Curriculum Vitae

PERSONAL DATA

Name: **Donata Orioli**
Position: Senior CNR Researcher
Present work address: CNR-Istituto di Genetica Molecolare (IGM)-L.L. Cavalli Sforza
Via Abbiategrasso, 207 - 27100 Pavia (Italia)
Tel: +39-0382-546330
e-mail: "orioli@igm.cnr.it"
ORCID ID: 0000-0002-3830-3408

EDUCATION

1997: *PhD* with first-class honours in Biological Science, University of Heidelberg, Germany.
1991: Graduated cum laude in Biological Sciences, University of Pavia.

RESEARCH EXPERIENCE

Since 2021: *Senior CNR researcher* (*II level*) at the IGM CNR Pavia.
2009-2020: *CNR researcher* (*III level*) at the IGM CNR Pavia.
2008: V*isiting scientist* in Prof. J.M. Egly's laboratory at the IGBMC, Strasbourg, France.
2003-2009: *Research contracts* at IGM CNR Pavia in M. Stefanini's laboratory.
2000-2002: *Principal investigator* of a Telethon research project at the IGM CNR Pavia.
1999-2000: *EMBO Postdoc Fellow* at the IGM CNR Pavia.
1993-1997: *PhD Fellow* at the EMBL, Heidelberg. Advisor: Dr. R. Klein.

ACADEMIC ACTIVITY

Adjunct Professor of the course Molecular Genetics, Master's degree in Molecular Biology and Genetics at the University of Pavia (since 2010).
Adjunct Professor of the course Ingegneria Genetica, Master's degree in Experimental and Applied Biology at the University of Pavia (2005-2009).
Adjunct Professor at Scuola di Specialità in Genetica Applicata, University of Bologna (1999).
Supervisor of Bachelor, Master and PhD theses at the University of Pavia.
Faculty Member of the PhD program in Genetics, Molecular and Cellular Biology, University of Pavia.
Invited speaker to several Italian doctoral (PhD) and post-graduate schools**.**
External Referee for National and International (Sorbonne Université de Paris, Université de Lyon, France and Ulm University, Germany) PhD thesis.

SCIENTIFIC ACTIVITY

Principal Investigator of the following research projects:
-2024-2029: *"UV damage repair disorders, the ideal model to dissect skin cancer proneness via multiple integrated approaches"* by Fondazione AIRC.
-2019-2023: *"Basis of the different skin cancer risk in human disorders caused by mutations in the same gene, XPD"* by the Italian Association for Cancer Research (AIRC).
-2016-2019: *"UV damage repair disorders and cancer: role of the repair transcription complex TFIIH"* by the Italian Association for Cancer Research (AIRC).
-2009-2011: *"Trichothiodystrophy as a model disease to dissect the basis of TFIIH transcriptional activity"* by CNR/CNRS, 2010-2011.
-2000-2002:*"Identification of Rac3-interacting proteins during neuritogenesis"* by the Italian Telethon Foundation.

Responsible of the Research Operative Unit for the project:
2023-2025: *"Progetto di Ricerca REte di Genomica Integrata per Nuove Applicazioni in medicina di precisione – REGINA*" by the Italian Health Minister.
2015-2017: Project international de coopération scientifique (PICS) *"Défauts transcriptionnels dans des maladies associées au complexe TFIIH"*, led by Dr. Emmanuel Compe, IGBMC Strasburg, France.

Invited speaker to several national and international meetings, workshops and congresses.
R*eferee* for several international scientific Journals; national (PRIN, FIRB, Italian Health Ministry, Italian Ministry of University and Research) and international research agencies (Agence Nationale de la Recherche; The Netherlands Organisation for Scientific Research, Deutscher Akademischer Austausch Dienst).
Member of the Scientific Committee of the Italian Association Xeroderma Pigmentosum;
Committee Member of the European Xeroderma Pigmentosum Society;
Member of the Medical Advisory Board of "Amy and Friends", charity group for patients affected by Cockayne syndrome or trichothiodystrophy and their families (http://www.amyandfriends.org/contacts.htm).
Member of the Editorial Board of DNA Repair (from 2022) and *Dermato* peer-reviewed scientific journals.

RESEARCH ACTIVITY
Extensive experience and publication record in the field of molecular biology and biochemistry. In the first period of her activity, she identified the *in vivo* function of Eph tyrosine kinase receptors in nervous system development by using the mouse gene targeting approach. Later, her research has been focusing on the human syndromes xeroderma pigmentosum (XP), trichothiodystrophy (TTD), Cockayne Syndrome (CS) and UV sensitive syndrome ($UV^{S}S$) associated to defects in nucleotide excision repair (NER), a versatile DNA repair system that removes a wide range of lesions, including UV photoproducts.
Major achievements in the field of NER defective disorders include: *i)* identification of novel causative genes; *ii)* identification of biomarkers accounting for some of TTD clinical features; *iii)* identification of a novel protein complexes in which NER proteins play a role.

PUBLICATIONS
Author of **39 publications** (37 in referred peer-reviewed journals and 2 in books)
H index*: 21 (Web of Science);*
Sum of citations*: 3034 (Web of Science)*

Recent selected publications:
-Arseni L, Lanzafame M, Compe E, Fortugno P, Afonso-Barroso A, Peverali FA, Lehmann AR, Zambruno G, Egly JM, Stefanini M and Orioli D (2015) TFIIH-dependent MMP-1 overexpression in trichothiodystrophy leads to extracellular matrix alterations in patient skin. **Proc. Natl. Acad. Sci.** *112*:1499-504
-Orioli D and Dellambra E. (2018) Epigenetic Regulation of Skin Cells in Natural Aging and Premature Aging Diseases. **Cells** *7(12). pii: E268. doi: 10.3390/cells7120268.*
-Theil AF, Botta E, Raams A, Smith DEC, Mendes MI, Caligiuri G, Giachetti S, Bione S, Carriero R, Liberi G, Zardoni L, Swagemakers SMA, Salomons GS, Sarasin A, Lehmann A, van der Spek PJ, Ogi T, Hoeijmakers JHJ, Vermeulen W* and Orioli D*. (2019) Bi-allelic TARS Mutations Are Associated with Brittle Hair Phenotype. **Am J Hum Genet.** 1;105(2):434-440. *doi: 10.1016/j.ajhg.2019.06.017*
-Lanzafame M, Branca G, Landi C, Qiang M, Vaz B, Nardo T, Ferri D, Mura M, Iben S, Stefanini M, Peverali FA, Bini L and Orioli D. (2021) Cockayne syndrome group A and ferrochelatase finely tune ribosomal gene transcription and its response to UV irradiation. **Nucleic Acids Res.** *49(19)*:10911-10930. *doi: 10.1093/nar/gkab819.*
-Lombardi A, Arseni L, Carriero R, Compe E, Botta E, Ferri D, Uggè M, Biamonti G, Peverali FA, Bione S and Orioli D. (2021) Reduced levels of Prostaglandin I2 Synthase: a distinctive feature of the cancer-free trichothiodystrophy. **Proc. Natl. Acad. Sci.** *118(26)*:e2024502118. *doi: 10.1073/pnas.2024502118*
-Lanzafame M, Nardo T, Ricotti R, Pantaleoni C, D'Arrigo S, Stanzial F, Benedicenti F, Thomas MA, Stefanini M, Orioli D and Botta E (2022) TFIIH stabilization recovers the DNA repair and transcription dysfunctions in thermo-sensitive trichothiodystrophy. **Hum Mutat.** *43(12)*:2222-2233. *doi: 10.1002/humu.24488*