Curriculum Vitae et Studiorum

Nome**:** **Manuela Lanzafame**
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FORMAZIONE

11/1/2010: PhD (first class honours) in Genetic and Biomolecular Sciences, Pavia University.
5/7/2006: Master Degree (cum laude) in Cell and Molecular Biology, Catania University.
20/7/2004: Bachelor Degree (cum laude) in Biological Science, Catania University.
14/7/2001: High School Diploma (100/100), Liceo Scientifico Leonardo, Giarre (CT).

ESPERIENZA PROFESSIONALE

2022-presente: Ricercatore, Istituto di genetica Molecolare, CNR, Pavia.

2019-2020: Coordinatore studi clinici presso il reparto di Oncologia, Istituti Clinici Scientifici Maugeri, Pavia. Supervisors: Dr. A. Bernardo and Dr. A Lanza.

2017-2019: Research Assistant presso l'Istituto di Patologia, Policlinico di Basilea (Svizzera). Supervisors: Dr LM. Terracciano e S. Piscuoglio. Attività di ricerca: "Discovery and characterization of cancer biomarkers".

2010-2017: Post-Doc presso l'IGM CNR (Pavia). Supervisors: Dr. M. Stefanini e D. Orioli. Attività di ricerca: molecular and functional alterations in NER defective disorders.

2006-2009: Dottorato di ricerca in Scienze Genetiche e Molecolari finanziato dall'Università di Pavia, presso l'IGM-CNR Pavia. Supervisor: Dr. M. Stefanini. Attività di ricerca: molecular and functional alterations in the hereditary disorder trichothiodystrophy.

2004-2006: internato di tesi sperimentale per la laurea triennale in Biologia e la laurea specialistica in biologia Cellulare e Molecolare presso università degli studi di Catania (Dip. di Biologia animale). Supervisor: Dr. G. Rappazzo. Attività di ricerca: analisi di polimorfismi genomici del *locus SPANX.*

PARTECIPAZIONE A PROGETTI DI RICERCA

2024-2025 Telethon. Evaluation of a new medical device shielding the skin of XP patients from UV-light.

2024-2029 AIRC. UV damage repair disorders, the ideal model to dissect skin cancer proneness via multiple integrated approaches.

2022-2024 NUTRAGE-FOE 2021. Alterazioni metaboliche e disfunzione mitocondriale nell'invecchiamento e nelle patologie legate all'invecchiamento: La sindrome di Cockayne come modello per lo studio delle alterazioni metaboliche e mitocondriali tipiche dell'invecchiamento. https://nutrage.it/il-progetto/

2019-2023 AIRC. Basis of the different skin cancer risk in human disorders caused by mutations in the same gene XPD2.

017-2018 University of Basel Research Fund Junior Researchers (grant DMS2381). Dissecting the transcriptional effects of somatic mutations in human cancers".

2017-2018 Lega svizzera contro il cancro (Oncoswiss). Genetic determinants for progression from cirrhosis to hepatocellular carcinoma

2017-2018 Lega svizzera contro il cancro (Oncoswiss). Identification of molecular targets in hepatocellular carcinomas associated with HMGA1 overexpression

2017-2018 Krebsliga beider Basel. The feasibility of genetic profiling using plasma-derived cell-free DNA in therapy-naïve hepatocellular carcinoma patients.

2016-2019 AIRC. UV-damage repair disorders and cancer: role of the repair/transcription complex TFIIH.

2015-2017. CNRS (Francia). Dissecting the transcriptional alterations of TFIIH-related disorders.

2013-2016 AIRC. Dissecting the molecular pathways relevant for cancer proneness in UV-damage repair disorders.

2010-2011 CNR-CNRS. Trichothiodystrophy as a model disease to dissect the basis of TFIIH transcriptional activity

2009-2010 AIRC. Functional basis of cancer proneness in UV-damage repair disorders.

2006-2009 Fondazione Cariplo, Progetto Nobel. Genetic and epigenetic control of genome stability.

2006-2009 MIUS- Progetto FIRB-RBIN042YJ7. *In vitro* and *in vivo* models to investigate the molecular mechanisms of oxidative DNA damage and repair.

PREMI E RICONOSCIMENTI SCIENTIFICI

2011 - Associazione Genetica Italiana (AGI)- Menzione Premio di Dottorato AGI/Zanichelli 2010

MEMBRO DI SOCIETÀ SCIENTIFICHE

2022-presente: EACR, Eauropean Association for Cancer Reasearch

2023-today: EUROoCS, European organ-on-chip society

PUBBLICAZIONI

1. Salemi M, Bosco P, Calì F, Calogero AE, Soma PF, Galia A, **Lanzafame M**, Romano C, Vicari E, Grasso G, Siragò P, Rappazzo G (2008) *SPANX-B* and *SPANX-C* (Xq27 region) gene dosage analysis in Sicilian patients with melanoma. *Melanoma Res*, 18: 295-299.
2. Stefanini M, Botta E, **Lanzafame M**, Orioli D (2010) Trichothiodystrophy: from basic mechanisms to clinical implications. *DNA Repair* (Amst), 9: 2-10.
3. **Lanzafame M**, Vaz B, Nardo T, Botta E, Orioli D, Stefanini M (2013) From laboratory tests to functional characterisation of Cockayne syndrome. *Mech Ageing Dev*, 134: 171-179.
4. Arseni L, **Lanzafame M**, Compe E, Fortugno P, Afonso-Barroso A, Peverali FA, Lehmann AR, Zambruno G, Egly JM, Stefanini M, Orioli D (2015) TFIIH-dependent MMP-1 overexpression in trichothiodystrophy leads to extracellular matrix alterations in patient skin. *Proc Natl Acad Sci U S A*, 112: 1499-1504.
5. **Lanzafame M**, Botta E, Teson M, Fortugno P, Zambruno G, Stefanini M, Orioli D (2015) Reference Genes for Gene Expression Analysis in Proliferating and Differentiating Human Keratinocytes. *Exp Dermatol*, 24: 314-316.
6. Kuschal C, Botta E, Orioli D, Digiovanna JJ, Seneca S, Keymolen K, Tamura D, Heller E, Khan SG, Caligiuri G, **Lanzafame M**, Nardo T, Ricotti R, Stephens R, Zhao Y, Lehmann AR, Baranello L, Levens D, Kraemer KH, Stefanini M (2016) GTF2E2 mutations destabilizing the general transcription factor complex TFIIE in two DNA repair proficient trichothiodystrophy patients. *Am J Hum Genet*, 98: 627-642.
7. Pascucci B, D'Errico M, Romagnoli A, De Nuccio C, Savino M, Pietraforte D, **Lanzafame M**, Calcagnile AS, Fortini P, Baccarini S, Orioli D, Degan P, Visentin S, Stefanini M, Isidoro C, Fimia GM, Dogliotti E (2016) Overexpression of parkin rescues the defective mitochondrial phenotype and the increased apoptosis of Cockayne Syndrome A cells. *Oncotarget*. doi: 10.18632/oncotarget.9913.
8. Quagliata L, Quintavalle C, **Lanzafame M**, Matter MS, Novello C, di Tommaso L, Pressiani T, Rimassa L, Tornillo L, Roncalli M, Cillo C, Pallante P, Piscuoglio S, Ng CK, Terracciano LM (2018) High expression of HOXA13 correlates with poorly differentiated hepatocellular carcinomas and modulates sorafenib response in in vitro models. *Lab Invest*, 98:95-105.
9. Kancherla V, Abdullazade S, Matter MS, **Lanzafame M**, Quagliata L, Roma G, Hoshida Y, Terracciano LM, Ng CKY, Piscuoglio S (2018) Genomic Analysis Revealed New Oncogenic Signatures in TP53-Mutant Hepatocellular Carcinoma. *Front Genet*, 9:2.
10. **Lanzafame M**, Bianco G, Terracciano LM, Ng CKY, Piscuoglio S (2018) The Role of Long Non-Coding RNAs in Hepatocarcinogenesis. *Int J Mol Sci*, 19.
11. Calmels N, Botta E, Jia N, Fawcett H, Nardo T, Nakazawa Y, **Lanzafame M**, Moriwaki S, Sugita K, Kubota M, Obringer C, Spitz MA, Stefanini M, Laugel V, Orioli D, Ogi T, Lehmann AR (2018) Functional and clinical relevance of novel mutations in a large cohort of patients with Cockayne syndrome. *J Med Genet*, 55:329-343.
12. Ng CKY, Di Costanzo GG, Tosti N, Paradiso V, Coto-Llerena M, Roscigno G, Perrina V, Quintavalle C, Boldanova T, Wieland S, Marino-Marsilia G, **Lanzafame M**, Quagliata L, Condorelli G, Matter MS, Tortora R, Heim MH, Terracciano LM, Piscuoglio S (2018) Genetic profiling using plasma-derived cell-free DNA in therapy-naïve hepatocellular carcinoma patients: a pilot study. *Ann Oncol*, doi: 10.1093/annonc/mdy083.
13. Paradiso V, Garofoli A, Tosti N, **Lanzafame M**, Perrina V, Quagliata L, Matter MS, Wieland S, Heim MH, Piscuoglio S, Ng CKY, Terracciano LM (2018) Diagnostic targeted sequencing panel for hepatocellular carcinoma genomic screening. *J Mol Diagn*. doi: 10.1016/j.jmoldx.2018.07.003.

14. Bombaci M, Pesce E, Torri A, Carpi D, Crosti M, **Lanzafame M**, Cordiglieri C, Sinisi A, Moro M, Bernuzzi F, Gerussi A, Geginat J, Muratori L, Terracciano LM, Invernizzi P, Abrignani S, Grifantini R (2019) Novel biomarkers for primary biliary cholangitis to improve diagnosis and understand underlying regulatory mechanisms. *Liver Int*. doi: 10.1111/liv.14128.

15. Ghosh S, Guimaraes JC, **Lanzafame M**, Schmidt A, Syed AP, Dimitriades B, Börsch A, Ghosh S, Mittal N, Montavon T, Correia AL, Danner J, Meister G, Terracciano LM, Pfeffer S, Piscuoglio S, Zavolan M. (2020) Prevention of dsRNA-induced interferon signaling by AGO1x is linked to breast cancer cell proliferation. *EMBO J*. 15;39:e103922. doi: 10.15252/embj.2019103922.

16. Witzigmann D, Grossen P, Quintavalle C, **Lanzafame M**, Schenk SH, Tran XT, Englinger B, Hauswirth P, Grünig D, van Schoonhoven S, Krähenbühl S, Terracciano LM, Berger W, Piscuoglio S, Quagliata L, Rommelaere J, Nüesch JPF, Huwyler J. (2021) Non-viral gene delivery of the oncotoxic protein NS1 for treatment of hepatocellular carcinoma. *J Control Release*. 10;334:138-152. doi: 10.1016/j.jconrel.2021.04.023.

17. Tlili H, Macovei A, Buonocore D, **Lanzafame M**, Najjaa H, Lombardi A, Pagano A, Dossena M, Verri M, Arfa AB, Neffati M, Doria E. The polyphenol/saponin-rich Rhus tripartita extract has an apoptotic effect on THP-1 cells through the PI3K/AKT/mTOR signaling pathway (2021) *BMC Complement Med Ther*. 21:153. doi: 10.1186/s12906-021-03328-9.

18. Montazeri H, Coto-Llerena M, Bianco G, Zangene E, Taha-Mehlitz S, Paradiso V, Srivatsa S, de Weck A, Roma G, **Lanzafame M**, Bolli M, Beerenwinkel N, von Flüe M, Terracciano LM, Piscuoglio S, Ng CKY. (2021) Systematic identification of novel cancer genes through analysis of deep shRNA perturbation screens. *Nucleic Acids Res*. 7;49:8488-8504. doi: 10.1093/nar/gkab627.

19. **Lanzafame M**, Branca G, Landi C, Qiang M, Vaz B, Nardo T, Ferri D, Mura M, Iben S, Stefanini M, Peverali FA, Bini L, Orioli D. Cockayne syndrome group A and ferrochelatase finely tune ribosomal gene transcription and its response to UV irradiation. (2021) *Nucleic Acids Res*. 49:10911-10930. doi: 10.1093/nar/gkab819.

20. **Lanzafame M**, Nardo T, Ricotti R, Pantaleoni C, D'Arrigo S, Stanzial F, Benedicenti F, Thomas MA, Stefanini M, Orioli D, Botta E. TFIIH stabilization recovers the DNA repair and transcription dysfunctions in thermo-sensitive trichothiodystrophy. Hum Mutat. 2022 Oct 19. doi: 10.1002/humu.24488.