Università di Genova

Renata Bocciardi

Associate Professor
Tel.+ 39 010/56362725
E-mail : renata.bocciardi@unige.it
URL : https://rubrica.unige.it/personale/VUZCUltu

Current Position

Associate Professor (MEDS-01/A – Medical Genetics, formerly 06/A1 - MEDICAL GENETICS) affiliated with the Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics and Maternal and Child Sciences (DiNOGMI, University of Genoa), serving at the Medical Genetics UOC of IRCCS Istituto Giannina Gaslini (Genoa), with an agreement with the National Health Service (SSN) since 2011.

National Scientific qualification as associate professor in the Italian higher education system, in the call 2021/2023 (Ministerial Decree n. 553/2021 and 589/2021) for the disciplinary field of 06/A1 - Medical genetics. (Academic Recruitment Field 06/A - Pathology and laboratory medicine, according to the national classification). The validity of the qualification is eleven years1, starting from the 07/11/2023 and will expire on the 07/11/2034.

Personal Identifiers

ORCID ID: 0000-0001-8415-3802
ResearchID: J-8577-2016
Scopus ID: 6602978425
Full scientific publications in indexed international journals:
https://pubmed.ncbi.nlm.nih.gov/?term=Bocciardi+R&sort=date
https://iris.unige.it/

Education

1999 – 2003: Specialization in Medical Genetics (50/50 cum laude), University of Genoa, Italy
1993 - 1997: PhD in Human Genetics (IX cycle), University of Turin, Italy
Qualification to practice as a Biologist. **1**
1988 - 1992: **Degree in Biological Sciences** (110/110 cum laude), University of Pisa, Italy

Affiliations

- Member of the **Italian Society of Human Genetics (SIGU)**.
- Member of the **European Society of Human Genetics (ESHG)**.
- Member of the **European Cystic Fibrosis Society (ECFS)**.

Professional Experience and Research Activities

Contracts and Scholarships supporting research activities (in reverse chronological order)

- **01/09/2010 to 31/07/2025** University Researcher (MEDS-01/A – Medical Genetics, formerly 06/A1 - MEDICAL GENETICS) affiliated with the DiNOGMI (University of Genoa).
- **01/01/2008 to 31/08/2010**: Fixed-term employment as **"Dirigente Biologo"** at the Molecular Genetics Service (current "UOC Genetica Medica") of IRCCS Istituto Giannina Gaslini.

- **16/06/2001 to 31/12/2007**: Holder of an Institute G. Gaslini Excellence Contract pursuant to Art. 36 D.P.R. 31 July 1980 n. 617 for the performance of research activities.
- **01/01/2001 to 15/06/2001**: Research contract (Assegno di Ricerca), University of Genoa.
- **01/03/1999 to 31/03/1999**: Holder of an Institute G. Gaslini Contract (Art. 36 D.P.R. 31 July 1980 n. 617) at the Molecular Genetics Service for research activities within the 1997 Ministerial Targeted Research project "Molecular biology and biotechnologies applied to medical genetics: mapping and identification of genes involved in rare genetic diseases and common diseases", provision no. 246 of March.
- **01/03/1998 to 28/02/1999**: Awarded a fixed-term position for foreign post-doc researchers (Poste Vert) at Unité 364 (U364) Institut National de la Santé et de la Recherche Médicale (INSERM) in Nice (France).
- **01/11/1997 to 01/04/1998**: Holder of an Institute G. Gaslini Contract (pursuant to Art. 36 D.P.R. 31 July 1980 n. 617) at the Molecular Genetics Service, following the award of an AIRC Scholarship for the research activities within the project "Molecular genetic study of the RET proto-oncogene" by provision no. 1406 of November 3, 1997.
- **01/07/1995 to 31/12/1995**: Research Stay at the Unité 364 (U364) of the Institut National de la Santé et de la Recherche Médicale (INSERM, Nice, France) for the development of a collaborative project aimed at the functional study of RET gene variants associated with familial thyroid tumors and Hirschsprung's disease, an autosomal dominant congenital condition. The stay was supported from 1/10/1995 to 31/12/1995 by the award of a dedicated European Molecular Biology Organization (EMBO short-term fellowship) scholarship for mobility and exchanges between European research laboratories.
- **18/01/1994 to 30/10/1997**: **PhD in Human Genetics** (University of Turin, cycle IX, duration: 4 years). Holder of a ministerial PhD scholarship.
- **01/01/1994 to 17/01/1994**: Holder of an Institute G. Gaslini Contract (pursuant to Art. 36 D.P.R. 31 July 1980 n. 617) at the Molecular Genetics Service for the performance of research activities within the 1991 Ministerial Targeted Research project "Gaslini North South Project in the prevention, treatment and rehabilitation of genetic diseases causing disability", by ordinance no. 947 of December 23.
- **15/04/1993 to 31/11/1993**: Holder of an Institute G. Gaslini Contract (pursuant to Art. 36 D.P.R. 31 July 1980 n. 617) at the Molecular Genetics Service for the performance of research activities within the Ministerial Targeted Research project "Study of the most frequent genetic causes and prevention models of chronic renal insufficiency", by ordinance no. 241 of April 6, 1993.
- **December 1993**: Awarded an annual "Margherita Divora Florica" scholarship from the Italian Foundation for Cancer Research, subsequently declined due to incompatibility with the doctoral scholarship.

Research Activities

Research Interests from 2006 to Present

- **Study of the post-transcriptional regulation mechanisms of *SLC2A1* gene** (OMIM 138140) expression, responsible for Glut-1 Deficiency (OMIM 606777), and STX1B gene (OMIM 601485) responsible for an AD form of epilepsy (GEFS+9, OMIM 616172), with particular emphasis on the role of non-coding RNAs, and possible use for the development of innovative therapeutic strategies.
- **Characterization of Cystic Fibrosis patients carrying rare and/or functionally understudied *CFTR* gene variants** (orphan variants). The project is based on the molecular characterization of these variants (complex allele search, qualitative and quantitative analysis of CFTR messenger), functional characterization, and response to currently available drugs for Cystic Fibrosis treatment (theratype) in primary nasal cell epithelia derived from patients (ex vivo).
- **Fibrodysplasia Ossificans Progressiva (FOP, OMIM #135100) and the *ACVR1* gene (OMIM # 102576)**. Study of transcriptional and post-transcriptional regulation of ACVR1 gene expression: identification of basic mechanisms and possible therapeutic targets. Generation of cellular systems useful for high-throughput screening (HTS) of small molecules with potential pharmacological effect on ACVR1 gene expression and on the intracellular signaling pathway mediated by the mutated receptor. Role of the innate immune system in FOP pathogenesis. Functional characterization of new

ACVR1 gene variants associated with FOP emerging from diagnostic activity. Over the years, this research activity has benefited from the ongoing collaboration with Prof. Silvia Brunelli (University of Milano-Bicocca) and Dr. Tiziano Bandiera (D3 Pharmachemistry) of the Italian Institute of Technology (IIT, Genoa).

- **Identification of new disease genes** through the characterization of chromosomal rearrangements or, more recently, through exome analysis, and associated with clinical pictures of particular interest.
- **Functional studies in support of Diagnostics**. Development and refinement of a standardized series of functional assays and molecular tools for the characterization of new variants associated with genetic diseases emerging from molecular diagnostics protocols based on massive parallel DNA sequencing (gene panels, WES, WGS), with particular attention to variants in gene expression, regulatory regions (promoter, 5' and 3'UTR) and splicing alterations. These studies have the dual purpose of facilitating, when possible, the interpretation (classification or re-classification) of variants for diagnostic and research purposes, to improve knowledge of basic pathogenetic mechanisms and potentially lead to the identification of molecular targets useful for developing innovative therapies.

Summary of Professional Experience and Research Interests from 1993 to 2005
January 1993 - January 1997: Serving at the Molecular Genetics Laboratory of the G. Gaslini Institute, director Prof. G. Romeo, first as an intern and contractor, then as a PhD student in Human Genetics (University of Turin).
Research projects developed:

- Determination of the physical map and genomic structure of the **RET proto-oncogene** (OMIM #164761).
- Search for mutations in the **proto-RET** in patients with Hirschsprung's disease (OMIM #142623) using non-radioactive SSCP.
- In vitro analysis of the effect of mutations in the **proto-RET** in patients with Hirschsprung's disease (HSCR, OMIM #142623) and Multiple Endocrine Neoplasia type 2A and 2B (MEN 2A, OMIM #171400 and MEN 2B, OMIM #162300).

July 1995 - December 1995 and subsequently September 1996 - December 1996: Research stay at the Cellular and Molecular Immunology Laboratory of INSERM (Unit 364) in Nice (France) directed by Dr. Bernard Rossi.
January 1997 - February 1998: Completion of the PhD in Human Genetics at the Molecular Genetics Laboratory of the G. Gaslini Institute in Genoa. Research project initiated during the PhD and subsequently the subject of post-doctoral work: Implication of the RET proto-oncogene in the genetic etiology of thyroid tumors. Characterization of the cellular signal transduction pathway mediated by the RET proto-oncogene: comparison of the transduction pathway mediated by the activated wild type form with that supported by forms carrying mutations characteristic of HSCR and MEN 2B patients (OMIM #162300). Functional analyses of allelic series and genotype-phenotype correlation.
March 1998 – February 1999: Assignment for foreign researchers at Unit 364 of INSERM, Nice, France, and continuation of the project started in the Molecular Genetics laboratory of the Gaslini Institute. The research topic included various aspects concerning the biological role of the RET proto-oncogene, with particular emphasis on the mechanism by which RET activation by its natural ligand (GDNF) is able to prevent the apoptotic process. The analysis was extended not only to the wild-type receptor but also to some interesting activating mutated forms associated with both HSCR disease (OMIM #142623) and MEN 2A (OMIM #171400) for the study of genotype-phenotype correlation.
March 1999 - January 2000: After completing the Post-doc, the undersigned returned to the Molecular Genetics Laboratory of the G. Gaslini Institute to continue the project concerning the possible role of apoptosis in the genesis of HSCR disease (OMIM #142623), functional analysis of the regulatory regions of the RET gene (OMIM #164761).
January 2000 - 2005: Study of the transcriptional regulation of the RET proto-oncogene (OMIM #164761) in vitro and in vivo, by generating transgenic mice. Identification and characterization of the basal promoter.

Research Projects Funded on a Competitive Basis (from the most recent)

- **2024 ongoing, PNRR-MR1-2023-12378412 (Ministry of Health)**: A precision medicine approach to assess protein function impairment and potential for CFTR rescue in CF and non-CF conditions. Duration 24 months. **Role: Collaborating Researcher**.
- **2023 ongoing**: Fondazione per la Ricerca sulla Fibrosi Cistica - Grant Application FFC 2023 - FFC#03/2023 A functional and molecular approach to address the heterogeneity of the response to CFTR modulators of the N1303K variant. Duration 24 months. **Role: Scientific Lead**.
- **2023 ongoing PRIN**: PROGETTI DI RICERCA DI RILEVANTE INTERESSE NAZIONALE – Call 2022 Prot. 2022TR9N4R control oF Ectopic oSTeOgenesis in Fibrodysplasia Ossificans Progressiva: from mouse to chip and back. (EFESTO). Duration 24 months. **Role: Unit Head**.
- **2021 – 2022 FFC#10/2021 funding from the Italian Foundation for Cystic Fibrosis Research (FFC)** "Theratyping orphan mutations in Italian Cystic Fibrosis patients: meeting unmet needs". **Role: Partner Unit Head**.
- **2022 - 2023, Telethon Seed Grant (renewal)**: GLUT1 deficiency: new therapeutic strategies to increase glucose transport across the Blood-Brain Barrier (BBB). **Collaborating Researcher**.
- **2015-2018 Telethon Foundation, funding No: GGP15196** "New treatment strategies for Fibrodysplasia Ossificans Progressiva" - PI: Prof Roberto Ravazzolo. Role: Head of the research group of the proposing unit and, after the PI's retirement, responsible for the funds.
- **PRIN 2011 (Call 2010-2011)**: Identification of new therapeutic molecules for inflammatory-based orphan muscle diseases. (36 months) - Participant Unit University of Genoa (**Role: Researcher**).
- **University Research Project 2010, University of Genoa (Call reserved for researchers under 45 years of age)** - Heterotopic ossification mechanisms and Fibrodysplasia Ossificans Progressiva (FOP): therapeutic approaches (12 months). **Role: Scientific Lead**.
- **PRIN 2007**: "Characterization of the functional properties of the mutated ACVR1 gene" (24 months). Participant Unit University of Genoa – **Researcher**.
- **FIRB 2001**: “PRIME: Integrated Project on Hereditary Diseases” (36 months). Participant Unit University of Genoa – **Researcher**.
- **PRIN 2001**: "Hereditary platelet disorders: from phenotype to genotype" (24 months). Participant Unit University of Genoa - **Contract Researcher**.

Teaching Activities

- **Academic Year 2010-2011 to present**: Supervisor for Bachelor's (three-year and master's, for Biological Sciences, Biotechnologies, and Biomedical Laboratory Techniques Degree Courses), PhD, and Medical Genetics Specialization School (University of Genoa) theses.
- **Academic Year 2017 – 2018 to present**: **Assignment of MEDICAL GENETICS teaching (code 66686)** for the Master's Degree Course in Applied and Experimental Biology (3 CFU, 24 hours), until 2020 Master's Degree Course in Molecular and Health Biology.
- **Academic Year 2017 - 2018 to present**: **Assignment of MEDICAL GENETICS II teaching (code 68495, 1 CFU)** for the Degree Course in Biomedical Laboratory Techniques. Coordinator of the integrated course of Medical Genetics II and Genetic Techniques (68493) (5 CFU).
- **Academic Year 2012 – 2013 to present**: **Assignment of MEDICAL GENETICS teaching** for the Integrated Course of Physics, Biology and Genetics of the Degree Course in Nursing Sciences (code 67693), and for the Degree Course in Physiotherapy (code 65454), University of Genoa, La Spezia Campus.
- **Academic Year 2011 – 2012**: **Assignment of MEDICAL GENETICS teaching** for the Integrated Course of Physics, Biology and Genetics of the Degree Course in Nursing Sciences (code 67693), and for the Degree Course in Physiotherapy (code 67693), University of Genoa, Savona and Pietra Ligure Campuses.
- **From 2013 to 2019**: **Lectures in Medical Genetics** within the Pediatrics Course, Degree Course in Medicine and Surgery, University of Genoa.
- **From 2011 to 2013**: **Medical Intern's Laboratory** for the Degree Course in Medicine and Surgery, University of Genoa.

- **From 2018 to present**: **Lectures in Medical Genetics** for the Specialization School in Medical Genetics (University of Genoa).

Institutional Activities

- **From 21/01/2025 to present**: **Referring person for Quality Assurance (RAQ)** of the Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics and Maternal and Child Sciences (DINOGMI).
- **Academic Year 2018 to present**: **Member of the Faculty Board** of the Specialization School in Medical Genetics, University of Genoa.
- **Academic Year 2013-2022**: **Member of the Faculty Board** for the PhD Course in Pediatric Sciences - Genetics Curriculum.
- **Academic Year 2011-2012 and 2012-2013**: **Member of the Faculty Board** for the PhD Course in Clinical, Genetic and Immunology of Developmental Age Diseases (Genetics Branch), University of Genoa.
- **Academic Year 2021-2022 to present**: **Member of the Examination Committee** for the admission exam to the three-year degree courses in health professions, School of Medical and Pharmaceutical Sciences, University of Genoa.

Orientation and Third Mission Activities (main initiatives)

- Since 2012, speaker for seminar activities within the initiatives related to the "DNA Day essay contest", sponsored by the European Society of Human Genetics (ESHG) (Science Education for High Schools in collaboration with the University of Genoa), scientific referent until 2019 Prof R. Ravazzolo (University of Genoa). Since 2019, the undersigned has taken over as scientific referent. The activity is aimed at students of secondary schools and involves the organization of seminars and lessons for students in the last two years, focusing on the theme provided annually by the European Society of Human Genetics (ESHG) for the European "DNA Day Essay contest". This activity has seen the participation of several Ligurian schools and, in the last three years alone, over 200 students and science teachers challenged to engage with current genetic topics and participate in the competition. The activity was interrupted during the pandemic and resumed remotely in 2021. For the academic year 2023-2024, the initiative has been planned and approved within the "Cross-Cutting Skills and Orientation Paths (Percorsi per le Competenze Trasversali e l'Orientamento, PCTO)" for which the University of Genoa is a host institution.
- **Consultancy and scientific support activities for the FOP Italia Odv Association**, which gathers patients with Fibrodysplasia Ossificans Progressiva and their families. From 2007 to the present, the undersigned has supported the Association's activities and participated in annual conferences both as support for the organization of the scientific program and as a speaker. - 2017-2021 scientific manager of funding from the FOP Italia Association for a research grant.
- **29/11/2019**: Participation as an expert in the "In-depth seminar on rare diseases" at the Titano Theater, Republic of San Marino.
- **October 2010**: Seminar activity within the Science Festival (Festival della Scienza, Genova) 2010 edition "The Horizons of Man" - Intervention on "Rare genetic diseases: the horizon of a therapy" in collaboration with the University of Genoa (Intervention no. 141 of 29/10/2010).

Other Teaching and Seminar Activities

Academic Year 2010-2011 and 2011-2012: Supplementary teaching activity within the Second Level Master's Degree in Clinical Genetics (University of Siena, inter-university master), held by Prof R. Ravazzolo.
21-22 April 2008: Invited speaker at the "International workshop on Endocrine involvement in developmental syndromes", at the Pontifical Urbaniana University (Rome) presentation titled "The C-type natriuretic peptide and skeletal overgrowth".
June 2000: Speaker at the Symposium on "Immune system and digestive system" for the Degree Course in Biotechnologies (medical specialization) of the University of Bologna.

Academic Year 1999–2000: Support activity (exercises and tutoring) for the Human Genetics Course of the Degree Course in Medicine and Surgery (free contract Art. 33), University of Genoa, chair holder Prof. Roberto Ravazzolo.
Academic Year 1999–2000: Lecturer in Genetics (General Genetics and Medical Genetics Course) for the University Diploma Course for Physiotherapists, University of Genoa, Santa Corona Hospital, Pietra Ligure (contract under Art. 32).
May 1998: At the in-depth seminar "New genetic and biomolecular acquisitions in digestive and endocrine pathology: the role of the RET proto-oncogene" for the Degree Course in Biotechnologies of the University of Bologna, lecture titled "Activating mutations in the RET Proto-oncogene associated with HSCR + MEN2A: possible genotype-phenotype correlation".
Academic Years 1993-1997: Support teaching activity in practical exercises provided by the Human Genetics Course, Faculty of Medicine and Surgery, University of Genoa, chair holder Prof. Giovanni Romeo.
Academic Year 1993-1994: Lecturer in Genetics for the Three-Year Professional Training Course for Music Therapists at the Italian Association of Music Therapy Studies.

Other Activities
Participation in Clinical Studies on Fibrodysplasia Ossificans Progressiva (FOP)

- **2018-2020**: A Phase 2, two-part, placebo-controlled, parallel-group, double-blind study to assess the efficacy and safety of 2 dosage regimens of oral IPN60130 for the treatment of fibrodysplasia ossificans progressiva in male and female participants 5 years of age and older (D-CA-60130-452). Sponsor CLEMENTIA PHARMACEUTICALS INC. PI: Dr. Maja Di Rocco. **Role:** "study coordinator".
- **2021-2024**: Participation in the Clinical Study "A randomized, placebo-controlled study to assess the safety, tolerability, pharmacokinetics, and effects on heterotopic bone formation of REGN2477 in patients with Fibrodysplasia Ossificans Progressiva (LUMINA-1)". Sponsor: REGENERON PHARMACEUTICALS, INC. PI: Dr. Maja Di Rocco and subsequently Prof. Marco Gattorno. **Role:** study coordinator.

Support Activities for the FOP Italia OdV Association

Thanks to her activity in the field of diagnostics and research on Fibrodysplasia Ossificans Progressiva, since 2007, the year in which she first performed the molecular diagnosis of FOP in an Italian patient, the undersigned has initiated a fruitful and active collaboration with the FOP Italia OdV Association, which gathers families and patients affected by FOP (http://www.fopitalia.it/consulenza-medica.html). Over the years, the undersigned has provided scientific support to the Association, participating in the organization of annual conferences and speaking as an expert. Over the years, the conferences have gone beyond the national context and the Family Association, with the participation of the major international research groups and clinical centers, and with the increased interest of various pharmaceutical companies involved in the development of drugs for the treatment of this rare and severe genetic condition (For details and scientific programs of the various annual meetings, please consult the page dedicated to FOP Congresses available at http://www.fopitalia.it/index.html). In 2017, this intense and fruitful collaboration led to the organization of the second international "Drug Development Forum" (2nd FOP DDF 13-14/10/2017) in Italy. The Association has also funded the undersigned's research work and, since 2019, has supported, with a liberal donation, the coverage of a research grant for two years.

Organization or Participation as a Speaker at Scientific Conferences in Italy or Abroad

- **Oral presentation selected at the 29th Annual Meeting of the European Society of Human Genetics (ESHG)** held in Genoa from 17-05-1997 to 20-05-1997. The presentation was awarded the "Young investigator award for the best oral presentation" by the ESHG Scientific Committee.
- **Oral presentation selected at the "Third International Meeting on Hirschsprung disease and Related neurocristopathies"** held in Evian (France) from 05-02-1998 to 08-02-1998. The presentation focused on functional studies of RET gene variants associated with Hirschsprung's disease and thyroid tumors.
- **Invited speaker at the "International Workshop on Endocrine involvement in Developmental Syndromes"** in the "Overgrowth syndromes" session with a presentation titled "The C-type natriuretic

peptide and overgrowth" held in Rome at the Pontifical Urbaniana University from 21-04-2008 to 22-04-2008.

- **Speaker at the "First European FOP consortium meeting"** at the VU University Medical Center (Amsterdam, the Netherlands) held from 23-11-2012 to 24-11-2012 with a presentation titled "Updates on FOP research in Italy: ACVR1 expression regulation".
- **Speaker at the "Second European FOP consortium meeting"** at the VU University Medical Center (Amsterdam, the Netherlands) held from 05-12-2014 to 06-12-2014 with a presentation titled "Update on FOP research in Italy: Screening of 1200 FDA-approved molecules to identify pharmacological modulators of the ACVR1 gene expression".
- **Speaker at the "2nd Skelethon Meeting"**, national meeting of Telethon-funded researchers involved in research projects on bone-related pathologies with a presentation on "New treatment strategies for Fibrodisplasia Ossificans Progressiva (FOP)", held in Rome from 12-05-2016 to 13-05-2016.
- **Participation in the organizing committee (Planning committee)** of the second international "FOP Drug Development Forum" held in Alghero (Italy) from 13-10-2017 to 14-10-2017 and moderator of a scientific session. This international scientific event brought together major academic research groups and representatives of pharmaceutical companies involved in scientific research on Fibrodysplasia Ossificans Progressiva (FOP). The Italian research group, of which the candidate is a part, and the candidate herself, were involved in the organization of the second edition of the forum held in Italy.
- **Invited speaker at the 1st National Congress of Metabolic Osteopathies in Pediatric Age New Diagnostic and Therapeutic Frontiers**, Session dedicated to Fibrodysplasia Ossificans Progressiva titled: "New therapeutic approaches and their biological bases", held in Rome at the AOU Policlinico Umberto I – Sapienza University of Rome from 13-12-2019 to 15-12-2019.
- **Invited speaker at the XXVIII Italian Congress of Cystic Fibrosis and XVIII National Congress of the Italian Society for the Study of Cystic Fibrosis** held at the Palazzo della Gran Guardia (Verona) in the session dedicated to genetics with a presentation titled "Complex CFTR alleles", from 09-11-2022 to 12-11-2022.
- **Consultancy and scientific support activities for the FOP Italia Odv Association**, which brings together patients with Fibrodysplasia Ossificans Progressiva and their families. From 2007 to the present, the undersigned has supported the Association's activities and participated in annual conferences both as support for the organization of the scientific program and as a speaker.
- **From 2022 to present**: **Co-Scientific Director of the Clinical Genetics Refresher Course**, IRCCS Istituto Giannina Gaslini, Genoa, now in its XXII edition in 2024. The course is divided into several days dedicated to relevant topics in the field of clinical genetics, addressed with their most recent acquisitions by expert speakers from Italy and abroad. Held at the IRCCS Istituto Giannina Gaslini (Genoa), it is aimed at all professional figures working in the clinical, diagnostic, and research fields of genetic diseases and is accredited for the ECM (Continuous Medical Education) pathway.

Molecular Diagnostic Activities under the Agreement with the National Health Service (SSN)

Since 2011, she has been included in the lists of university personnel in agreement with the SSN. In Responsible for molecular diagnostic activities as Reporting Manager for the following genetic pathologies:

- **Fibrodysplasia Ossificans Progressiva (FOP, OMIM#135100, *ACVR1* gene)**: molecular diagnosis for Italian patients identified to date and, upon request, for patients referred from foreign centers.
- **Nail-Patella Syndrome (NPS, OMIM#161200, *LMX1B* gene)**;
- **EEC Syndrome (EEC type 3, OMIM #604292)** and other conditions related to the ***TP63* gene (OMIM#603273)**;
- **Congenital cataracts and anterior ocular segment dysgeneses** (complex gene panels; exome analysis).
- In collaboration with the Pediatric and Endocrinology Unit at the IRCCS Giannina Gaslini, responsible for the genetic diagnosis of **Inborn error of Metabolism, diabetes, and dysglycemias** (gene panels and exome analysis).

The undersigned also declares to be informed that the personal data collected are processed by the University of Genoa according to EU Regulation 2016/679 (GDPR – General Data Protection Regulation) and Legislative Decree 30.6.2003, n. 196 (Personal Data Protection Code), as amended by Legislative Decree 10.8.2018 n. 101.

Genoa, 15/07/2025