



PHD COURSE OF NATIONAL INTEREST IN

PRECISION MEDICINE

SITE



**Università
degli Studi
di Palermo**

COORDINATOR

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PIANO NAZIONALE
DI RIPRESA E RESILIENZA

THE CONTEXT OF PRECISION MEDICINE

In the Italian context of innovation in the Health sector, the systematisation of major skills and Key Enabling Technologies at the service of National and European objectives represents one of the major challenges for growth. The investments of the PNRR (Mission 4 Component 2) aimed at creating Research and Innovation process chains - ranging from basic research to technology transfer - which involve networks between Universities, Research Centers and Organizations and Companies, and which have identified research in the field of Innovative Diagnoses and Therapies for Precision Medicine among the priority objectives of the country, have been included in this framework.

In fact, complex diseases, including monogenic diseases (rare diseases), polygenic diseases (cardiovascular and metabolic diseases) and cancer are the main causes of mortality in the world, for which an incidence and a mortality rate are estimated to increase in the coming decades. The etiopathogenetic and prognostic complexity and heterogeneity of these pathologies make the application of precision medicine imperative, which deals with individualized paths for diagnosis and treatment, in order to obtain an optimal clinical result. Therefore, using a holistic and multidisciplinary

approach, supported by the use of transversal KETs and going beyond the classic sectoral training paradigm, it is now necessary to create Higher Education courses, which allow, among other things, to study the molecular mechanisms of noncommunicable diseases, identifying potentially prognostic and pathogenic disease biomarkers to develop innovative strategies for personalized measures of prevention, prediction, diagnosis, monitoring and for precision therapeutic planning.

This approach requires, alongside bio-medical skills, to enhance training in the field of enabling technologies 4.0 applied to health, which make it possible to train new professionals capable of applying the skills learned for the acquisition of broad-spectrum omics data, the complex processing of big data, the development of advanced predictive experimental/computational models, the study of the most advanced methods of precision diagnostics, the development of innovative and personalized therapies, the design of cutting-edge devices of interest for the market, the development of prevention strategies and clinical validation of new diagnostic/therapeutic solutions.

Under this premise, it is proposed the establishment of a Doctorate of National Interest (DIN) in Precision Medicine with administrative headquarters at the University of Palermo which is also the proponent of the project “HE-ALITALIA - Health Extended Alliance for Innovative Therapies, Advanced Lab-research, and Integrated Approaches of Precision Medicine” funded under the PNRR (Mission 4 “Education and Research” Component 2 “From Research to Business”) which represents the first Italian multidisciplinary research network of Universities, IRCCS and businesses to support translational research for advanced diagnosis and therapies in the fight against cardiovascular, metabolic, rare diseases and cancer.

Through the National Doctorate in Precision Medicine, PhD students will be offered the opportunity to operate in a highly collaborative context rich in tools and utilities such as:

- An already available platform of shareable clinical and instrumental data and biological samples of populations, with related biobanks, followed for years and/or cohorts of patients affected by complex polygenic pathologies;
- A nationwide network of cell factories and biobanks, accessible through collaboration with HEAL ITALIA project partners and with universities participating in the National Doctorate, which includes the collection of diagnostic images according to standardized protocols to allow the creation of prospective studies for the identification of prognostic biomarkers, the screening of off-label drugs and the prevention of disease risk.

11 UNIVERSITIES INVOLVED

UNIVERSITY OF ROME “LA SAPIENZA”

Radiological, oncological
and pathological anatomy sciences

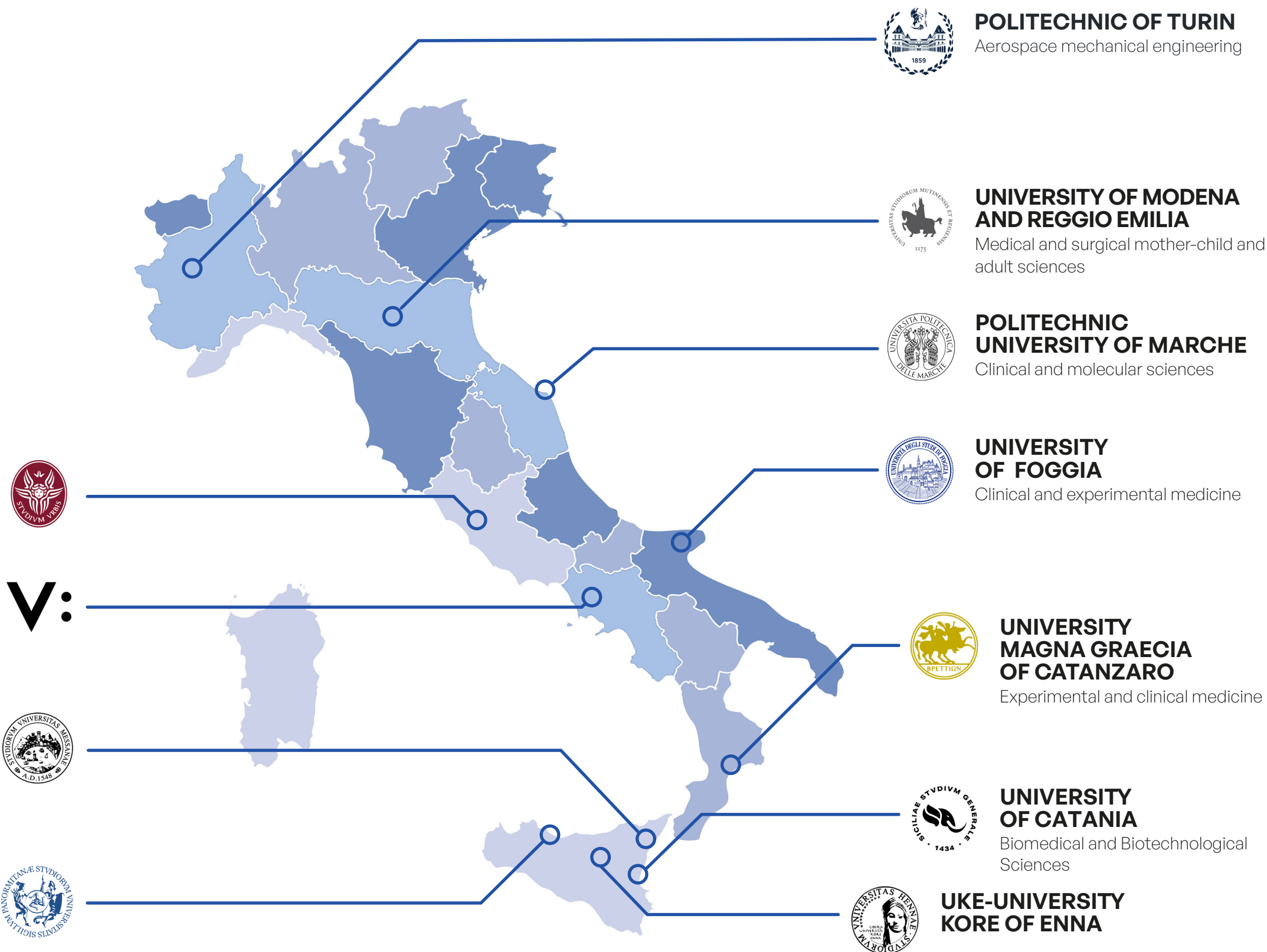
UNIVERSITY OF CAMPANIA LUIGI VANVITELLI

Precision medicine

UNIVERSITY OF MESSINA

UNIVERSITY OF PALERMO

Surgical, Oncological and
Stomatological Disciplines





AZIENDA OSPEDALIERA UNIVERSITARIA POLICLINICO PAOLO GIACCONE

The “Paolo Giaccone” Policlinico University Hospital in Palermo represents a center of excellence in health care, biomedical research and academic training.

Closely integrated with the University of Palermo, it actively contributes to teaching and specialization in the medical and surgical fields. The center offers a wide range of high-complexity clinical and care services, with operating units dedicated to disciplines such as oncology, cardiology, neurology and advanced surgery.



CEINGE BIOTECNOLOGIE AVANZATE Franco Salvatore S.c.a. r.l.

The CEINGE - Advanced Biotechnology “Franco Salvatore” S.c.a. r.l. in Naples is a center of excellence for research in the field of biotechnology applied to medicine. Specialized in genomics, proteomics and molecular biology, CEINGE conducts innovative studies on genetic, oncological and infectious diseases, with a strong focus on translational research.



Laboratorio CRIMM, AOU Careggi

The CRIMM Laboratory (Center for Research and Innovation in Precision Medicine) at AOU Careggi in Florence is a reference center for translational research and innovation in personalized medicine. CRIMM is focused on the development and application of advanced technologies for the diagnosis and treatment of complex diseases, with particular emphasis on oncology, rare diseases and pharmacogenomics.



CNR Napoli, Istituto di Genetica e Biofisica Adriano Buzzati Traverso

“Adriano Buzzati-Traverso” Institute of Genetics and Biophysics (IGB) of the CNR in Naples is a center of excellence for research in genetics, molecular biology and biophysics.



A.R.N.A.S. Ospedali Civico Di Cristina Benfratelli

The A.R.N.A.S. (Azienda di Rilievo Nazionale e di Alta Specializzazione) Ospedali Civico Di Cristina Benfratelli in Palermo is one of the main hospital facilities in Sicily, with a leading role in high-complexity healthcare, clinical research and specialist training.



Ospedale P. Pederzoli

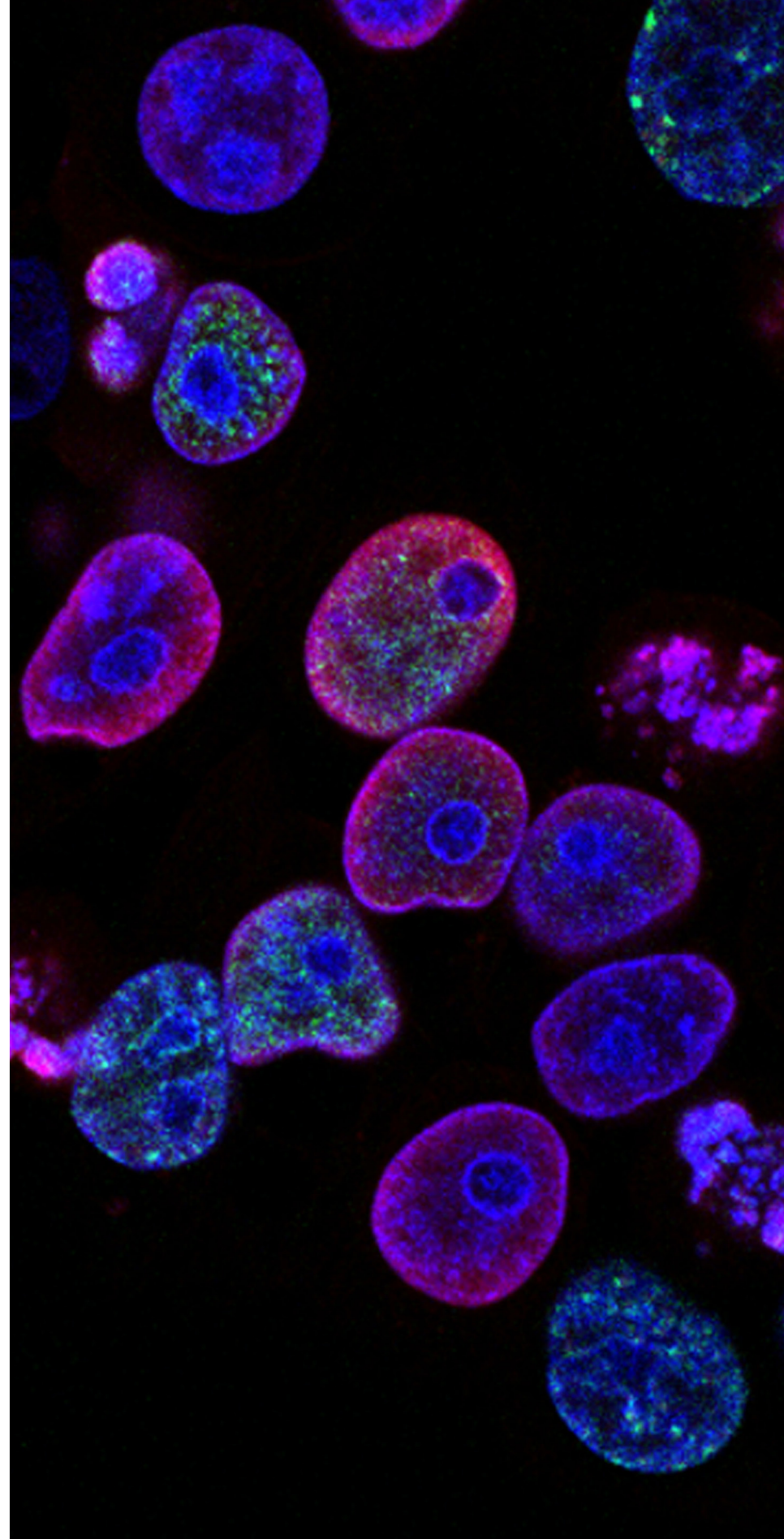
The P. Pederzoli Hospital in Peschiera del Garda is an important health facility, recognised for the high quality of its medical and surgical care. Founded as a privately run hospital, it offers a wide range of specialist services, with particular excellence in areas such as oncology, general and minimally invasive surgery, cardiology and orthopaedics.

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CURRICULA

1. Oncology

The Precision Medicine in Oncology course focuses on applying personalized approaches to the diagnosis, management and treatment of cancer patients. Precision medicine represents a significant breakthrough in oncology, as it is based on the identification of molecular, genetic and environmental characteristics of tumors and of the patients themselves, in order to provide targeted and personalized therapies. The course offers in-depth education on different types of cancers, such as breast, lung, colon, prostate cancer and many more. Students will acquire advanced knowledge on the biology of cancer, on the molecular mechanisms involved in its origin and progression, as well as on innovative diagnostic and therapeutic methodologies. A central aspect of the course concerns the analysis of the genetic profile of tumors to identify the genetic mutations and the molecular alterations that determine their development and progression. Students will be trained in the use of DNA sequencing techniques and advanced genetic analysis to identify oncogenic mutations and genetic abnormalities that can influence response to treatments. The course addresses the importance of profile analysis of gene expression and epigenetic alterations in tumors. Students will be introduced to RNA microarray and sequencing technologies, to analysis methodologies of data to identify gene expression patterns characteristic of tumors and to identify prognostic biomarkers and useful predictors for personalizing treatments.



2. Rare Diseases

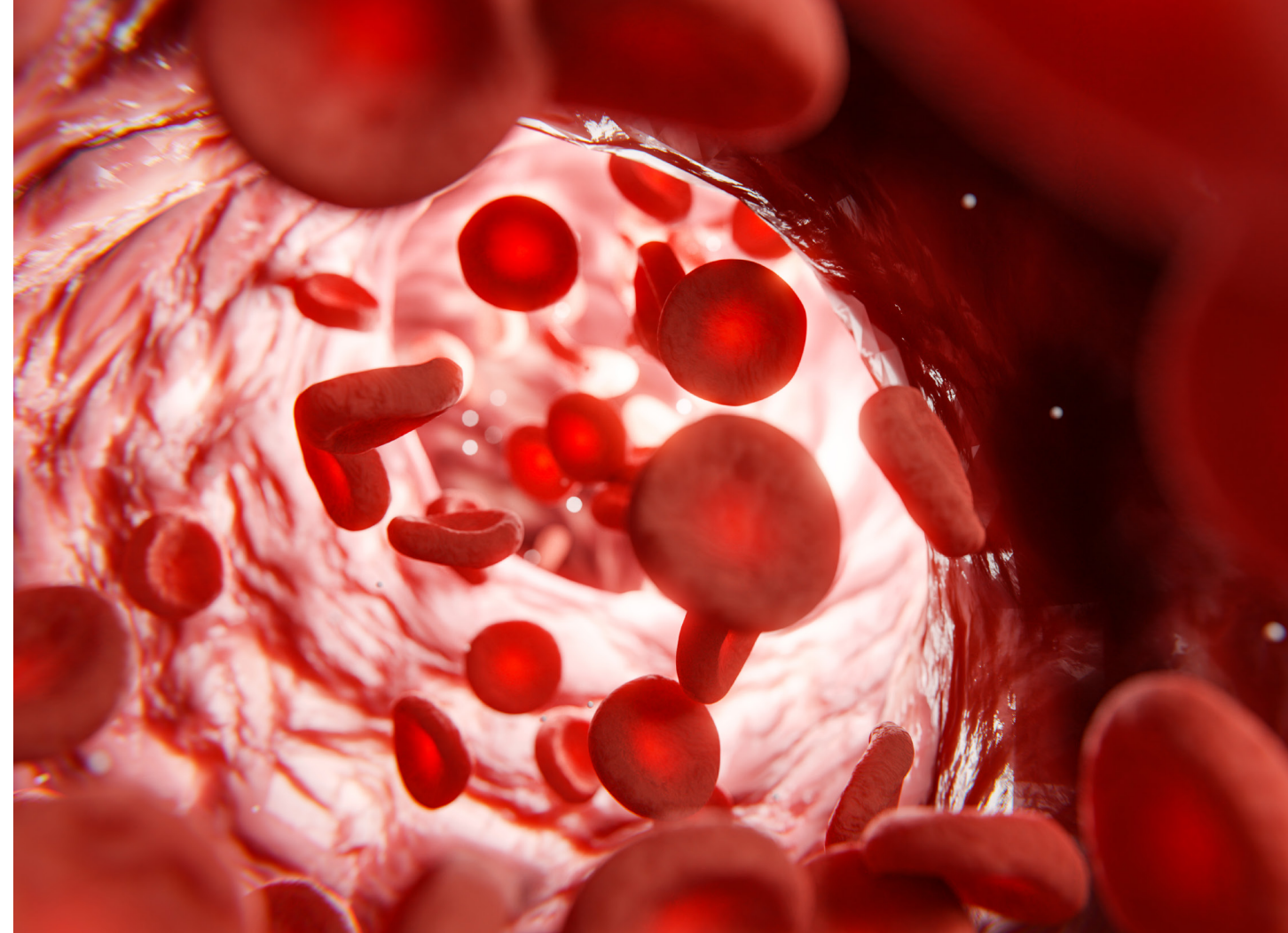
The PhD course in Precision Medicine on the topic “Rare Diseases” aims to provide advanced training on the challenges and opportunities related to the diagnosis, treatment and management of rare diseases. Rare diseases represent a diverse set of medical conditions, often genetic in nature, that affect a limited number of individuals in the population. Precision medicine offers a promising approach to addressing these complex and often debilitating diseases. One of the fundamental aspects of the course concerns the diagnosis of rare diseases. PhD students will be trained in the use of advanced DNA sequencing technologies, such as full exome or genome sequencing, to identify genetic variants responsible for rare diseases. They will acquire knowledge about strategies for analyzing and interpreting genetic data, as well as using genetic databases to compare the results of patients with known genetic variants. Another central theme concerns the development of personalized therapies for rare diseases. Students will explore the new frontiers of gene therapy, cell therapy and the use of targeted drugs to treat specific rare diseases. They will be trained in the identification of specific therapeutic targets and the use of precision medicine approaches to identify patients who can benefit from these innovative therapies. Furthermore, the course will address the clinical and management aspects of rare diseases. PhD students will learn to recognize the signs and symptoms of rare diseases and adopt a multidisciplinary approach in patient management. The challenges of collecting and sharing clinical data among specialists will be explored in order to facilitate timely diagnosis and coordinated management of rare diseases.

3. Metabolic Diseases

The Precision Medicine in Metabolic Diseases course focuses on the application of personalized approaches for the diagnosis, management and prevention of metabolic diseases. This field is based on the identification of genetic, epigenetic and environmental factors that influence individual metabolism, in order to develop targeted and personalized interventions. The course provides a solid foundation of knowledge on various metabolic diseases, such as diabetes mellitus, obesity, dyslipidemia and hereditary metabolic diseases. Students will gain an in-depth understanding of the genetic and molecular basis of these diseases and their impact on human metabolism. A fundamental aspect of the course concerns the importance of analyzing the individual genetic profile to identify genetic predispositions to metabolic diseases and to personalize prevention and treatment strategies. PhD students will learn the methods of DNA sequencing and analysis of genetic variants, as well as techniques for assessing genetic risk and interpreting results. Furthermore, the course will address the application of precision medicine approaches in the field of nutrigenomics and pharmacogenomics. PhD students will be trained to identify interactions between diet, nutrients and the individual genome in order to develop personalized food plans for the management of metabolic diseases. They will also be introduced to the concepts of pharmacogenomics, which studies the interaction between drugs and individual genes, and their influence on drug response and susceptibility to adverse effects.

4. Cardiovascular Diseases

The Precision Medicine in Cardiovascular Diseases course is designed to provide advanced training in the field of personalized medicine applied to cardiovascular pathologies. Precision medicine represents a significant evolution in the management of diseases, especially cardiovascular diseases, as it aims to identify risk factors and specific characteristics of patients to personalize diagnosis, treatment and prevention. The course focuses on the application of advanced molecular, genomic and imaging approaches to understand the genetic basis, molecular and physiological aspects of cardiovascular disease. Stu-



dents will acquire in-depth knowledge of various cardiovascular diseases, including myocardial infarction, heart failure, arterial hypertension and cardiac arrhythmias, and their connections with individual genetic makeup. The key concepts of pharmacogenomics, which studies the interaction between drugs and individual genes, and their impact on drug response and susceptibility to adverse effects, will be introduced. Students will be trained to interpret genomic and molecular data and to use this information in choosing the most appropriate pharmacological treatments for each patient. Furthermore, the course will address the importance of advanced imaging technologies, such as echocardiography, cardiac magnetic resonance and computed tomography, in the evaluation of cardiovascular diseases. Students will learn how to acquire and interpret imaging data for accurate diagnosis and prognostic evaluation of patients with cardiovascular disease.

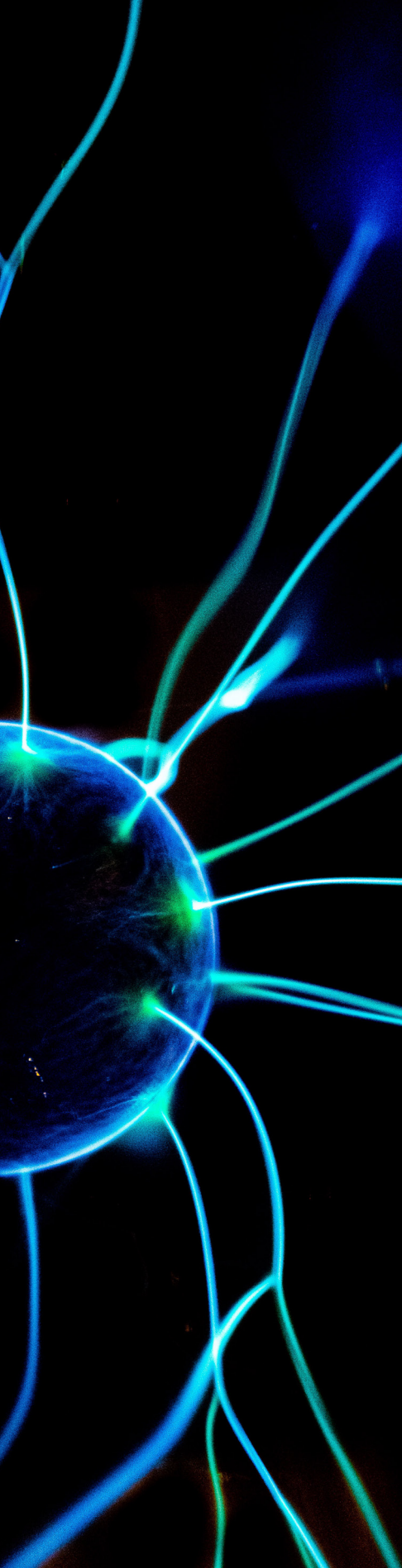


5. Neurodegenerative Diseases

The PhD course in Precision Medicine on the topic “Precision Medicine in Neurodegenerative Diseases” offers specialized training to understand and address the challenges associated with neurodegenerative diseases, such as Alzheimer’s, Parkinson’s disease and amyotrophic lateral sclerosis (ALS). These diseases represent a growing problem in the context of an aging population and require a personalized approach for diagnosis, prognosis and treatment. One of the main objectives of the course is to provide a solid foundation of knowledge on the genetic and molecular basis of neurodegenerative diseases. PhD students will acquire an in-depth understanding of the pathogenic mechanisms leading to these diseases, including aspects related to the accumulation of abnormal proteins, inflammation and cellular damage. The main genes associated with neurodegenerative diseases and the interactions between genetic and environmental factors will also be introduced. Another fundamental aspect of the course concerns the use of precision medicine approaches for the early diagnosis and prognosis of neurodegenerative diseases. PhD students will learn to apply advanced brain imaging techniques, such as functional magnetic resonance imaging (fMRI) and positron emission tomography (PET), to identify early signs of these diseases and assess disease progression over time. In addition, they will be trained in the use of biomarkers in blood and cerebrospinal fluid for more accurate diagnosis and personalized assessment of patients. Another relevant issue concerns the development of personalized therapies for neurodegenerative diseases. PhD students will explore the new frontiers of gene therapy, cell therapy and the use of targeted drugs to treat specific subtypes of neurodegenerative diseases.

6. Digital Health

The PhD program in Precision Medicine on the topic of “Digital Health” has been designed to provide advanced and specialized training on the intersection between digital technology and healthcare. In an era where technological innovation is radically transforming the health sector, it is crucial to understand the role and potential of digital health in providing personalized and improved care. Digital health refers to the use of digital technologies, such as electronics, information technology, communications and artificial intelligence, to manage and optimize health and well-being. In the course, students will explore the different applications of digital health, including smart medical devices, mobile health applications, remote monitoring systems, artificial intelligence for health data analysis and much more. A fundamental aspect of the course concerns the analysis of digital health data. PhD students will learn to use advanced data analysis methods to extract relevant insights from the large volumes of data generated by digital technologies. This analysis can help identify patterns, trends and correlations that are significant for early diagnosis of diseases, prediction of clinical outcomes and identification of personalized therapies. PhD students will be trained on the use of algorithms and machine learning techniques for data processing and the creation of predictive models. Another relevant issue concerns the security and privacy of digital health data. With the rise of digital technologies, it is essential to protect personal information and ensure the security of patient data. PhD students will be trained on health data privacy and security policies and regulations, as well as strategies to mitigate the risks associated with the interoperability of digital systems.



7. Materials and technology in precision medicine

Precision medicine is an emerging field that relies on personalizing medical treatments based on the individual characteristics of patients. In a world where human health is influenced by multiple factors, the one-size-fits-all approach is no longer enough. The course aims to provide advanced training on how innovative materials and technologies can be used to improve clinical outcomes and promote personalized medicine. A crucial aspect of precision medicine is the use of advanced biomaterials for the development of medical devices, implants and diagnostic tools. These materials must be safe, biocompatible and able to interact specifically with the human body. Throughout the course, students will explore the latest breakthroughs in biomaterial design and synthesis, as well as their applications in precision medicine. The physical and chemical properties of materials will be examined, as well as their interaction with biological tissues, in order to develop innovative solutions for the diagnosis and treatment of diseases. Advanced technologies play a vital role in precision medicine. PhD students will acquire in-depth knowledge of the latest imaging technologies, such as high-resolution computed tomography, magnetic resonance and advanced ultrasound. They will learn how to use these technologies to obtain high precision and resolution images, enabling more accurate diagnosis and personalized assessment of patients. Another important topic covered in the course concerns DNA sequencing technologies and genomic analysis. PhD students will learn how to interpret genetic data and identify genetic variants that can influence disease susceptibility and drug response.

11 COURSES

1. Oncology and molecular diagnostics

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Provide an in-depth understanding of the fundamental principles of oncology, including mechanisms of cancer formation and the molecular characteristics associated with different types of tumors.

2. Drug delivery systems based on extracellular vesicles

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Understand the principles and applications of extracellular vesicles as drug delivery vectors, explore extracellular vesicle isolation, characterization and engineering methodologies, understand the mechanisms of drug storage and release by the extracellular vesicles.

3. Privacy and legal management of health data

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Provide an overview of the laws and regulations governing privacy in the healthcare context, understand the fundamental principles of privacy and key concepts such as informed consent, explore the specific risks and challenges related to managing healthcare data, understand cybersecurity measures to protect healthcare data.

4. Diagnostic and therapeutic aspects of genetic diseases mediated by nonsense mutations

Duration	Objectives of the course
First second and third year, 8 hours (an intensive day of training)	Understand the concept of nonsense mutations and their impact on genetic diseases, familiarize yourself with the advanced diagnostic techniques used to identify nonsense mutations. Examine treatment options available for genetic nonsense disorders. Explore emerging innovative therapies used in precision medicine to address nonsense genetic diseases.

5. Mediterranean diet in the prevention and treatment of metabolic and chronic-degenerative pathologies

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Deepen the knowledge of the Mediterranean diet as a preventive and therapeutic tool for metabolic and chronic-degenerative diseases, examine the scientific evidence that supports the effectiveness of the Mediterranean diet in prevention and management of these pathologies, to provide practical tools for the adoption and integration of the Mediterranean diet in the clinical context.

6. Identification of specific biomarkers of response to innovative therapies: development and validation

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Provide an in-depth understanding of the importance of biomarkers in precision medicine and in the assessment of response to innovative therapies, examine the strategies and techniques used for the identification and development of specific biomarkers of response to innovative therapies, delve into biomarker validation methodologies and practical considerations in their clinical implementation.

7. Inorganic/organic nanomaterials for biomedical applications

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Provide in-depth understanding of inorganic and organic nanomaterials and of their applications in the biomedical field, explore the unique properties of nanomaterials and their potential in the field of Precision Medicine, analyze the different synthesis methodologies and characterization of nanomaterials for biomedical applications to delve into the specific applications of inorganic and organic nanomaterials in diagnosis, therapy and biomedical imaging.

8. Identification and validation of diagnostic biomarkers and prognostic in cardiovascular disease

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Deepen the knowledge of the biomarkers used for the diagnosis and prognosis of cardiovascular diseases, examine the methodologies and the techniques used for identification and validation of cardiovascular biomarkers, analyze scientific and clinical evidence supporting the use of cardiovascular biomarkers in medical practice, provide practical skills for the application of cardiovascular biomarkers in precision medicine.

9. Genetic basis in mitochondrial diseases and identification of the genes underlying neurodegenerative diseases

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Provide an in-depth understanding of the genetic basis of mitochondrial diseases and neurodegenerative diseases, explore the pathogenetic mechanisms of mitochondrial diseases and neurodegenerative diseases, analyze the techniques used for the identification of genes involved in these pathologies, deepen the relationship between mitochondrial diseases and neurodegenerative diseases and their impact on precision medicine.

10. Bioprinting, Microfluidics and Alternative Models to Animal Models: Integration of Technologies for the Advancement of Biomedical

Duration	Objectives of the course
First second and third year, 3 modules from 8 hours	Understand the principles and applications of bioprinting and microfluidics in biomedical research, explore methodologies for designing and fabricating biological structures via bioprinting, and evaluate the challenges and opportunities in integrating these technologies for the advancement of biomedical research.

11. Agnostic drugs: an innovative approach in Precision Medicine

Duration

First second and third year, 3 modules from 8 hours

Objectives of the course

Explore the concept of agnostic drugs and their role in Precision Medicine, provide an overview of the molecular basis of agnostic drugs and the mechanisms of action that distinguish them from traditional drugs, delve into the most recent developments in the research and development of agnostic drugs, analyze the clinical aspects and therapeutic implications of agnostic drugs in management of diseases.

Objectives of the PhD

The DIN in Precision Medicine aims to provide interdisciplinary theoretical/practical training in the biomedical sciences, and in the technological disciplines for health in order to create new professionals equipped with methodological tools and knowledge of advanced strategies, necessary for the implementation of patient-centred models of diagnosis and treatment, promoting innovative and efficient solutions. Advanced training based on a training project characterized by a strong interdisciplinarity, multidisciplinarity and transdisciplinarity, is guaranteed by the curricula structuring of the doctorate, which will provide specific skills in the student’s sector of interest. The training course is aimed at stimulating the continuous growth of doctoral students within the scientific community of reference, creating the conditions for the training of high quality professionals.

Employment opportunities and expected professionals

The PhD is strongly correlated to the analysis of training and employment needs directly expressed by the academic, clinical and industrial network that participates directly in its establishment and recorded in the design phase by the reference clinical and production sectors. The National Doctorate in Precision Medicine will provide trainees with specific and transversal skills useful for covering the role of new professional figures, including hybrid ones, such as, for example, those of Molecular Geneticist, Bioinformatician for Precision Medicine, Pharmacogenist, Biomedical Engineer for Precision Medicine, Molecular Oncologist, Data Scientist, Machine Learning Specialist for Precision Medicine, Manager for Healthcare Organizations and Precision Medicine.

The other figures of the PhD in Precision Medicine

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