Dario Cocciadiferro

Curriculum Vitae

Personal Details

Date and place of birth: 15/04/1988, Catania (Italy)

Academic Education

Academic Year 2019–2022 Residency (Specialisation School) in Medical Genetics at the University of Rome "Tor Vergata"
Final grade: 50/50 cum laude.
Project title: *The diagnostic odyssey of patients with rare genetic diseases: exome analysis as a first-line test*

Academic Year 2014–2017 PhD in Experimental and Regenerative Medicine at the University of Foggia**.** Research carried out at I.R.C.C.S. Casa Sollievo della Sofferenza, San Giovanni Rotondo
Project title: *Cellular modelling to functionally dissect Kabuki Syndrome, implications for diagnostic, therapeutic and clinical care*

Academic Year 2013–2014 - Master's degree in Genetics and Molecular Biology, Faculty of Mathematical, Physical and Natural Sciences, Sapienza University of Rome.
Final grade: **110/110 cum laude.**
Thesis title: *Molecular basis of the oculo-auriculo-vertebral spectrum (OAVS).*

Academic Year 2010–2011 - Bachelor's degree in Biological Sciences, Faculty of Mathematical, Physical and Natural Sciences, University of Palermo.
Final grade: 110/110 cum laude.
Thesis title: *Assay of the anticancer molecule Kahalalide F on cisplatin-sensitive and cisplatin-resistant cell lines by flow cytometry.*

Academic Year 2008–2009 - Winner of an *Erasmus* scholarship. Academic year spent at the University of Salford, Manchester (UK)
Exams taken: Biochemistry of Disease (10 ECTS), Medical Biochemistry (10 ECTS), Medical and Public Health (10 ECTS), Human Physiology (10 ECTS).

Professional Experience

01/01/2024 – present Senior Biologist, Specialist in Medical Genetics
U.O.C. Medical Genetics Laboratory, Translational Cytogenomics Research Unit, IRCCS Bambino Gesù Children's Hospital

01/03/2018 – 31/12/2023 Research Contract at U.O.C. Medical Genetics Laboratory, Translational Cytogenomics Research Unit, IRCCS Bambino Gesù Children's Hospital

01/12/2014 – 01/12/2017 Research activity at "I.R.C.C.S. Casa Sollievo della Sofferenza", Medical Genetics Unit, San Giovanni Rotondo (IT)
Activities carried out: Characterisation of the pathogenicity of missense mutations in *KMT2D/MLL2* in patients with Kabuki Syndrome.

18/01/2016 – 18/04/2016 Research activity at "Columbia University, Institute for Cancer Genetics", New York, NY
Activities carried out: Functional characterisation of missense mutations in *KMT2D/MLL2*, including analysis of enzymatic activity through in vitro nucleosomal methylation assays.

21/11/2012 – 21/06/2014 - Internship at "Istituto C.S.S. Mendel", Rome.
Activities carried out: Research on OAVS using automated sequencing, SNP array and Real-Time PCR techniques.

01/2009 – 07/2009 - Internship at the University of Salford, Manchester.
Activities carried out: Research testing anti-cancer molecules on cervical cells.

Scholarships and Awards

2025 Oral presentation at the autism conference "From genetics to clinical care to life project", Varese (VA)

2024 Oral presentation at the international congress PediRhythm XI – World Congress of Pediatric & Congenital Heart Rhythm Disorders, Rome

2024 Oral presentation at the training course "Genomics and the new frontiers of medicine", CSS Mendel Institute, Rome

2019 Oral presentation at the course "Precision pharmacological therapy: evolving pathways and strategies", Bambino Gesù Children's Hospital, Rome

2019 Oral presentation at the PerkinElmer webinar**:** From manual to automated library prep: Implementation and analytical validation

2018 Oral presentation at the Agilent workshop – 20th National Congress of the Italian Society of Human Genetics (SIGU), Catania

2017 Scholarship winner for participation in the "Clinical Genomics and NGS – 30th course", University of Bertinoro

2017 Oral presentation at the "5th Rare Diseases Summer School", University of Zurich (CH)

2017 Scholarship winner for participation in the "5th Rare Diseases Summer School", University of Zurich

2016 Oral presentation at the 19th National Congress of the Italian Society of Human Genetics (SIGU), Turin

Scientific Contributions

Guillouet C, Agostini V, Baujat G, **Cocciadiferro D**, Pippucci T, Lesieur- Sebellin M, Georget M, Schatz U, Fauth C, Louie RJ, Rogers C, Davis JM, Konstantopoulou V, Mayr JA, Bouman A, Wilke M, VanNoy GE, England EM, Park KL, Brown K, Saenz M, Novelli A, Digilio MC, Mastromoro G, Rongioletti MCA, Piacentini G, Kaiyrzhanov R, Guliyeva S, Hasanova L, Shears D, Bhatnagar I, Stals K, Klaas O, Horvath J; University of Washington Center for Mendelian Genomics; Bouvagnet P, Witmer PD, MacCarrick G, Cisarova K, Good JM, Gorokhova S, Boute O, Smol T, Bruel AL, Patat O, Broadbent JR, Tan TY, Tan NB, Lyonnet S, Busa T, Graziano C, Amiel J, Gordon CT. *Bi-allelic MED16 variants cause a MEDopathy with intellectual disability, motor delay, and craniofacial, cardiac, and limb malformations*. Am J Hum Genet. 2025 Apr 3;112(4):829-845. doi:10.1016/j.ajhg.2025.02.016. Epub 2025 Mar 12. PMID: 40081376; PMCID: PMC12081314.

Vecchio D, Macchiaiolo M, Gonfiantini MV, Panfili FM, Petrizzelli F, Liorni N, Cortellessa F, Sinibaldi L, Rana I, Agolini E, **Cocciadiferro D,** Colantoni N, Semeraro M, Rizzo C, Deodati A, Cotugno N, Caggiano S, Verrillo E, Nucci CG, Alkan S, Saraiva JM, De Sá J, Almeida PM, Krishna J, Buonuomo PS, Martinelli D, Dionisi Vici C, Caputo V, Bartuli A, Novelli A, Mazza T. *Widening the infantile hypotonia with psychomotor retardation and characteristic Facies-1 Syndrome's clinical and molecular spectrum through NALCN in-silico structural analysis*. Front Genet. 2024 Dec 11;15:1477940. doi: 10.3389/fgene.2024.1477940. PMID: 39722796; PMCID: PMC11668739.

3: Vecchio D, Panfili FM, Macchiaiolo M, Dentici ML, Trivisano M, Medina CB, Capolino R, Salzano E, Cortellessa F, Busè M, Pantaleo A, **Cocciadiferro D**, Gonfiantini MV, Niceta M, De Dominicis A, Specchio N, Piccione M, Digilio MC, Tartaglia M, Novelli A, Bartuli A. *Molecular and clinical Insights into KMT2E-Related O'Donnell-Luria-Rodan syndrome in a novel patient cohort*. Eur J Med Genet. 2025 Feb;73:104990. doi: 10.1016/j.ejmg.2024.104990. Epub 2024 Dec 19. PMID: 39709003.

Moroni A, Pedrini E, Tremosini M, Di Cecco A, **Cocciadiferro D**, Novelli A, Santoro L, Cordiali R, Sangiorgi L, Gnoli M. Further Evidence of Early-Onset Osteoporosis and Bone Fractures as a New FGFR2 Related Phenotype. Int J Mol Sci. 2025 Apr 29;26(9):4204. doi: 10.3390/ijms26094204. PMID: 40362441;PMCID: PMC12071962.

Granata P, Zito A, **Cocciadiferro D**, Novelli A, Pessina C, Mazza T, Ferri M, Piccinelli P, Luoni C, Termine C, Fasano M, Casalone R. *Unveiling genetic insights: Array-CGH and WES discoveries in a cohort of 122 children with essential autism spectrum disorder*. BMC Genomics. 2024 Dec 10;25(1):1186. doi: 10.1186/s12864-024-11077-5. PMID: 39654053; PMCID: PMC11629504.

Lomuscio S, **Cocciadiferro D**, Petrizzelli F, Liorni N, Mazza T, Allegorico A, Ullmann N, Novelli G, Cutrera R, Novelli A. *Two Novel Biallelic Variants in the FARSA Gene: The First Italian Case and a Literature Review*. Genes (Basel). 2024 Dec 5;15(12):1573. doi: 10.3390/genes15121573. PMID: 39766840; PMCID: PMC11675362.

Cocciadiferro D, Mazza T, Vecchio D, Biagini T, Petrizzelli F, Agolini E, Villani A, Minervino

D, Martinelli D, Rizzo C, Boenzi S, Panfili FM, Buonuomo PS, Macchiaiolo M, Bartuli A, Novelli A. *Exploiting in silico structural analysis to introduce emerging genotype-phenotype correlations in DHCR24-relatedsterol biosynthesis disorder: a case study.* Front Genet. 2024 Jan 4;14:1307934. doi: 10.3389/fgene.2023.1307934. PMID: 38239854; PMCID: PMC10795535.

Guadagnolo D, Mastromoro G, Torres B, Marchionni E, di Palma F, Goldoni M, **Cocciadiferro D**, Novelli A, Bernardini L, Pizzuti A. Duplication at 19q13.32q13.33 *Segregating with Neuropsychiatric Phenotype in a Three-Generation Family: Towards the Definition of a Critical Region.* Genes (Basel). 2023 Nov 29;14(12):2157. doi: 10.3390/genes14122157. PMID: 38136979; PMCID: PMC10742575.

Marchionni E, D'Apice MR, Lupo V, Lattanzi G, Mattioli E, Lisignoli G, Gabusi E, Pepe G, Helmer Citterich M, Campione E, Nardone AM, Spitalieri P, Pucci N, **Cocciadiferro D**, Picchi E, Garaci F, Novelli A, Novelli G. *Clinical and functional characterization of COL2A1 p.Gly444Ser variant: From a fetal phenotype to a previously undisclosed postnatal phenotype.* Bone Rep. 2023 Nov 27;19:101728. doi: 10.1016/j.bonr.2023.101728. PMID: 38076483; PMCID:PMC10698255.

Murdocca M, Citro G, Centanini E, Giannini R, Latini A, Centofanti F, Piano Mortari E, **Cocciadiferro D,** Novelli A, Bernardini S, Novelli G, Sangiuolo F. *COVID-19: S-Peptide RBD 484-508 Induces IFN-γ T-Cell Response in Naïve-to-Infection and Unvaccinated Subjects with Close Contact with SARS-CoV-2-Positive Patients*. Viruses. 2023 Jun 22;15(7):1417. doi:10.3390/v15071417. PMID: 37515105; PMCID: PMC10386297.

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Granata P, **Cocciadiferro D,** Zito A, Pessina C, Bassani A, Zambonin F, NovelliA, Fasano M, Casalone R. *Whole Exome Sequencing in 16p13.11 Microdeletion Patients Reveals New Variants Through Deductive and Systems Medicine Approaches*. Front Genet. 2022 Mar 15;13:798607. doi: 10.3389/fgene.2022.798607. PMID: 35368691; PMCID: PMC8965081.

Novelli G, Liu J, Biancolella M, Alonzi T, Novelli A, Patten JJ, **Cocciadiferro D,** Agolini E, Colona VL, Rizzacasa B, Giannini R, Bigio B, Goletti D, Capobianchi MR, Grelli S, Mann J, McKee TD, Cheng K, Amanat F, Krammer F, Guarracino A, Pepe G, Tomino C, Tandjaoui-Lambiotte Y, Uzunhan Y, Tubiana S, Ghosn J; COVID Human Genetic Effort; French COVID Cohort Study Group; CoV- Contact Cohort, Notarangelo LD, Su HC, Abel L, Cobat A, Elhanan G, Grzymski JJ, Latini A, Sidhu SS, Jain S, Davey RA, Casanova JL, Wei W, Pandolfi PP. *Inhibition of HECT E3 ligases as potential therapy for COVID-19.* Cell Death Dis. 2021 Mar 24;12(4):310. doi: 10.1038/s41419-021-03513-1. PMID: 33762578; PMCID: PMC7987752.

Zito A, Lualdi M, Granata P, **Cocciadiferro D,** Novelli A, Alberio T, Casalone R, Fasano M. *Gene Set Enrichment Analysis of Interaction Networks Weighted by Node Centrality*. Front Genet. 2021 Feb 24;12:577623. doi: 10.3389/fgene.2021.577623. PMID: 33719329; PMCID: PMC7943873.

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Morlino S, Micale L, Ritelli M, Rohrbach M, Zoppi N, Vandersteen A, Mackay S, Agolini E, **Cocciadiferro D**, Sasaki E, Madeo A, Ferraris A, Reardon W, Di RoccoM, Novelli A, Grammatico P, Malfait F, Mazza T, Hakim A, Giunta C, Colombi M, Castori M. COL1-related overlap disorder: *A novel connective tissue disorder incorporating the osteogenesis imperfecta/Ehlers-Danlos syndrome overlap.* Clin Genet. 2020 Mar;97(3):396-406. doi: 10.1111/cge.13683. Epub 2019 Dec 12. PMID:31794058.

Giannantonio S, Agolini E, Scorpecci A, Anzivino R, Bellacchio E, **Cocciadiferro D**, Novelli A, Digilio MC, Marsella P. *Genetic identification andmolecular modeling characterization of a novel POU3F4 variant in two Italian deaf brothers.* Int J Pediatr Otorhinolaryngol. 2020 Feb;129:109790. doi: 10.1016/j.ijporl.2019.109790. Epub 2019 Nov 22. PMID: 31786483

Gentile M, Agolini E, **Cocciadiferro D**, Ficarella R, Ponzi E, Bellacchio E, AntonucciMF, Novelli A. *Novel exostosin-2 missense variants in a family withautosomal recessive exostosin-2-related syndrome: further evidences on the phenotype.* Clin Genet. 2019 Jan;95(1):165-171. doi: 10.1111/cge.13458. Epub 2018Oct 24. PMID: 30288735.

Marano M, Nicoletti F, Pro S, Goffredo BM, Cecchetti C, Piervincenzi E, Agolini E, **Cocciadiferro D**. *Phenytoin intoxication associated with omeprazole administration in a child with defective CYP2C9.* Eur J Clin Pharmacol. 2020 May;76(5):731-732. doi: 10.1007/s00228-019-02821-y. Epub 2020 Jan 15. PMID: 31938857.

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Giannantonio S, Agolini E, Scorpecci A, Anzivino R, Bellacchio E, **Cocciadiferro D,** Novelli A, Digilio MC, Marsella P. *Genetic identification and molecular modeling characterization of a novel POU3F4 variant in two Italian deaf brothers*. Int J Pediatr Otorhinolaryngol. 2020 Feb;129:109790. doi: 10.1016/j.ijporl.2019.109790. Epub 2019 Nov 22.

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Lucia Micale, Carmela Fusco, Andrea Fontana, Raffaela Barbano, Bartolomeo Augello, Pasquelena De Nittis, Massimiliano Copetti, Maria Teresa Pellico, Barbara Mandriani, **Dario Cocciadiferro**, Paola Parrella, Vito Michele Fazio, Lucia Maria Cecilia Dimitri, Vincenzo D'Angelo, Chiara Novielli, Lidia Larizza, Antonio Daga and Giuseppe Merla.
TRIM8 downregulation in glioma affects cell proliferation and it is associated with patients survival. ***BMC Cancer*** 15(1):470. June 2015

Carmela Fusco, Lucia Micale, Maria Teresa Pellico, Ester Valentina D'Addetta, Bartolomeo Augello, Barbara Mandriani, Pasquelena De Nittis, **Dario Cocciadiferro**, Natascia Malerba, Michele Sacco, Leopoldo Zelante and Giuseppe Merla *Genomic and Genetic Disorders Biobank*. ***Open Journal of Bioresources****, 2*:e1,DOI: http://dx.doi.org/10.5334/ojb.ag. March 2

Language Skills

Italian (native): Full command of written and spoken language.
English: Excellent command of written and spoken language.
French: Good command of written and spoken language.

Computer Skills

Advanced knowledge of the Internet and the Microsoft Office suite.
Excellent knowledge of Mac OS, Windows and Linux operating systems.

Personal Characteristics

Research, analytical and synthesis skills. Flexibility and adaptability. Commitment, tenacity and a *problem-solving* attitude. Ability to work both in a team and independently.

I authorise the processing of the personal data contained in my curriculum vitae pursuant to Art. 13 of Legislative Decree 196/2003