





# Postgraduate Diploma

Thrombosis Genomics

Course Modality: **Online** Duration: **6 months**.

Certificate: TECH Technological University

Official N° of hours: 450 h.

Website: www.techtitute.com/us/medicine/postgraduate-diploma/postgraduate-diploma-thrombosis-genomics

# Index

> 06 Certificate

> > p. 30

# 01 Introduction

Genetics is one of the main factors causing thrombosis, in addition to other environmental causes, such as diet or smoking. Advances in genomics make it possible to improve treatments for people suffering from these pathologies, so it is essential for professionals to improve their knowledge in this field.



### tech 06 | Introduction

Thrombosis is an often undiagnosed pathology that can affect anyone, regardless of age, and that can become a serious disease. Early detection of venous thrombosis is essential to treat the disease and reduce sequelae in patients. There are also preventive measures, such as physical or pharmacological ones.

Throughout this Postgraduate Diploma, students will focus on Genomic Medicine applied to the treatment of venous thrombosis. The program has been designed by specialists in the field, so students will receive a complete and specific training delivered by experts on the subject.

This program aims to establish the basis of knowledge in this field, starting with genome-wide association studies (GWAS), massive sequencing studies and gene expression regulation.

Therefore, after completing and passing the Postgraduate Diploma, students will have acquired the theoretical knowledge necessary to carry out effective treatment of venous thrombosis in the main areas of professional practice.

This **Postgraduate Diploma in Thrombosis Genomics** contains the most complete and up-to-date scientific program on the market. Its most notable features are:

- Case studies presented by experts in Thrombosis Genomics
- The graphic, schematic, and practical contents with which they are created, provide scientific and practical information on the disciplines that are essential for professional development
- The latest news on Thrombosis Genomics
- Practical exercises where self-assessment can be used to improve learning
- Special emphasis on innovative methodologies in Thrombosis Genomics
- Theoretical lessons, questions to the expert, debate forums on controversial topics, and individual reflection assignments
- Content that is accessible from any fixed or portable device with an Internet connection



Don't miss the opportunity to study this Postgraduate Diploma in Thrombosis Genomics with us. It's the perfect opportunity to advance your career"



This Postgraduate Diploma may be the best investment you can make when selecting a refresher program for two reasons: in addition to updating your knowledge of Thrombosis Genomics, you will obtain a qualification endorsed by TECH Technological University"

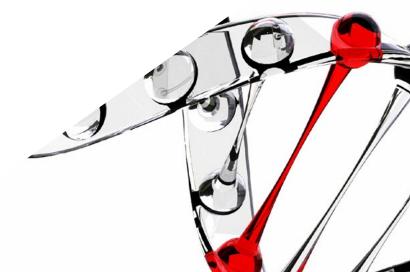
The teaching staff includes professionals belonging to the field of Thrombosis Genomics, who contribute their vast work experience to this program, in addition to recognized specialists from leading societies and prestigious universities.

The multimedia content, developed with the latest educational technology, will provide the professional with situated and contextual learning, i.e., a simulated environment that will provide immersive specialization programmed to learn in real situations.

This program is designed around Problem-Based Learning, whereby the professional must try to solve the different professional practice situations that arise throughout the program. To that end, the professional will have the help of an innovative, interactive video system made by recognized and extensively experienced experts in Thrombosis Genomics.

This program comes with the best didactic material, providing you with a contextual approach that will facilitate your learning.

This 100% online Postgraduate Diploma will allow you to combine your studies with your professional work while expanding your knowledge in this field.







## tech 10 | Objectives

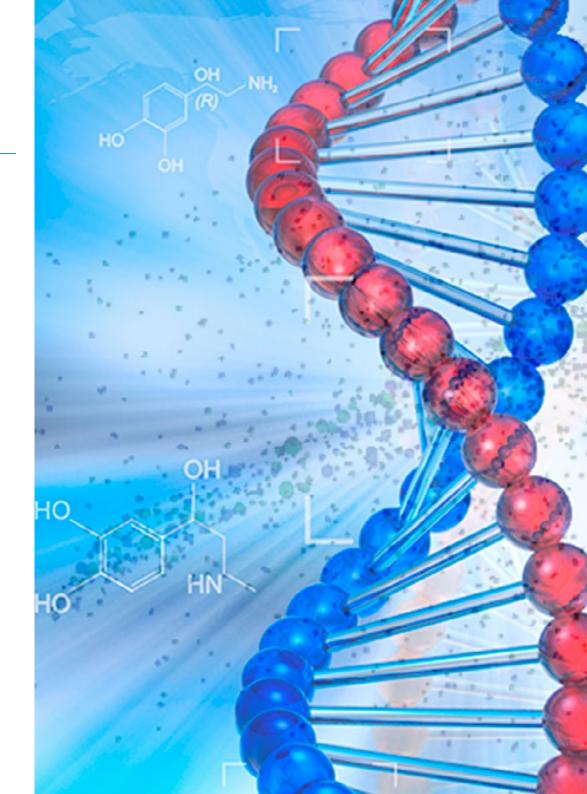


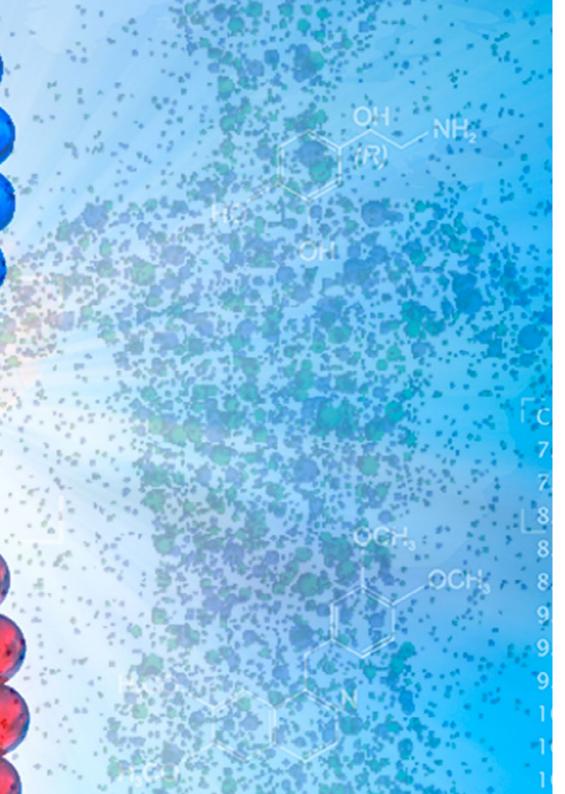
### **General Objectives**

- Delve into the knowledge of venous thromboembolism as a complex disease
- Specialize in the field of omic data and bioinformatic methods applied to precision medicine
- Keep up with the latest updates on the disease



The broad view on the multidisciplinary approach that managing autoimmune diseases requires, including the essential guidelines and knowledge in this scientific discipline"





### Objectives | 11 tech



### **Specific Objectives**

# Module 1. Thrombosis in the Genomic Era I: Genome-Wide Association Studies (GWAS)

- Provide an overview of genetics, and in particular of genome-wide association studies
- Show the current status of the use of genetics in venous thromboembolism

#### Module 2. Thrombosis in the Genomic Era II: Massive Sequencing Studies

- Know the genetic basis and molecular study of thrombosis and hemostasis
- Identify DNA sequencing techniques
- \* Acquire knowledge of bioinformatic analysis of NGS data
- Learn how to interpret NGS results in thrombosis and hemostasis
- Learn about the future perspectives of NGS technologies

## Module 3. Thrombosis in the Genomic Era III: Regulation of Gene Expression Studies (RNA and miRNA)

- Gain knowledge about RNA-seq
- Know the experimental designs and quality control for RNA-seg studies





### tech 14 | Course Management

### Management



### Dr. Soria, José Manuel

- Genomics Group of Complex Diseases
- Sant Pau Hospital Research Institute (IIB Sant Pau)
- Santa Creu i Sant Pau Hospital Barcelona

#### **Professors**

#### Dr. Sabater Lleal, María

- Degree in Biology, University of Barcelona, 2000
- Specialized in Biomedicine
- PhD in Genetics, University of Barcelona, 2006
- Genomics Group of Complex Diseases Sant Pau Hospital Research Institute (IIB Sant Pau) Santa Creu i Sant Pau Hospital Barcelona
- Cardiovascular Genetics Research Associate, Cardiovascular Medicine Unit (KI)

#### Dr. Vidal, Francisco

- Degree in Biology, Universitat of Barcelona
- Official Doctoral Program in Biochemistry, Molecular Biology and Genetics University of Barcelona
- \* Executive Master's Degree in Healcare Organization ESADE Business School/ Ramon Llull University
- \* Specialist Physician, Blood and Tissue Bank (BST) Barcelona







### tech 18 | Structure and Content

# **Module 1.** Thrombosis in the Genomic Era I: Genome-Wide Association Studies (GWAS)

1.1. Introduction to Genetics
-------------------------------

- 1.1.1. Introduction and Basic Concepts
  - 1.1.1.1. Genes
  - 1.1.1.2. Polymorphisms, Alleles and Loci
  - 1.1.1.3. Haplotypes
  - 1.1.1.4. Concept of Linkage Disequilibrium
  - 1.1.1.5. Genotype
  - 1.1.1.6. Phenotype
- 1.1.2. Genetics to Study Complex Diseases
  - 1.1.2.1. Complex and Rare Diseases
  - 1.1.2.2. Study of Candidate Genes vs. Global Genome Studies
- 1.1.3. Types of Polymorphism, Nomenclature and Genome Versions
- 1.1.4. Genotyping Chips
- 1.2. Introduction to Global Genome-Wide Analysis Studies (GWAS)
  - 1.2.1. What Is a GWAS?
  - 1.2.2. GWAS Study Design
    - 1.2.2.1. Heritability
    - 1.2.2.2. Case-Control vs. Quantitative Trait Analysis
    - 1.2.2.3. Sample Size and Statistical Power
    - 1.2.2.4. Biases by Population Substructure
    - 1.2.2.5. Phenotypes: Standardization and Outliers
  - 1.2.3. The Genetic Association Test
  - 1.2.4. Useful Software for GWAS
- 1.3. Genetic Imputation
  - 1.3.1. Concept of Imputation
  - 1.3.2. Reference Panels
    - 1.3.1.1. Hap Map Project
    - 1.3.1.2. 1000 Genomes Project
    - 1.3.1.3. Haplotype Reference Consortium Project
    - 1.3.1.4. Other Population-Specific Projects



### Structure and Content | 19 tech

1.4. Quality Control and Filt
-------------------------------

- 1.4.1. Pre-Imputation Filters
  - 1.4.1.1. Minor Allele Frequency
  - 1.4.1.2. Hardy-Weinberg Equilibrium
  - 1.4.1.3. Genotyping Errors (Call Rate)
  - 1.4.1.4. Excess Heterozygosity
  - 1.4.1.5. Mendelian Errors
  - 1.4.1.6. Sex Errors
  - 1.4.1.7. Chain Direction
  - 1.4.1.8. Family Relationships
- 1.4.2. Post-Imputation Filters
  - 1.4.2.1. Monomorphic Variants, Frequencies
  - 1.4.2.2. Imputation Quality
- 1.4.3. Post GWAS Filters
- 1.4.4. Quality Control Software
- 1.5. Analyzing and Interpreting GWAS Results
  - 1.5.1. Manhattan Plot
  - 1.5.2. Multiple Testing Correction and Genome-Wide Significant Results
  - 1.5.3. Concept of Genetic Locus
- 1.6. Meta-Analysis and Replication
  - 1.6.1. Common Workflow in GWAS Studies
  - 1.6.2. Meta-Analysis
    - 1.6.2.1. Meta-Analysis Methods
    - 1.6.2.2. Required Information for Meta-Analyses
    - 1.6.2.3. Meta-Analysis Result
    - 1.6.2.4. Meta-Analysis Software Examples
  - 1.6.3. The Most Relevant Consortia
- 1.7. Post GWAS Analysis
  - 1.7.1. Fine-Mapping and Regional Graphic
  - 1.7.2. Conditional Analysis
  - 1.7.3. Selecting the Best Gene Candidate (from Locus to Gene)
    - 1.7.3.1. Exploiting Information on Expression
    - 1.7.3.2. Gene Set Enrichment Analyses
    - 1.7.3.3. Study of the Potential Functional Effect of Polymorphism

#### 1.8. The Era of GWAS

- 1.8.1. GWAS Data Repositories
- 1.8.2. Taking Stock of the GWAS Era Results
- 1.9. Use of GWAS Results
  - 1.9.1. Risk Estimation Models
  - 1.9.2. Mendelian Randomization Studies
- 1.10. Genetic Analysis of Venous Thromboembolism (VTE)
  - 1.10.1. Some History
  - 1.10.2. The Most Relevant GWAS Studies on VTE
  - 1.10.3. Latest Studies Results
  - 1.10.4. Clinical Implications of Genetic Results: The Importance of Coagulation Cascades and New Metabolic Pathways
  - 1.10.5. Future Strategies

# **Module 2.** Thrombosis in the Genomic Era II: Massive Sequencing Studies

- 2.1. Genetic Basis and Molecular Study in Thrombosis and Hemostasis
  - 2.1.1. Molecular Epidemiology in Thrombosis and Hemostasis
  - 2.1.2. Genetic Study of Congenital Diseases
  - 2.1.3. Classical Approach to Molecular Diagnostics
  - 2.1.4. Indirect Diagnosis or Genetic Linkage Techniques
  - 2.1.5. Direct Diagnostic Techniques
    - 2.1.5.1. Mutation Screening
    - 2.1.5.2. Direct Mutation Identification
- 2.2. DNA Sequencing Techniques
  - 2.2.1. Sanger's Traditional Sequencing
    - 2.2.1.1. Characteristics of the Technique, Limitations and Application in Thrombosis and Hemostasis
  - 2.2.2. Next-Generation Sequencing (NGS)
    - 2.2.2.1. NGS Platforms in Molecular Diagnostics
    - 2.2.2.2. General Information on the Technology, Possibilities and Limitations of NGS vs. Traditional Sequencing
  - 2.2.3. Third-Generation Sequencing (TGS)

### tech 20 | Structure and Content

2.3.	Different Approaches to Genetic Studies Using NGS					
	2.3.1.	Gene Panel Sequencing				
	2.3.2.	Whole Exome Sequencing and Whole Genome Sequencing				
	2.3.3.	Transcriptomics by RNA-Seq				
	2.3.4.	MicroRNA Sequencing				
	2.3.5.	Mapping Protein-DNA Interactions with ChIP-Seq				
	2.3.6.	Epigenomics Analysis and DNA Methylation Using NGS				
2.4.	Bioinformatics Analysis of NGS Data					
	2.4.1.	. The Challenge of Bioinformatics Analysis of Massive NGS Generated Da				
	2.4.2.	IT Requirements for NGS Data Management and Analysis				
		2.4.2.1. NGS Data Storage, Transfer and Sharing				
		2.4.2.2. Computing Power Required for NGS Data Analysis				
		2.4.2.3. Software Requirements for NGS Data Analysis				
		2.4.2.4. Bioinformatics Skills Required for NGS Data Analysis				
	2.4.3.	Base Calling, FASTQ File Format and Base Quality Scoring				
	2.4.4.	NGS Data Quality Control and Pre-processing				
	2.4.5.	Read Mapping				
	2.4.6.	Variant Calls				
	2.4.7.	Tertiary Analysis				
	2.4.8.	Structural Variation Analysis by NGS				
	2.4.9.	Methods to Estimate Copy Number Variation from NGS Data				
2.5.	Concept and Types of Mutation Detectable by NGS					
	2.5.1.	Molecular Etiology of Thrombotic and Hemorrhagic Disorders				
	2.5.2.	Mutation Nomenclature				
	2.5.3.	Functional Implication of Identified Variants/Mutations				
	2.5.4.	Differentiation between Mutation and Polymorphism				
2.6.	Fundamental Molecular Databases in NGS					
	2.6.1.	Locus Specific Databases (LSDB)				
	2.6.2.	Previous Mutation Descriptions in Databases				
	2.6.3.	Databases of Variants Detected in Healthy Population by NGS				
	2.6.4.	Molecular Databases with Clinical Annotations				
2.7.	Analysis and Interpretation of NGS Results on Thrombosis and Hemostasis					
	271	Mutation Validation				

2.7.2. Concept of Mutation Pathogenicity

	2.7.3.	Genotype-Phenotype Correlation			
		2.7.3.1. In Silico Studies			
		2.7.3.2. Expression Studies			
		2.7.3.3. In Vitro Functional Studies			
2.8.	Role of NGS in Genetic Counseling and Prenatal Diagnosis				
	2.8.1.	2.8.1. Genetic Counseling in the NGS Era			
	2.8.2.	Ethical Issues Specific to NGS and Whole Genome Sequencing for Genetic Counseling and Clinical Diagnostics			
	2.8.3.	Conventional Prenatal Diagnosis and Methods			
	2.8.4.	Genetic Pre-implant Diagnosis			
	2.8.5.	Noninvasive Prenatal Diagnosis			
		2.8.5.1. Use of Fetal DNA in Maternal Circulation for Prenatal Diagnosis			
		2.8.5.2. Sequencing of SNPs from Circulating Fetal DNA			
		2.8.5.3. Limitations and Challenges of NGS-Based Non-invasive Prenatal Testing			
		2.8.5.4. Clinical Implementation of Non-Invasive Prenatal Testing for Aneuploidies			
2.9.	Future Perspectives in NGS Technologies and Data Analysis				
	2.9.1.	Technological Development of Sequencing in the Mid-Term			
	2.9.2.	Evolution of Bioinformatics Tools for High-Throughput Sequencing Data Analysis			
	2.9.3.	Standardization and Rationalization of NGS Analytical Processes			
	2.9.4.	Parallel Computation			
	2.9.5.	Cloud Computing			

# **Module 3.** Thrombosis in the Genomic Era III: Regulation of Gene Expression Studies (RNA and miRNA)

- 3.1. Introduction to RNA-seq
  - 3.1.1. Technique Description
  - 3.1.2. Advantages over Expression Arrays
  - 3.1.3. Limitations

### Structure and Content | 21 tech

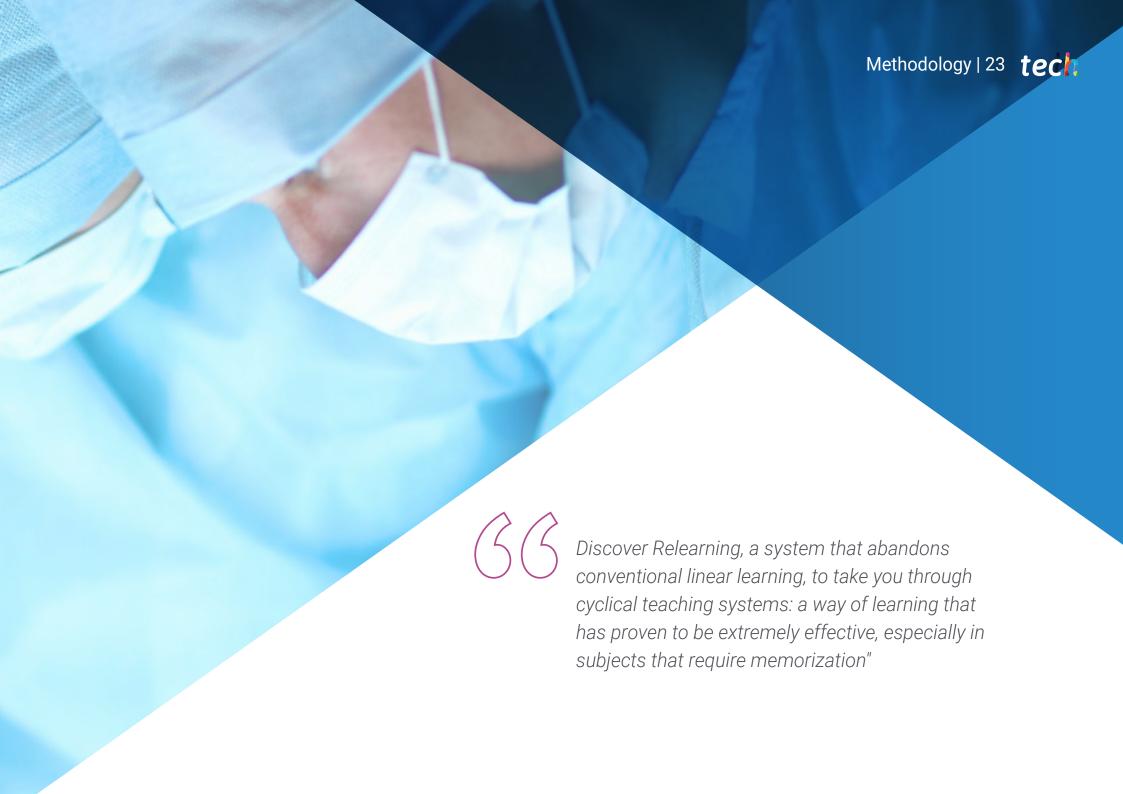
3.2.	Experimental	Design for	r RNA-sea	Studies
0.2.	LAPCITICITUAL	DCGIGITIO	1 1 11 17 1 0 0 9	Ottadico

- 3.2.1. Concept of Randomization and Blocking
- 3.2.2. Biological Replicas vs. Technical Replicas
- 3.2.3. Number of Replicas
- 3.2.4. Sequencing Depth
- 3.2.5. Type of Library
- 3.3. Quality Control for RNA-seq
  - 3.3.1. Quality Metrics for RNA-seq
  - 3.3.2. Programs Designed for RNA-seq Quality Control
- 3.4. RNA Alignment and Quantification
  - 3.4.1. Reference Genome (Genome-based)
  - 3.4.2. Reference Genome (Transcriptomics-Based)
- 3.5. De Novo Assembly and RNA Annotation
  - 3.5.1. Pipeline without Reference Transcriptome
  - 3.5.2. Annotation of Coding and Non-Coding Transcripts
- 3.6. Differential Expression with RNA-seq
  - 3.6.1. Standardization
  - 3.6.2. Latent Variable Elimination
  - 3.6.3. Programs and Statistics Methods
  - 3.6.4. Functional Enrichment
- 3.7. Other Applications of RNA-seq Technology
  - 3.7.1. Alternative Splicing Detection
  - 3.7.2. Chimera Transcript Detection
  - 3.7.3. Mutation Detection
  - 3.7.4. Allele-specific Expression Detection
- 3.8. Small RNA-seq
  - 3.8.1. Small RNA-seq Library Building3.9.8.1. Quality Control for Small RNA-seq
  - 3.8.2. Alignment and Quantification for Small RNA-seq
  - 3.8.3. miRNA Annotation
  - 3.8.4. miRNA targets

- 3.9. Gene Coexpression Networks
  - 3.9.1. Concept of Gene Coexpression Networks
  - 3.9.2. Differential Coexpression vs. Differential Expression
  - 3.9.3. Weighted Gene Coexpression Networks Analysis (WGCNA)
  - 3.9.4. Gene Coexpression Networks Visualisation
- 3.10. Gene Expression Regulation Analysis in Venous Thromboembolism (VTE)
  - 3.10.1. Some History
  - 3.10.2. Relevant Studies on VTE
  - 3.10.3. Latest Studies Results
  - 3.10.4. Clinical Implications in the Results
  - 3.10.5. Practical Examples and Exercises







### tech 24 | Methodology

#### At TECH we use the Case Method

What should a professional do in a given situation? Throughout the program, students will face multiple simulated clinical cases, based on real patients, in which they will have to do research, establish hypotheses, and ultimately resolve the situation. There is an abundance of scientific evidence on the effectiveness of the method. Specialists learn better, faster, and more sustainably over time.

With TECH you will experience a way of learning that is shaking the foundations of traditional universities around the world.



According to Dr. Gérvas, the clinical case is the annotated presentation of a patient, or group of patients, which becomes a "case", an example or model that illustrates some peculiar clinical component, either because of its teaching power or because of its uniqueness or rarity. It is essential that the case is based on current professional life, trying to recreate the real conditions in the physician's professional practice.



Did you know that this method was developed in 1912, at Harvard, for law students? The case method consisted of presenting students with real-life, complex situations for them to make decisions and justify their decisions on how to solve them. In 1924, Harvard adopted it as a standard teaching method"

#### The effectiveness of the method is justified by four fundamental achievements:

- Students who follow this method not only achieve the assimilation of concepts, but also a development of their mental capacity, through exercises that evaluate real situations and the application of knowledge.
- 2. Learning is solidly translated into practical skills that allow the student to better integrate into the real world.
- 3. Ideas and concepts are understood more efficiently, given that the example situations are based on real-life.
- Students like to feel that the effort they put into their studies is worthwhile.
   This then translates into a greater interest in learning and more time dedicated to working on the course.





### **Relearning Methodology**

At TECH we enhance the case method with the best 100% online teaching methodology available: Relearning.

This university is the first in the world to combine the study of clinical cases with a 100% online learning system based on repetition, combining a minimum of 8 different elements in each lesson, a real revolution with respect to the mere study and analysis of cases.

Professionals will learn through real cases and by resolving complex situations in simulated learning environments. These simulations are developed using state-of-the-art software to facilitate immersive learning.



### Methodology | 27 tech

At the forefront of world teaching, the Relearning method has managed to improve the overall satisfaction levels of professionals who complete their studies, with respect to the quality indicators of the best online university (Columbia University).

With this methodology, more than 250,000 physicians have been trained with unprecedented success in all clinical specialties regardless of surgical load. Our pedagogical methodology is developed in a highly competitive environment, with a university student body with a strong socioeconomic profile and an average age of 43.5 years old.

Relearning will allow you to learn with less effort and better performance, involving you more in your specialization, developing a critical mindset, defending arguments, and contrasting opinions: a direct equation to success.

In our program, learning is not a linear process, but rather a spiral (learn, unlearn, forget, and re-learn). Therefore, we combine each of these elements concentrically.

The overall score obtained by TECH's learning system is 8.01, according to the highest international standards.

### tech 28 | Methodology

This program offers the best educational material, prepared with professionals in mind:



#### **Study Material**

All teaching material is produced by the specialists who teach the course, specifically for the course, so that the teaching content is highly specific and precise.

These contents are then adapted in audiovisual format, to create the TECH online working method. All this, with the latest techniques that offer high-quality pieces in each and every one of the materials that are made available to the student.



#### **Surgical Techniques and Procedures on Video**

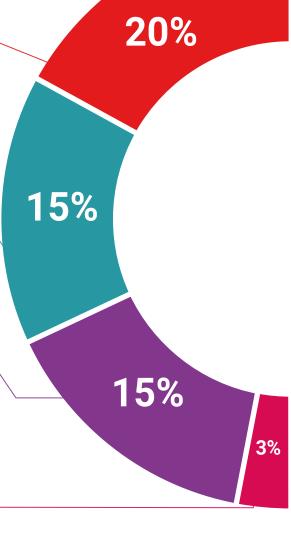
TECH introduces students to the latest techniques, the latest educational advances and to the forefront of current medical techniques. All of this in direct contact with students and explained in detail so as to aid their assimilation and understanding. And best of all, you can watch the videos as many times as you like.



#### **Interactive Summaries**

The TECH team presents the contents attractively and dynamically in multimedia lessons that include audio, videos, images, diagrams, and concept maps in order to reinforce knowledge.

This exclusive educational system for presenting multimedia content was awarded by Microsoft as a "European Success Story".





#### **Additional Reading**

Recent articles, consensus documents and international guidelines, among others. In TECH's virtual library, students will have access to everything they need to complete their course.

### **Expert-Led Case Studies and Case Analysis**

Effective learning ought to be contextual. Therefore, TECH presents real cases in which the expert will guide students, focusing on and solving the different situations: a clear and direct way to achieve the highest degree of understanding.



#### **Testing & Retesting**

We periodically evaluate and re-evaluate students' knowledge throughout the program, through assessment and self-assessment activities and exercises, so that they can see how they are achieving their goals.



#### Classes

There is scientific evidence on the usefulness of learning by observing experts.

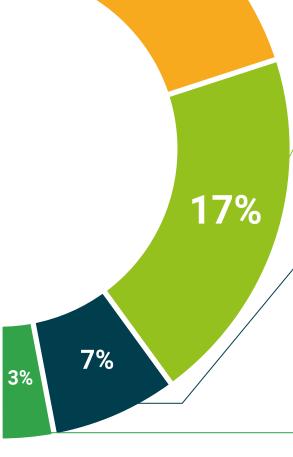
The system known as Learning from an Expert strengthens knowledge and memory, and generates confidence in future difficult decisions.



#### **Ouick Action Guides**

TECH offers the most relevant contents of the course in the form of worksheets or quick action guides. A synthetic, practical, and effective way to help students progress in their learning.









### tech 32 | Certificate

This **Postgraduate Diploma in Thrombosis Genomics** contains the most complete and up-to-date scientific program on the market.

After the student has passed the assessments, they will receive their corresponding Postgraduate Diploma issued by **TECH Technological University** via tracked delivery\*.

The diploma issued by **TECH Technological University** will reflect the qualification obtained in the Postgraduate Diploma, and meets the requirements commonly demanded by labor exchanges, competitive examinations, and professional career evaluation committees.

Title: Postgraduate Diploma in Thrombosis Genomics

Official No of hours: 450 h.



<sup>\*</sup>Apostille Convention. In the event that the student wishes to have their paper diploma issued with an apostille, TECH EDUCATION will make the necessary arrangements to obtain it, at an additional cost.

future
health confidence people
education information tutors
guarantee accreditation teaching
institutions technology learning
community commitment



### Postgraduate Diploma Thrombosis Genomics

Course Modality: Online Duration: 6 months.

Certificate: TECH Technological University

Official No of hours: 450 h.

