of Human Genetics - Società Italiana di Genetica Umana – SIGU** 2013-active
Member	**Italian Society of Paediatric Genetic Diseases and Congenital Disabilities – Società Italiana di Malattie Genetiche Pediatriche e Disabilità Congenite – SIMGePeD** 2018-active
Member	**European Society of Human Genetics - ESHG** 2019-active
Member	**Italian Association of Pediatric Hematology and Oncology - Associazione Italiana di Ematologia e Oncologia Pediatrica – AIEOP** 2022-active
Member	**European Federation for Immunogenetics - EFI** 2023-active

EXPERTISE

Languages	Italian (native) English(excellent) Spanish (good)
Certificates	GCP (Good Clinical Practice) NIDA Clinical Trials Network – August 22, 2021 PBLS-D Esecutore Sanitario (Supporto di base delle funzioni vitali e defibrillazione precoce in età evolutiva) – March 22, 2022 PALS (Pediatric advanced life support) AHA Provider – March 31, 2022

Turin, March 27, 2023
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