European School of Molecular Medicine

2025 summer call PhD selections

AVAILABLE POSITIONS

Principal Investigator	Graziano Pesole
Affiliation	University of Bari
Title of the proposed project:	Charting Tissue-Specific Transcriptome and Epitranscriptome in Humans Through Long-Read Sequencing Data
Short description of the project	Eukaryotic organisms exhibit complex and dynamic transcriptomes whose regulation is essential for all cellular processes and for maintaining homeostatic. Technological advances in RNA deep sequencing have revolutionized gene expression analysis, offering an unbiased approach to gene detection and quantification, and enabling the discovery of novel isoforms, alternative splicing events, and fusion transcripts. Although short-read sequencing is the de facto standard technology for gene expression profiling, it cannot resolve full-length transcripts or complex isoforms. In contrast, long-read sequencing technologies, such as PacBio and Oxford Nanopore (ONT), allow the profiling of full-length transcripts. However, a comprehensive map of full-length transcriptomes across human tissues is still lacking. In addition, eukaryotic RNAs are decorated with hundreds of RNA chemical modifications, collectively referred to as the epitranscriptome, which can modulate gene expression and regulation. ONT direct RNA sequencing enables the detection of unique signatures of these modifications at level of individual RNA molecules, although their computational identification remains a challenge. Here, we aim to sequence full-length transcripts from diverse human tissues using both PacBio and ONT direct RNA sequencing to generate a comprehensive transcriptome and epitranscriptome map. This effort will pave the way for deeper functional insights by charting the repertoire of full-length isoforms is crucial for investigating tissue-specific transcriptional dynamics and facilitating systematic investigation of tissue-specific transcriptional dynamics, the distribution and location of RNA modifications, and their functional interplay.
Main research area for the project	Computational Biology
Second research area for the project	
3 key words for project	High-throughput diretct RNA sequencing, transcriptome profiling, RNA modifications
Main topic/s of the lab	Bioinformatics, Comparative Genomics, High-throughput second and third generation sequencing data, Integrative omics
Short description of the lab activity	The Graziano Pesole group has been a pioneer in the fields of Bioinformatics, Comparative Genomics, and Molecular Evolution in Italy since the early 1990s. The current research activities are focused on the development and application of computational approaches for the management and analysis of high-throughput sequencing data, including single-cell and spatial resolution technologies. Key areas of focus include: i) Genome assembly and annotation;

	ii) Transcriptome analysis of both coding (mRNAs) and non-coding RNAs (miRNAs, lincRNAs, circRNAs), including the identification of novel splicing isoforms, for investigating gene expression in physiological and pathological conditions, as well as for discovering diagnostic and prognostic molecular biomarkers; iii) Identification and functional characterization of pathogenic mutations; iv) Genome-wide data analyses for epigenomic and epitranscriptomics profiling in normal and disease conditions; v) Metagenomic studies to characterize the microbial composition of clinical and environmental samples and to assess their functional roles. The group has developed several internationally recognized specialized databases and has designed numerous widely used software tools and algorithms for bioinformatics analyses.
Recent bibliography	**1:** Cox SN, Varvara AS, Pesole G. MitSorter: a standalone tool for accurate discrimination of mtDNA and NuMT ONT reads based on differential methylation. Bioinform Adv. 2025 Jul 10;5(1):vbaf135. doi: 10.1093/bioadv/vbaf135. PMID: 40688360; PMCID: PMC12275464. **2:** Salsi V, Losi F, Fosso B, Ferrarini M, Pini S, Manfredi M, Vattemi G, Mongini T, Maggi L, Pesole G, Henras AK, Kaufman PD, McStay B, Tupler R. Nucleolar FRG2 lncRNAs inhibit rRNA transcription and cytoplasmic translation, linking FSHD to dysregulation of muscle-specific protein synthesis. Nucleic Acids Res. 2025 Jul 8;53(13):gkaf643. doi: 10.1093/nar/gkaf643. PMID: 40637237; PMCID: PMC12242771. **3:** Fonzino A, Mazzacuva PL, Handen A, Silvestris DA, Arnold A, Pecori R, Pesole G, Picardi E. REDInet: a temporal convolutional network-based classifier for A-to-I RNA editing detection harnessing million known events. Brief Bioinform. 2025 Mar 4;26(2):bbaf107. doi: 10.1093/bib/bbaf107. PMID: 40112338; PMCID: PMC11924403. **4:** D'Addabbo P, Cohen-Fultheim R, Twersky I, Fonzino A, Silvestris DA, Prakash A, Mazzacuva PL, Vizcaino JA, Green A, Sweeney B, Yates A, Lussi Y, Luo J, Martin MJ, Eisenberg E, Levanon EY, Pesole G, Picardi E. REDIportal: toward na integrated view of the A-to-I editing. Nucleic Acids Res. 2025 Jan 6;53(D1):D233-D242. doi: 10.1093/nar/gkae1083. PMID: 39588754; PMCID: PMC11701558. **5:** Grasso L, Fonzino A, Manzari C, Leonardi T, Picardi E, Gissi C, Lazzaro F, Pesole G, Muzi-Falconi M. Detection of ribonucleotides embedded in DNA by Nanopore sequencing. Commun Biol. 2024 Apr 23;7(1):491. doi: 10.1038/s42003-024-06077-w. PMID: 38654143; PMCID: PMC11039623.
Group composition	The group consists of approximately 40 members, including faculty staff, postdoctoral researchers, PhD students, scholars, and technicians from both the University of Bari and the National Research Council (Consiglio Nazionale delle Ricerche, CNR).

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