

Claudio Bruno, M.D. Ph.D.
Curriculum Vitae

Personal data

Name: Claudio
Last name: Bruno
Gender: Male
Birth date: 27/12/1965
Place of birth: Milan
Nationality: Italian

Academic Position

Associate Professor in Pediatrics, Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health - DINOGLMI, University of Genova

Position title

Head- Center of Experimental and Translational Myology, IRCCS Istituto Giannina Gaslini, Genova, Italy

Working address

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Education and training

1990 Medical School, College of Physicians and Surgeons, Univ. of Genova, magna cum laude
1995 Residency in Pediatrics, College of Physicians & Surgeons, Univ. of Genova, magna cum laude
2003 PhD in Developmental Neurology, University of Siena, Italy

Employment and research experience

-1996-1999: Research Fellow, Muscle Biochemistry and Pathology Laboratory, H. Houston Merritt Clinical Research Center for Muscular Dystrophy and Related Disorders, Dept. of Neurology, Columbia University, College of Physicians and Surgeons, New York, USA (Director: Prof. S. DiMauro)
-1999-2001: Telethon Scientist, Laboratory of Muscle Pathology, Neuromuscular Disease Unit, Istituto Giannina Gaslini, Genova
-2001-2009: Assistant Medical Researcher, Laboratory of Muscle Pathology, Neuromuscular Disease Unit, Istituto Giannina Gaslini, Genova
-2010: Director, Muscle Pathology Lab, Neuromuscular Disease Unit, Institute Stella Maris, Pisa
-2012-2017: Head, U.O.S.D. Center for Translational Myology and Neurodegenerative Diseases, Department of Neuroscience and Rehabilitation, Giannina Gaslini Institute, Genova
-2018: Head, U.O.S.D. Center for of Experimental and Translational Myology, Department of Neuroscience and Rehabilitation, Giannina Gaslini Institute, Genova

University Memberships

2025- Fellow of the Fudan Scholar Program, Fudan University, Shanghai, China
2024-present: Delegate for the Internationalization Program, DiNOGLMI, Faculty of Medicine, University of Genova
2011-present: Member of the Academic board of PhD School in Pediatrics, University of Genova
2013-present: Member of the Academic board of Residency Program in Pediatrics, University of Genova

Honors and Prize

1991 Winner of a bursary from the Italian Association against Muscular Dystrophy, Genova section.
1997 Winner 1st price-Italian Society of Pediatrics for Young Researcher, Montecatini Terme, 04Sep1997

Appointments in independent Institutions

- 2023-present: Member of the Territorial Ethics Committee-Liguria Region (Pediatrician)
- 2022-present: Representative of the IRCCS Istituto Giannina Gaslini for the European Neuromuscular Reference Network (ERN NMD)
- 2020-present: President of the National Medical-Scientific Commission of the Italian Union of Muscular Dystrophy (UILDM)
- 2015-present: Member of the Scientific Advisory Board (SAB) of SMA Europe Organization
- 2015-present: Member of the Scientific Advisory Board (SAB) SAB of the Italian SMA Family

Scientific Society Membership

- 2025-2027: Member of the Scientific Committee of the Italian Association of Myology (AIM)
- 2015-present: Member of the World Muscle Society (WMS)
- 2001-present: Member of the Italian Association of Myology (AIM) (Founding Member)
- 2000-present: Member of the Italian Society of Paediatrics (SIP)

Organisation of Scientific Meetings

Organizer of numerous courses and specialized workshops on neuromuscular diseases at national and international level. Actively involved in the organization of AIM annual congresses, training events, and the promotion of multicenter collaborations and study groups

Clinical Activity

- 2011-present: Director of UOSD Center for Translational and Experimental Myology, Department of Pediatric Sciences, IRCCS Istituto Giannina Gaslini, Genova
- 2011-present: Principal Investigator in more than 20 clinical trials involving Duchenne Muscular Dystrophy and Spinal Muscular Atrophy patients (GCP certified)
- 2010: Director of UOC Neuromuscular Diseases, IRCCS Stella Maris Foundation, Pisa. Established a Center for Morphological and Biochemical Diagnosis for Muscle Pathologies and an Outpatient Clinic for Muscle Diseases.
- 2002-2009: Attending Physician, UOC Muscular and Neurodegenerative Diseases, University of Genoa, IRCCS Istituto Giannina Gaslini, Genova, Italy

Publications and Research Impact

- Total peer-reviewed publications: 338
- H-index: 62
- Total citations: >14,0912

Key research contributions:

- Clinical trials leading to approval of novel therapies for SMA and DMD
- Diagnostic approaches for congenital myopathies and metabolic myopathies
- Pathophysiological mechanisms and therapeutic targets in muscular dystrophies
- Application of proteomics and machine learning to neuromuscular disorders
- Natural history studies and clinical outcome measures in SMA and DMD

Research Activity

Since 2011, Principal Investigator at the Center for Translational and Experimental Myology. The principal research subjects are: 1) Clinical and molecular diagnosis of neuromuscular disorders with particular emphasis on metabolic myopathies, congenital myopathies, and muscular dystrophies; 2) Study of pathophysiological mechanisms of muscular dystrophies using mouse models; 3) Development and testing of new therapeutic approaches for neuromuscular disorders; 4) Coordination of natural history studies and disease registries; 5) Principal investigator in multiple clinical trials for SMA and DMD. Since 2005 principal investigator of several competitive research grants.

Referee activity for scientific journals

Neuromuscular Disorders, Muscle Nerve, Clinical Genetics, Neurology, Journal of Inherited Metabolic Disorders, PlosOne, Journal of Neuromuscular Diseases.

Personal Statement

My primary focus in both research and clinical practice is on the study and treatment of individuals affected by neuromuscular disorders, with a particular emphasis on metabolic myopathies, congenital myopathies, muscular dystrophies, and SMA.

Throughout my career, I have dedicated myself to unraveling the molecular mechanisms underlying these conditions and developing innovative therapeutic approaches for rare diseases. Notably, I have specialized in pediatric neuromuscular disorders and have played a significant role in establishing clinical and research infrastructure for these conditions.

From March 1996 to March 1999, I was a Research Fellow at the “Muscle Biochemistry and Pathology Laboratory” of the H. Houston Merritt Clinical Research Center for Muscular Dystrophy and Related Disorders, Department of Neurology, Columbia University, New York, directed by Prof. S. DiMauro. There, I conducted clinical research and molecular genetics studies in the field of muscle glycogenoses and mitochondrial encephalomyopathies. This experience provided me with invaluable expertise in the diagnosis and management of metabolic muscle disorders.

In 2010 I served as the Director of UOC Neuromuscular Diseases, IRCCS Stella Maris Foundation, Pisa, where I established a Center for Morphological and Biochemical Diagnosis for Muscle Pathologies and an Outpatient Clinic for Muscle Diseases.

Since January 2012, I have served as the Director of the UOSD Center for Translational and Experimental Myology at the IRCCS Istituto Giannina Gaslini in Genova, a specialized clinical, diagnostic, and research unit dedicated to neuromuscular diseases. In this role, I oversee Day Hospital activities and myopathological consultations within the Institute. Since January 2022, I have also been responsible for the Muscle Pathology Laboratory affiliated with the Center for Translational and Experimental Myology.

Since 2015 I have been actively involved in numerous clinical trials, serving as Principal Investigator for more than 20 clinical trials on Spinal Muscular Atrophy (SMA) and Duchenne Muscular Dystrophy (DMD). My contributions to the field extend beyond clinical practice, as I have secured competitive research grants as principal investigator since 2005 and have authored 338 peer-reviewed publications with an H-index of 62.

My commitment to advancing the understanding and treatment of neuromuscular disorders is also reflected in my leadership roles in professional organizations. I currently serve as the President of the National Medical-Scientific Commission of the Italian Muscular Dystrophy Union (UILDM) for the 2020-2022 and 2023-2025 terms, and I am a member of the Scientific Committee of SMA Families and the Advisory Board of SMA Europe for the four-year period 2022-2025.

As a founding member of the Italian Association of Myology (AIM), I have a long-standing commitment to the field, having served as Treasurer for two consecutive terms (2009-2015) and now as a member of its Scientific Advisory Board Committed.

I authorize the use of my personal data according the D.L. 196/2003 and GDPR 679/16.

Genova, 05 June 2025

Prof. Claudio Bruno

