DOCTORAL POSITION
ENTRY-DM: Marie Skłodowska-Curie Doctoral Networks (MSCA DN)

The complexity of DM repeat expansions: new challenges in developing personalised molecular therapeutics

UNIVERSITA DEGLI STUDI DI ROMA TOR VERGATA
Medical-surgical biotechnologies & translational medicine

Introduction: The objective of Doctoral Networks (MSCA DN) is to implement doctoral programmes through partnerships with organisations from different sectors across Europe and beyond to train highly skilled doctoral candidates, stimulate their creativity, enhance their innovation capacities and boost their employability in the long term. The Laboratory of Medical Genetics of the Biomedicine and Prevention Department of the University of Rome Tor Vergata is a distinctive hub for multidisciplinary collaboration, innovation, and excellence, dedicated to advancing cutting-edge research and education, with the aim to train highly qualified researchers in the use of advanced methodologies in the Genetics sector.

Network Description: ENTRY-DM is an interdisciplinary training and research programme focused on RNA-based therapeutics for myotonic dystrophy (DM). It offers 14 fully funded positions across top European institutions, integrating fundamental science, translational medicine, and clinical applications. The network integrates academic leaders, biotech experts, and patient advocates to develop disease models, optimise antisense oligonucleotide (ASO) therapies, and identify clinical biomarkers. Doctoral candidates will receive advanced training in genomics, bioinformatics, stem cell research, bioengineering, and neuropsychology. With host institutions in France, Spain, Italy, the Netherlands, Germany, and Poland, ENTRY-DM provides exceptional mobility, cross-sector training, and world-class supervision. It will equip doctoral candidates with the expertise to drive future breakthroughs in RNA therapeutics for rare disease treatment.

Project: The underlying mechanisms of genotype-phenotype correlation in DMs rare still poorly known, especially for DM2 patients. Moreover, about 10% of DM1 patients carry CTG expansions interrupted by naturally occurring variant repeat (VR) motifs, which are considered major cis-modifiers of molecular and phenotypic consequences. Heterogeneity across tissues, somatic instability, and the relative technical difficulty of measuring repeat length distributions using PCR-based methods currently hamper the possibility to establish and refine these crucial correlations. This project will exploit a diagnostic protocol based on a combination of NGS and PCR-based methods enabling the full characterization of pure and interrupted DM expansions. DC2 will correlate the length, structure and methylation pattern of the DM loci with the clinical phenotype in a selected cohort of DM patients, to identify relevant genotype-phenotype correlations. Moreover, DC2 will produce cell lines overexpressing different motifs of C/CTG interruptions, as tools to study the molecular and functional impact of VRs on RNA foci dynamics and develop personalized molecular therapeutic strategies.

Candidate's Profile: We are looking for a highly motivated and skilled DC with a solid background in Medical Genetics and Cellular Biology to join our research project aimed to explore the structural, molecular, and functional aspects of expanded alleles in DM patients. The candidate should hold Master's degree (or equivalent) in Genetics, Biology, Molecular Biology, Biomedicine, or a related field. A strong interest in genetic diseases and motivation to contribute to the development of therapeutic strategies for patients is essential. Prior experience in key molecular genetics techniques, including DNA/RNA extraction, PCR, RT-PCR, genotyping and NGS data analysis is required. Hands-on experience in cell culture and plasmids construction is highly desirable. The ideal candidate should demonstrate ability to plan and conduct experiments independently, analyse data, and draw conclusions. Excellent communication and teamwork skills are necessary to work in an international and interdisciplinary context, including academic researchers and physicians.

Offer Starting Date: 1 Sep 2025

Application Deadline: 30 May 2025 - 12:30 (Europe/Paris)

For more information, please contact:
Associate Professor, Supervisor, Annalisa Botta: botta@med.uniroma2.it

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Università di Roma Tor Vergata
DIPARTIMENTO DI BIOMEDICINA E PREVENZIONE

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